Biobanking for Genomic Research:

Public awareness and stakeholders' attitudes towards risks and benefits in Malta.

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"A society grows great when it plants trees whose shade they know they shall never sit in." – **Greek Proverb**

Dedicated to those contributing to potential medical breakthroughs.

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Abstract

Participant motivation is essential for the success of biobanking for genomic research. The benefits which emerge from the process play an important role in this respect, however, issues of risk and trust also have a direct impact. This mixed-methods study includes preliminary quantitative research on the level of local public awareness of biobanking for genomic research, as well as in-depth qualitative analysis of the attitudes, beliefs and perspectives of the key stakeholders towards the donation of biospecimens to a biobank for research. A survey questionnaire was administered amongst a sample of 387 respondents stratified on the basis of gender and educational background. The key result is that the significant majority of the local population does not know what a 'biobank' is, with a mere 4.9% (19 participants) of the sample mentioning the process of storing biospecimens for the purpose of biomedical research at some point during the survey, although only 2.3% (9 participants) knew that the premises which serves this purpose is called a 'biobank'. These findings were used to inform the production of audio-visual tools used during data collection for the qualitative exploration of stakeholders' attitudes towards risk and benefits of biobanking for genomic research.

The data for this qualitative phase of the study were collected through focus group sessions with individuals with existing medical conditions (henceforth referred to as patients), parents of patients, and the general public, as well as in-depth expert interviews with the researchers, the biobank manager and representatives of a local patient support group. Findings show that the perceptions of both the general public and patients towards the medical system at large, and even towards the wider social system or the establishment, have a direct impact on their concerns about donating their biospecimens for genomic research and biobanking, yet this does not necessarily impact their willingness to participate. Irrespective of the level of concern conveyed by the participants, the motivation to participate in genomic research studies remained consistent, as the perceived benefits overruled the potential negative repercussions. Moreover, the patients' focus groups demonstrated more enthusiasm towards participation when compared to their non-patient counterparts. Whilst non-patients are motivated by a need for self-fulfilment, patients are motivated by a more basic need for enhancing personal health or that of future generations. Findings highlight a form of stratification in reflexivity based on health status.

It is revealed that the discrepancy in motivation levels is linked to a disparity in perceptions towards risk, with patients being significantly less preoccupied with issues such as privacy risk, and more concerned about research not reaching its full potential. All stakeholder groups (experts and focus group participants) expressed the desire to attain some level of power and control over how the data is processed, particularly with regards to specific issues such as consenting, data sharing and incidental findings.

Keywords: biobanking, genomic research, biobank awareness, motivation, risk, trust, power, control

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1. Introduction

1.1 Background information

The main focus of this dissertation is biobanking, and since the preliminary research conducted for this dissertation reveals that the local levels of awareness of biobanks are alarmingly low, it seems pertinent to start with a definition and background information. A biobank is a repository that "collects, stores and distributes biological samples", ranging from human blood, saliva, or tissue, to animal or plant tissue (Schmilden, 2016, p.2). The focus of this dissertation is biobanks which store human biospecimens, as well as associated personal data for the purpose of genomic research.

Large-scale biobanks have become indispensable for medical research, as it is often not feasible for solo scientists to acquire the required pool of samples and to recollect new samples for each study. Biobanks counter this issue as they allow researchers to store and share samples, as well as results and associated personal and medical information, thus facilitating the research process (Schmilden, 2016). The collection and storage of human biospecimens for research started over a 100 years ago, but technology and computerisation have revolutionised biobanking, and nowadays various biobanks operate in collaboration to provide the best possible outcomes (Souza and Greenspan, 2013).

The following are the key stakeholders serving as pillars to the process of biobanking: a) the researchers, who study and conduct research intended at making ground-breaking discoveries in the hopes of developing novel cures, innovative bio-therapeutics, diagnostic procedures, or even methods of disease prevention, b) profit or non-profit investors, who provide the funds for research by financing the work of the former stakeholder group, and c) the research

participants, be it patients or non-patients, who donate biological samples, such as blood, saliva, tissue or even dysfunctional organs removed during surgery, to be used as tools for research (Ducournau and Strand, 2009).

The management of a biobank entails the coordination of a set of key components, namely the collection and storage of biological material collected from research participants, metadata and informatics as a description of the stored samples and pseudonymised personal data of research participants, such as medical records, as these provide essential added value to the biospecimens (Holub et al, 2018). Biobanking also involves the management of ethical, legal and societal issues related to the processes of collecting, storing and sharing the biospecimens and personal data, including the acquisition of informed consent and ensuring the privacy and protection of research participants (Vaught and Lockhart, 2012).

1.2 The University of Malta biobank

Recent developments in health and medicine indicate a key role of genetics in understanding human illnesses and conditions, therefore the past few decades saw a surge of biobanks being established all over the world (Chen and Pang, 2015), including the biobank at the University of Malta. It was inaugurated in 1989 and consists of a clinical bank and a population bank (University of Malta Biobank, n.d.). The clinical bank curates samples of individuals who are diagnosed with illnesses or diseases, and those of their families, whereas the population bank holds samples of healthy individuals from the general public. The biobank at the University of Malta nowadays forms a part of the Centre for Molecular Medicine and Biobanking and is a member of the Biobanking and BioMolecular Resources Research Infrastructure, European Research Infrastructure Consortium (BBMRI-ERIC); a hub aimed at facilitating research

through sample and data sharing (Mayrhofer et al, 2016). One of the projects being carried out at the biobank is that of collecting samples which add up to 1% of the Maltese population, with the aim of discovering new DNA variants causing illness in Malta, and thus facilitating the search for new diagnostic procedures, novel treatments and hopefully cures. Other collections at the biobank situated at the University of Malta include those being used for research on type 2 diabetes, ALS, muscular dystrophy, cystic fibrosis, colorectal cancer and thalassemia (University of Malta Biobank, n.d.).

1.3 Biobanking: a hope for rare diseases

Approximately 80% of rare diseases are genetic disorders, hence why most rare disease research falls under the umbrella of genomic research. Eight per cent of the Maltese population is born with, or will develop a rare disease throughout their lifetime; that amounts to around 25,000 people. However, only 3258 had been identified until 2018, which is 13% of the 25,000 that are currently affected or will be affected by a rare disease. Amongst the 3258 cases exist 350 variations of diagnosis (Agius, 2018).

Biobanks are especially crucial to rare disease research. The discovery of new cures, treatments or diagnostic procedures depends on research conducted on numerous samples, however, those suffering from a rare disease are typically a few, and are most likely to be geographically distant. Biobanks offer a solution through the long-term storage of samples, and the sharing of samples and other data amongst research institutions (Garcia et al, 2018).

As cited by Agius (2018), the European Organisation for Rare Diseases (EURORDIS) has identified the main concerns of individuals who suffer from rare diseases. The key obstacles

are difficulty in getting the right diagnosis, lack of research and knowledge on diseases, lack of adequate healthcare to deal with the symptoms, unaffordable drugs and sometimes no treatment option at all.

Although biobanks are particularly linked with rare diseases, biobank research is not limited to this field. Biobanks are also useful in studying disease patterns in the general population; case in point the UK biobank is currently undertaking a research project aimed at understanding the disparities in the severity of symptoms of Covid-19 amongst the population of the UK. The study is being carried out using 20,000 blood samples from the general public (UK Biobank, 2020).

1.4 Aims and objectives of this study

Biobanks are entirely dependent on donors, and this research shall precisely look into the two main elements which impact participation: awareness and attitudes towards biobanks in the Maltese context. This project provides a scientific, quantitative measure of the awareness of the Maltese public about biobanks, as well as an in-depth, qualitative analysis of the attitudes and perceptions towards donating human biospecimens for biomedical research. The findings emerging from the preliminary quantitative research are used to develop the right instruments for the collection of qualitative data. The latter part of this project considers the attitudes and perspectives of the key stakeholders in genomic research and biobanking, particularly patients[1],

[1] Throughout this dissertation the term 'patients' shall be used to refer to individuals with existing medical conditions, in order to distinguish them from participants who do not depend on regular medical management and intervention.

parents of patients, the general public and local experts, including researchers, the biobank manager and representatives from a local patient support group.

The choice of the topic emerged from a need for a sociological assessment of the societal aspects of genomic research and biobanking in Malta. There is a vacuum of information about perceptions regarding the topic in question, within the local scenario. The need for such a research project was put forward by the Centre for Molecular Medicine and Biobanking, through a call for social scientists to join the team at the centre, a role which together with principal investigator Dr Gillian Martin I occupied for over two years whilst carrying out the research for this dissertation. I decided to embark on this journey because I was always profoundly interested in issues of health and illness.

My own experience of living with an endocrine condition always left me wanting to know more. As I seek to gain more knowledge, I typically find myself challenging medical authorities. My own interpretation of living with a chronic condition enabled me to recognise the importance of having the perspectives of the patients, the public and the experts laid out into one research project with the ultimate aim of attaining a dynamic analysis which highlights the key issues associated with participating in genomics and biobanking.

1.5 Dissertation structure

The next chapter will offer a review of the literature on issues related to biobanking and genomic research vis-à-vis the key topics of awareness, motivation, risk and trust. Chapter 3 describes the research methods used to conduct this research and the reasons behind the choices that were made. The two subsequent chapters offer a thorough analysis of the primary data; Chapter 4 deals with the analysis of the quantitative data which explores awareness in Malta

of biobanking for genomic research, and Chapter 5 highlights the findings which emerged from the qualitative data about related attitudes and perceptions. The sixth chapter concludes the dissertation by offering a synopsis of the key findings.

2. Literature Review

2.1 The awareness of biobanks

Since most genetic and genomic research requires the active participation of donors who provide their biospecimens to the research community, awareness of biobanks is crucial to the success of research (Gaskell and Gottweis, 2011). Prior to this research, there were no local studies which looked into the awareness of biobanking in the Maltese context. However, findings from pan-European studies show a severely low level of awareness. Gaskell and Gottweis (2011) found that only a third of Europeans have heard of biobanks, urging biobank managers to start promoting them, or else "they could fail".

Whilst the success of biobanking depends on awareness and the motivation of potential participants, motivation is essentially dependant on the transparency and trustworthiness of biobanks, and well as the ethical and legal governance of samples and related data (Locock and Boylan, 2016). Also integral is a thorough understanding by the public of the biobanks' potential to revolutionise health for future generations (Goisauf and Durnová, 2019). Thus, awareness and motivation are the key driving forces behind active participation in biobanking and the success of most genetic and genomic research (Gaskell and Gottweis, 2011).

2.2 The key motivations behind the donation of biospecimens

Research shows that participants are generally neither aware nor bothered about the risks that can emerge through genetic or genomic research. However, when it comes to benefits participants are typically better informed, and thus, generally speaking, individuals are more

likely to be motivated to participate in medical research and biobanking, than to decline requests for participation (Domaradzki and Pawlikowski, 2019).

A study conducted by Tupasela and Snell (2012) reveals that the key participant motivators for donating biosamples for genomic research are the following: a) the benefit of improving knowledge on disease prevention, and knowledge as power over disease, b) the benefit of increasing the chances for finding ways to diagnose diseases earlier, c) the benefit of increasing the chances for finding a cure, d) the motivation of altruism - solidarity towards the nation, future generations and towards the scientific research community (Tupasela and Snell, 2012).

Since biobanks allow for the preservation of biospecimens for long periods they increase the chances for breakthroughs (De Souza and Greenspan, 2013; Schmilden, 2016) and thus they tend to instil a sense of hope in those suffering from incurable or untreatable illnesses, who often feel isolated and helpless (Boat and Field, 2011). Research hubs which encourage the involvement of patient research participants in the process of research create a sense of hope amongst participants and thus enhance trust towards the system (Kaye et al, 2014).

Different stakeholders are motivated by different factors; however, research indicates that the greater good is typically a common motivating factor amongst all main stakeholders, namely investors, biobank managers, researchers and research participants. Nonetheless, researchers tend to also be motivated by personal gains as they typically earn a wage from the research hub. Non-profit investors are motivated to get involved for philanthropic reasons, and venture capitalists for the purpose of developing a profitable product. Research participants do not typically participate for a return, especially in the case of non-patient participants, where the motive is generally purely altruistic. Patient-participants are also often altruistically motivated since they are usually aware that genomic research involves a longitudinal process which might

last longer than a lifetime. However, the hope for a breakthrough is still a common motivating factor amongst the latter stakeholder group, who perceive biomedical research as a way to potentially improve their health, that of their kin, or their descendants (Ducournau and Strand, 2009).

Maslow's (1943) theory of human motivation lies in the premise that individuals develop motivation based on the satisfaction of basic human needs. Maslow claims that the need for satisfying such needs has a direct influence on human behaviour and illustrates his theory on a pyramid which indicates that as individuals satisfy the most basic needs at the bottom of the pyramid, they develop the motivation to satisfy the needs which follow. Maslow (1943) places physiological needs at the very bottom of the pyramid, meaning that such needs are the most essential and are the first that individuals seek to satisfy. The pyramid is then shaped up with safety needs, the need for love and belonging, esteem needs, and finally the need for self-fulfilment at the very top of the pyramid. Research shows that different stakeholders express varying levels of motivation towards participating in genomic research and biobanking (Ducournau and Strand, 2009), and Maslow's theory can possibly shed light on this finding. Whilst some perceive participation in medical research as a mere altruistic action which satisfies the need for self-fulfilment, others are motivated by a more basic need, the need for safety through improved personal health (Ducournau and Strand, 2009).

2.3 Theories of risk and trust, and the medical system

> "When the life-world is colonized by medical insecurity, medicalised subjects come to suspect the messenger and the knowledge they bear" (Crawford, 2004, p.524).

Progress and innovation allow for considerable benefits but also contemporary challenges and risks (Schmilden, 2016). The issue of risks associated with the donation of biospecimens for research is complex. The sociological theory of risk provides insight into how social actors think, decide and act based on how they predict future consequences (Meyer et al, 2008). In the late 1970s, academia saw a growth curve in studies in the field of risk as the public started to change perceptions towards danger. There was a splurge of challenges on institutions: capitalists were confronted for labourer injuries, governments confronted for covering up for the capitalists, technological progress was challenged due to its impact on the environment, and there was a rise in challenges aimed at defending human rights. On the other hand, the establishment reacted by claiming that the concerns of the public were blown out of proportion and warned the public about the risks they incur every day without ever thinking about, such as being in the sun and crossing the road. Suddenly, the concept of risk, which was previously an unpopular academic area, gained momentum and became a core topic in fields such as sociology, anthropology and health studies (Douglas, 2013).

Social change is at the core of the work of Anthony Giddens and Ulrich Beck, who focus their sociological theory around the transition from pre-modern to modern societies, considering 'risk' to be a central characteristic of modernity. The concepts of 'risk' and 'trust' are entangled as they occur simultaneously; acting in spite of potential anticipated risk depends on trust, and trust is only necessary in the presence of an anticipated risk, however, in itself, placing trust in a person, entity or institution is a risk (Meyer et al, 2008; Luhmann, 1979). The notion of reflexivity brings the two concepts together (Ward, 2006).

Reflexive modernisation is characteristic of societies where individuals are exposed to further information which equips them to question the world around them (Beck, Giddens and Lash, 1994). Reflexivity has led modern societies into a social reality of constantly trying to

anticipate what the future holds, seeking for ways to combat the associated risks and questioning trustworthiness (Giddens, 1990; Giddens, 1991; Beck, 1992; Beck, Giddens and Lash, 1994).

The general belief in reflexive societies is that actions might introduce unforeseen, undesirable aftermaths. Climate change, lower fertility, birth defects due to prescribed medication, higher asthma rates and hundreds of other real-life circumstances are proof that "we don't, and can't know [...] new risk situations" including those created by human intervention itself, i.e. manufactured risks (Giddens, 1999, p.2). The key characteristic of risk societies is uncertainty. As the world innovates and makes progress, quite frankly everyone is relatively uncertain of what we are in for (Giddens, 1999).

The birth of risk societies is, according to Giddens, directly linked with the scientific and technological momentum. He particularly speaks of two types of revolutions behind risk societies; "the end of nature" and "the end of tradition". "For hundreds of years, people worried about what nature could do to us - earthquakes, floods, plagues, bad harvests and so on. At a certain point, somewhere over the past fifty years or so, we stopped worrying so much about what nature could do to us, and we started worrying more about what we have done to nature" (Giddens, 1999, p.3). Similarly, there was a shift from the traditional customs and norms of pre-modern societies to "individualisation". These unclear transitions are the point at which the new era of risk society emerged (Beck, 1992).

Some of the current realities were not predicted in the past, therefore the present is proof that the future can never be certain, ergo risk persists (Beck, 1996). A 'risk society' is constantly "dealing with hazards and insecurities induced and introduced by modernisation itself" (Beck, 1992).

However, risk essentially isn't danger (Beck, 1992). Risk societies are not necessarily more prone to hazard but rather more aware (Giddens, 1999). Pre-modern societies experienced dangers, yet the notion of risk did not exist, as they tended to perceive danger superstitiously; whereas modern, reflexive societies recognise their capability of controlling situations, and that human intervention impacts the world extensively (Beck, 1992). Consequently, risk emerges as societies aspire to predict and manage the future (Giddens, 1999). Societies seek for ways to cope with possible risks, either through actively acting to counter a known risk, accepting risk without responding or through denial (Elliott, 2002).

Traditional societies relied on intellects, experts or expert systems to comprehend the world around them and assumed that experts were infallible in their advice about the future. However, the future quickly became present and modern societies started to realise that expert systems were neither capable of predicting the future nor of controlling the unfavourable and as a consequence started to question their faith and trust in experts, and started to become more reflexive (Giddens, 1999). Whilst acknowledging that individuals still rely on expert systems in circumstances where they lack knowledge, it must be acknowledged that individuals no longer exclusively see experts as infallible beings who can predict consequences. This leads to what Giddens calls "lay re-skilling" which refers to a situation where individuals start to reject or challenge expert advice and regain control over their lives, such as by challenging medicine and pharmaceuticals and considering alternative ways of medication (Meyer et al, 2008). Trust requires both emotion and cognition, and as individuals become more reflexive, entrusting expert bodies becomes more difficult and consequently societies start to develop what Giddens (1991) calls, a sense of 'ontological insecurity'.

An example which Giddens (1999) himself gives is that of smoking. Some fifty years ago the practice of smoking was encouraged by medical practitioners who claimed that smoking can

serve as a stress reliever. The same medical system had not yet predicted the unfavourable consequences which emerge from smoking. Suddenly the discourse changed, and new risks started being discussed. Laypersons would grasp onto new knowledge and information provided to them, but ultimately they would still have to face the repercussions of the previous ill-advice. This is just one example of how access to information in reflexive modern societies leads social actors into a culture of ontological insecurity where they constantly seek for ways to combat the risks whilst questioning the knowledge of (medical) experts, the system, and their trustworthiness (Giddens, 1999). As a consequence of the uncertainty caused by misinformation, the health system, along with various other institutions, have suffered a decline in trust from laypersons in modern societies, as individuals are doubting the validity of "medical knowledge" (Ward, 2006). Therefore, trust in healthcare systems within reflexive societies cannot be treated as a given, but rather as an entitlement which must be earned and sustained (Beck, Giddens and Lash, 1994).

Paul Ward (2006) claims that reflexivity should not be seen as linear since individuals from different strata do not possess similar levels of reflexivity. Ward and Coates (2006) introduce the concept of 'stratified reflexivity', claiming that one's socio-economic position within a social hierarchy has a direct influence on their level of reflexivity, and thus rational action.

Risk is typically associated with a negative outcome since the term in itself refers to the possibility of an undesirable eventuality. However, one can also perceive risk positively. Risk-takers are more likely to face dangers, but also more likely to succeed. Case in point, entrepreneurial motivation is not enough for a business to flourish, unless the entrepreneur is willing to take a risk in the hopes of succeeding, and likewise, healthcare cannot make progress unless researchers and research participants are willing to take calculated risks with the hopes of developing therapeutic or diagnostic novelties. Avoiding anything risky and consistently

playing safe might decrease the chances for negative outcomes, however, it also minimises the chances of success. Hence, modern societies often perceive the determination to avoid risk at all costs as a driving force behind mediocrity (Giddens, 1999).

Avoiding progress out of fear of possible repercussions is a risk in itself. As Giddens (1999) points out, it might be evident that science and technology have led to catastrophes and epic disasters, however, the same innovations have eradicated illnesses, found treatments to serious conditions and cured millions of individuals. It is thus proper to strike a balance between the development of new risks due to progress and the eradication of old risks also as a result of progress. "The percept of staying close to nature or of limiting innovation rather than embracing it, can't always apply. [...] We may need quite often to be bold rather than cautious in supporting scientific innovation or other forms of change" (Giddens, 1999, p.9).

A direct link exists between risk and responsibility. Beck speaks of 'organised irresponsibility' to argue that the main problem with modernity is that dangers and hazards keep being created, yet no one is accountable for them and thus the situation cannot be improved. In an interview with Joshua Yates, Ulrich Beck had claimed that:

> "Politicians say they are not in charge, that they at most regulate the framework for the market. Scientific experts say they merely create technological opportunities: they don't decide how they are implemented. Businesses say they are simply responding to consumer demand. Society has become a laboratory with nobody responsible for the outcome of the experiment" (2003, p.5).

The shift from external to manufactured risk is leading to a "crisis of responsibility". As risk becomes less predictable, the attribution of responsibility becomes less clear and uncertainty starts to prevail as individuals struggle to calculate and transfer responsibility onto third parties (Giddens, 1999), such as insurance companies. The transfer of responsibility, where possible,

is common nowadays as individuals seek to embark on risky business whilst trying to ensure peace of mind (Baker, 2002).

The study of risk has unveiled various anomalies. It has been established that opinions about risk are very different between laypersons and experts; the two perceive risk disparately and consequently react differently (Douglas, 2013). This disparity has given rise to sub-disciplines aimed at classifying risks and communicating them. Nonetheless, the perplexing behaviours of the public persist, as they refuse to avoid foods which harm their bodies, as they avoid insuring against earthquakes, keep using gadgets which might cause accidents, and not bother about educational campaigns on the various forms of risk (Douglas, 2013). This in itself could be the result of a lack of trust towards those sending out the messages about possible dangers.

Moreover, Giddens (1990) speaks of distinct ways in which individuals react to and adapt to (mis)trust, risk and danger. The first 'adaptive reaction' he speaks of is that of 'pragmatic acceptors'; this refers to individuals who are aware of the limitations of science but accept the system anyway because they are uninterested in challenging the status quo. He then speaks of the 'sustained optimists' who have a quasi-religious attitude towards medicine and science since they believe that the ultimate goal of science is to achieve the best for the greater good and who also believe that ultimately science will give us more benefits than penalties, even if this is not indisputable yet. There are then the 'cynical pessimists' who pretty much have an antithetical view to that of the previous group. The last group are referred to as the 'radical engagers' and are individuals who recognise the limitations and dangers of science, and seek practical ways to mitigate the challenges by either eradicating the dangers, when possible, or seeking for ways to overstep them (Giddens, 1990).

2.3.1 Trust towards health systems

> "To show trust [...] is to behave as though the future were certain" (Luhmann, 1979).

Broad and extensive research has been carried out on the notion of trust in the social and behavioural sciences (Ward, 2006), however, in recent years, the concept of trust has also been rapidly gaining momentum in the field of healthcare. A study conducted by Schlesinger et al (2005) confirmed that in the mid-1990s, trust started to become a key concept in medical and health literature, with 1612 journal articles being published on the matter between 1995 and 2003, in contrast to a total of 764 journal articles being published on the matter in the preceding 15 years (1979-1994). Trust is nowadays considered a key pillar of healthcare and is often analysed through the sociological lens, primarily because a person's willingness to trust, or not to trust the system, is typically based on several social factors. The sociological theory of trust is therefore used as an analytical tool to understand how social factors affect trust towards the health and medical system (Meyer et al, 2008).

Giddens (1990) claims that trust and information are intertwined. Access to information, which is key in reflexive modern societies, leads individuals to base their actions more on rationality (reflexivity) and less on faith and trust. Trust in the healthcare system is the result of a public which supposes that the intentions of those who make it up are genuine and aim for achieving what is best for patients and the public (Ward, 2006). Giddens speaks of trust as a form of faith which results from "ignorance", whereas reflexivity challenges the intentions and knowledge of experts, leading to decreased levels of trust, where hard work and consistency are crucial for trust to be gained and nurtured (Giddens, 1990). In circumstances where individuals have complete knowledge, trust is not necessary; trust becomes key when individuals cannot

comprehend a situation due to a lack of knowledge and therefore must rely on faith, hoping that their best interests are secured (Giddens, 1991).

Giddens (1991) introduces the concept of faceless and facework commitments; two main types of relationships which foster trust or hinder it. Whilst trust (or mistrust) built through facework commitments is fostered and nurtured through interpersonal interactions with flesh and blood representatives of a system, such as general practitioners, faceless commitments refer to trust (or mistrust) created through system-based relations which do not involve any interpersonal interaction, but which are still key to the cultivation of trust or mistrust through the public's comprehension and perception of abstract systems. Key examples of trust in abstract systems are entrusting that the food we eat at a restaurant would not have been contaminated (Meyer et al, 2008); or entrusting an ATM with access to all bank account details (Giddens, 1991); or entrusting that a plane will take-off and land safely, without the need for interpersonal interaction with the pilot; or consuming medications without really understanding what they are made of, who concluded that they are beneficial, and how they impact the body.

In a nutshell, through these access points, the public meets with representatives of the system either through faceless interactions (abstract system) or through facework interactions (interpersonal relations), and trust is either fostered or eradicated, either enhanced or declined, at these access points (Schlichter, 2010). Access points serve as a reminder to the public that the abstract system is ultimately made of and operated by human beings who could be "fallible" at times. "Giddens is essentially exposing a difference between 'expertise' (the system of knowledge) and the 'expert' (representative), and locating trust as being a factor between the two" (Ward, 2006, p.125).

Several scholars have adopted Giddens' theoretical analysis and implemented it in their work, often observing that the two facets of trust are highly interrelated and interdependent. Therefore, the level of trust which stems from facework/ interactive relations impacts trust towards faceless relations or the abstract system, and vice versa. Representatives of the system, such as general practitioners (GPs) who interact face-to-face with recipients of health services as well as the abstract system, must seek to create an environment which feels, and is, safe and trustworthy (Rhodes and Strain, 2005, as cited in Meyer et al 2008).

Luhmann's structural-functionalism emphasises the important functions of trust within (and for) society, claiming that it is the glue which holds social life together as it prevents individuals from constantly having to make sense of the complexities of life. Trustworthy relations smooth out the process of decision making as individuals rely on others for assurance, thus trust minimises complexities allowing for social order (Luhmann, 1979). Luhmann's perspective of trust, as suggested in his 'systems theory', opposes that of Giddens, as he claims that trust is not to be analysed bilaterally as interpersonal or abstract, but rather as a multifaceted and multi-layered notion, relating to multiple social systems on multiple levels. Trust in systems (such as the medical system) or individuals (such as doctors or biomedical researchers) is dependent on trust towards vast social systems.

Whilst the claims of Luhmann and Giddens are essentially theories and lack empirical evidence (Meyer et al, 2008), in recent years, scholars have conducted empirical research to determine the variables which correlate with trust, using their theories as a basis. Ward's (2006) research findings support Luhmann's perspective, claiming that "there is a need to reconsider the two-layer conceptualization of trust". Ward's (2006) qualitative research investigated levels of trust towards general practitioners and the medical system, and how trust is gained or destroyed. The data suggest that in general those who participated in the research project mistrusted the

system but their mistrust was not simply based on an interpersonal relationship or due to inconsistency from the medical system, but rather as a result of a general sense of mistrust in authorities and institutions. Giddens (1990) claims that individuals determine whether or not to trust experts based on the accuracy of knowledge which they circulate, however, Ward's (2006) study suggests that cultivating trust is more complex, stating that individuals might not always trust based on what they make of their relations within the particular system, in this case the health system, but rather based on a wider feeling.

Ward's participants' feeling of social exclusion and lack of attachment led to a sense of mistrust towards organisations and institutions in general, including the health system. Their attitudes and perceptions were not necessarily rooted in personal experiences of lack of competence from a GP or the abstract system, but rather they primarily spoke about their perception of the wider system, including the government, as the culprit of their general feeling of mistrust.

Ward and Coates (2006) speak of a strong correlation between reflexivity, dependence and trust. The discourse amongst those who participated in their study revealed a sense of mistrust towards doctors, the medical system, and other government institutions, however, they also clearly communicated a sense of dependence on the knowledge of general practitioners and pharmaceuticals. Thus, these individuals might be reflexive in their critical perspective towards the systems, however, their undeniable dependence on the same system prohibits them from being reflexive actors who are willing to challenge the authorities, deny the experts and abandon the system (Ward and Coates, 2006).

Regaining control over one's of health is not always straightforward; Lupton (2012) speaks of a correlation between reflexive action and socio-economic status. She states that those who are disadvantaged socio-economically tend to have less access to information, educational

resources and enlightening publications, whereas those who are in an advantageous position are exposed to richer information, which empowers them to be reflexive social actors who are critical and challenging; and thus when experiencing mistrust in the system they would be more likely to act upon it. Elliot (2002) raises a similar argument in his discussion on the 'information-poor' and the 'information-rich'. This is evident in healthcare as the 'information-rich' are typically lay experts who have access to information which they can utilise to their benefit, such as through alternative medicine, international advice etc., whereas the 'information-poor' are deprived of such privilege, resulting in inequality amongst patients.

This is linked to Bourdieu's (1986) theory of capital. Various forms of capital, namely social, cultural, symbolic and economic capital are interchangeable and thus those possessing one form of capital are advantaged in the enhancement of the other forms of capital. Cultural capital is a non-financial resource which allows for, and is concurrently a result of, better education and enhanced knowledge. Social capital relates to strong social networks, economic capital relates to wealth, whilst symbolic capital relates to prestige (Tittenbrun, 2016). The interchangeability between the forms of capital means that those who occupy a high position within the socio-economic hierarchy tend to possess the various types of capital and are thus provided with the means to perpetuate their position. These understandings of capital and class distinction are also relevant to biobanking; those with higher educational backgrounds are advantaged when it comes to understanding the research process. *Habitus*, the physical embodiment of capital in terms of habits, tastes and skills, exposes resources and in turn allows for building richer networks and attaining social capital, and thus connecting with individuals who could transfer further knowledge (Veenstra and Burnett, 2014). Health literacy is a key predictor of experiences of individuals within the health system; case in point, those who are

better informed have a better understanding of their rights as participants and the implications of participating in genomic research and biobanking (Sentell et al, 2014).

In Foucault's words, 'knowledge is power', however, Ward (2006) claims that even if the information is widely distributed, the distinct ways through which the same information is accessed and internalised by different groups leads to stratified reflexivity, since "knowledge does not empower to the same extent everyone who possesses it" (Fuller, 1999, p.28). Reflexivity needs to be analysed in the context of class stratification, where the majority are passive individuals who receive and accept information from experts, whereas those with the social assets, wealth and cultural capital are considerably more likely to take control of their situation by acting upon their mistrust towards the system and challenging the experts and the authorities (Ward, 2006).

Data derived from genomic research are typically unavailable to the general public, or when available, information is rather complex and would not be comprehendible by non-experts (Robinson et al, 2013), thus Giddens' notion of reflexive modernity is challenged here as, on a general level, individuals in modern societies still tend to be dependent on medical experts (Ward, 2006). From accepting a diagnosis through interpersonal contact with a medical practitioner (facework interaction) to consuming prescribed medications without comprehending much about what they are made of, by whom and how they work (faceless interaction), the relationship between laypersons and the expert system and its experts is predominantly dependent on trust (Ward, 2006).

Several scholars claim that acting reflexively depends on a variety of factors, from social networks, to levels of autonomy, to socio-economic status, to hundreds of other social, economic, cultural elements, and Giddens has been thoroughly criticised for his linear

approach, and for failing to consider not only social class as a determining factor but also gender, age, nationality and other factors which could be critical in fostering trust (Meyer et al, 2008). Luhmann (1979), in contrast, speaks of trust as an extremely complex concept and admits that he cannot identify how trust is built; "social order does not stand and fall by the few people one knows and trusts. There must be other ways of building trust which do not depend on the personal element", and here Luhmann opens a huge opportunity for further research as he goes on to ask "But, what are they?"

When knowledge is insufficient, reliance on experts is inevitable. However, various contemporary scholars have emphasised that, in modern times, trust is not to be seen as given but rather as an ongoing process which requires prolonged consistency; trust is to be seen "as a learned, trained, and skilled activity" (Everts and Jackson, 2009).

2.3.2 Trust as a pillar of biobanking and genomic research

The importance of trust within the research process is not to be discounted, especially since donors are indispensable for genomic research, and research participation is encouraged if a trusting relationship exists between researchers and researched. Research participants must feel that their privacy will be safeguarded, that they will be protected from risk (Laurie et al, 2010). Trust is not attained by simply verbally reassuring participants and by claiming that efforts will be made to prevent harm, rather, trustworthiness is a process; it is achieved after consistently being transparent and clear about the processes involved. A detriment to trust is promising a degree of protection which cannot truly be guaranteed, such as ensuring complete anonymity when such safeguard is impossible; or by leaving out information about possible risks (Laurie et al, 2010).

Evidence shows that trust towards medical representatives, such as clinicians, usually leads to a positive impact on engagement in medical research. Research conducted by Robinson et al (2013), with research participants who were also patients, or the parents of patients with a genetic disease, showed a very clear correlation between trust in clinicians and the ease with which participants chose to give full consent for the use of their genetic data and samples. However, ethical considerations must also be carefully considered here (Carrieri et al, 2015).

Trustworthy relations between patients and clinicians might lead to a lack of clarity about whether an activity is being carried out for clinical or research purposes (Carrieri et al, 2015). The distinction between the two practices might sometimes become obscure for patients who are also research participants, thus creating ethical issues. Thus, best practice research requires researchers/ clinicians to preserve the trust-based relationships they have with participants/ patients by ensuring that a clear distinction is made between the two practices (Carrieri et al, 2015).

Research conducted by Ducournau and Strand (2009), amongst donors of a population bank in France, shows three patterns of trust amongst research participants. The first group identifies those who experienced a "natural trust" towards the institution and were thus unconcerned about the process of consent and disinterested in the information. The second group includes individuals who challenged the trustworthiness of the institution but participated nonetheless. The final group includes those individuals who showed interest in the consenting process and welcomed the information provided as an indicator of professionalism.

2.4 Issues of risk and trust when donating biospecimens for research

For most, at first glance, the donation of biospecimens for research seems not to be risky, however delving deeper into the matter reveals that the theory of risk and trust is immensely relevant to genomic research and biobanking (Ducournau and Strand, 2009). Technological developments are swiftly leading to great advancements within the field of biomedical research (Oliver et al, 2011). As a result, lawmakers and ethicists are constantly trying to combat the risks which arise as a result of such advancements with the aim of minimising, or when possible eradicating, harm on research participants.

Donating biospecimens for research typically requires the storage of genetic information on large-scale genomic databases, and thus risk in biobanking cannot be denied (Laurie et al, 2017). Some claim that the risks are minimal because data is encrypted and pseudonymised, however, it is known that the variations in one's genome can be considered as very accurate identifiers of the person. Additionally, as technology continues to develop, "correlations between genotype[2] and phenotype[3] will be more easily ascertained and associated with an individual" (Balaji and Terry, 2015).

A balance is sought to be achieved between a) allowing science to research its full potential, and b) ensuring the privacy and safety of data subjects. Alongside scientific and genomic advancements, legal and ethical considerations must be taken seriously to avert any social risks for genomic research participants (Oliver et al, 2011).

[2] Set of genes inside DNA.
[3] Observable expressions of genes.

2.4.1 Data sharing amongst biobanks and for future research

Genomic research is becoming increasingly dependent on data sharing amongst biobanks (Goisauf et al, 2019). As a matter of fact, one of the main issues linked to biobanking is the risk of breaching personal privacy as a result of data sharing (Laurie et al, 2010). In order to exploit data in the most effective way, collaboration amongst scientists is crucial. Medical researchers are urged to share "generated sequence data" with other medical researchers for the benefit of the research community (Robinson et al, 2013), with the aim of making sensible findings which would ultimately benefit the whole of society. Thus, data sharing and the reuse of samples for future projects are indispensable for genomic research (Balaji and Terry, 2015).

There has recently been a development in guidelines about the use of scientific data; specifically, through what is referred to as the FAIR Data Principle. FAIR stands for findability, accessibility, interoperability and reusability. Holub et al (2018) claim that while the FAIR principle is a good start, a more specific approach is required to battle the challenges of medical research. Whilst the FAIR principle might be adequate for other sciences, medical research is facing challenges which are incomparable to the other sciences when it comes to the use and reuse of data. Reproducibility and privacy protection are considered to be the two major challenges for medical researchers. Thus, Holub et al (2018) propose the principle of FAIR-Health. They argue that for a biobank to be compliant to FAIR-Health three principles must be fulfilled, over and over the basic four FAIR principles:

1. "Quality and traceability": Quality storage is imperative for future reuse of specimens and medical data. ISO certification is encouraged to ensure that the required standards

are met by the biobank (Souza and Greenspan, 2013). For reusability, detailed documentation of methods, tools used and results is crucial (Holub et al, 2018).

2. “Incentives: Not only should researchers have incentives to use existing resources but resource providers also need clear incentives to promote and facilitate the reuse of their resources” (Holub et al, 2018, p.5).

3. “Privacy regulation compliance”: At the base of medical research exists a struggle between three naturally occurring competing interests. Firstly, the privacy protection of research participants who provide the researchers with sensitive personal data. Secondly, the need to reuse data to achieve utmost benefits for society and the research community, and thirdly the complex conditions of ownership, and financial interests (Holub et al, 2018). With regards to personal data privacy and data protection, the General Data Protection Regulation (GDPR) is generally used a basis.

“Consent should not include promises regarding privacy and control over personal information in the event of data sharing, as they cannot be fulfilled. Participant data cannot be de-identified (e.g. pseudonymised or anonymised) in the clinical context, and data pathways should be transparent to participants. Researchers should ensure that their information governance is compliant with best current practices and legal guidance” (Carrieri et al, 2015, p. 2).

Laurie et al (2010) point out that not only is data sharing considered to be ethical when conducted in a compliant manner, but that not sharing data might be considered unethical, and in some instances even illegal. When researchers collect biospecimens, they typically ensure research participants that their tissue or blood will be used for achieving data which can bring about beneficial developments for the greater good. By keeping data exclusive to oneself and

being resistant to data sharing, researchers would be breaking the commitment of using the biospecimens for the greater good, and instead, they would be prioritising personal achievement (Laurie et al, 2010). Research participants perceive genomic research as a means for developing novel technologies for diagnostic purposes, or for developing novel medications for curing or treating illness, and ultimately their decision to donate their biospecimens is rooted in the desire for healthcare to flourish. According to Laurie et al (2010), research participants do not generally expect a financial reward for participating in research, they do however expect that their data are used to their optimum potential, to increase the chances for breakthroughs.

Biobanking increasingly calls for less individualism and more solidarity amongst research professionals, research participants and the general public, where data is shared on an ever-increasing scale; from local to national, to European, to global (Laurie et al, 2010). It must be acknowledged that data sharing does, to a certain extent, increase the risk of intrusion into personal data, however, without data sharing many findings would not be possible, as genomic data depends on large quantities of samples which are usually impossible to gather individually. Additionally, research institutions which follow ethical and legal guidelines almost entirely protect their research participants from harm. Thus, best practice biomedical research calls for compliant data sharing to achieve the best results for the greater good (Laurie et al, 2010).

A study held by Goisauf et al (2019) amongst 273 professionals who work in the field of genomic research and biobanking sought to unveil what truly happens in practice within the field of biobanking. The study reveals that informing research participants about the sharing of data with third parties at the stage of consent is still not typically practised. However, it is interesting to note that professionals are not ambivalent towards this, and they welcome it. Furthermore, in the case of data sharing with commercial entities, researchers are sceptical of

informing participants at the stage of consent, whereas other biobank professionals such as managers, and ethical, legal and social (ELSI) consultants are more certain of the importance such practice. The reasons behind the discrepancy in opinions between both groups have not been observed in this study, and thus the researchers call for further investigation (Goisauf et al, 2019).

2.4.2 **Privacy risk**

"While it is clear that [privacy] risks must be minimised wherever possible, it is impossible to eliminate them completely" (Laurie et al, 2010, p. 328). Biobanking and the storing of genomic and other personal information, such as medical histories, naturally create privacy risks for donors. However, efforts at protecting the privacy of research participants and to safeguard them from harm perpetuate trust towards the system (Laurie et al, 2010).

The core privacy concerns which emerge with biobanking and genomic research are; a) the possibility of personal information being breached and used in ways which might harm the donors, b) the possibility that an individual or group is stigmatised due to leaked information, and c) the possibility of intrusion into the private life of donors (Laurie et al, 2010).

Non-compliant research practice can lead to data leaks which threaten the participants' privacy. Pointed out below are the different dimensions of privacy; biobankers and researchers must consider all dimensions of privacy for the sake of best practice genomic research.

1. Physical privacy: this relates to storing and testing on genetic samples. If researchers store and test samples without obtaining consent, they would be breaching physical privacy.

2. Informational privacy: a breach of informational privacy happens when researchers misuse information, thus creating the possibility of genetic discrimination.

3. Decisional privacy: this is about research participants having control over what happens with the resources which they provide. Best practice biobank research puts research participants at the core of decision-making. This includes, but is not limited to, participants' choices regarding issues of data sharing with third-parties, as well as participants' choices about whether to maintain or withdraw participation (Laurie et al, 2010).

A substantial increase in privacy risk occurs when personal data is shared with third parties since the initial data receiver would no longer hold full control over information. Donor consent and anonymisation are two key safeguards against the harm which could be caused due to information sharing (Laurie et al, 2010). Absolute anonymity eradicates privacy risk, as through anonymisation research participants are completely unidentifiable and thus protection is ensured (Laurie et al, 2010). However, Laurie et al (2010) question the possibility of anonymising data when it comes to genomic research. Best practices call for safeguards which do not hinder research and which allow for maximised benefits, whereas anonymisation can indeed be problematic to the research process as it entails the irreversible disconnection between samples and the identity of donors (Laurie et al, 2010; Schlünder, 2018). The personal data linked to the biosamples are essential for most research projects, and such data can only be updated and revised if a link between the data and the donors' identity exists (Schlünder, 2018). Anonymity in itself can be unethical as it completely blocks off the possibility of contacting donors for re-consenting to further research. Furthermore, a genetic data set can never be completely anonymised as the genetic code of each individual is unique. These key

points prove why absolute anonymity is typically not a possibility for genomic research (Schlünder, 2018).

A trade-off between anonymity and access to private information needs to be made when the goal of achieving both is not plausible (Laurie et al, 2010). The solution is pseudonymity, or the use of coded links to the donors' identities. The General Data Protection Regulation (GDPR) defines the pseudonymisation of data as "personal data [that] can no longer be attributed to a specific data subject without the use of additional information, provided that such additional information is kept separately and is subject to technical and organisational measures to ensure that the personal data are not attributed to an identifiable natural person" (Bovenberg et al, 2017, p.7). Best practice genomic research uses pseudonymisation, which allows those who have access to the sensitive data to retrace donors if required, and also allows donors to adjust their consent preferences over time, if desired. Pseudonymity also allows for the possibility to give feedback to the research participants about any relevant research information (Schlünder, 2018).

In conclusion, Schlünder (2018) suggests that even though privacy risk persist since anonymity is not viable, it is in the public interest that commitments to anonymity do not hinder genomic research, especially since pseudonymisation is sufficient to safeguard research participants from privacy risk. Regulations such as the EU's GDPR allow for such research to be carried out without the hindrance of anonymisation, whilst allowing for maximised protection to research participants (Bovenberg et al, 2017).

2.4.3 The risk of genetic discrimination

Whilst laws and regulations are aimed at protecting research participants, and whilst legitimate research bodies are well-trained to conduct best practice research under high ethical standards, the risk of a data breach or a data leak remains possible (Laurie et al, 2010). A breach of privacy can directly harm research participants if it facilitates genetic discrimination. Two key institutions which may seek to use breached data for their advantage are employers and health insurance brokers; genetic information would equip both of these institutions with enough data to make selections of new employees or insurance customers on the basis of the genetic make-up (Gostin, 1991).

According to Oliver et al (2011), 28% of their research participants claimed that they feared that their participation in genomic research might lead to health insurance discrimination. Genetic discrimination is not only detrimental to the research participants but also inimical to public health, medicine and society in general (Gostin, 1991).

2.4.4 **The issue of time**

Functionalist sociologist Talcott Parsons (1951) speaks of social actors as being motivated by circumstances which lead to the "optimisation of gratification". Social situations are the result of social actors engaging in social action aimed at balancing the optimisation of gratification and the minimisation of deprivation (Parsons, 1951). Furthermore, Parsons argues that social actors have to constantly make decisions between immediate and deferred gratification. Certain circumstances offer greater levels of gratification if individuals were to postpone gratification to the future; therefore, social actors often deliberate between minimised immediate gratification or optimised deferred gratification (Parsons, 1951).

> "Action may be oriented to the achievement of a goal which is an anticipated future state of affairs, the attainment of which is felt to promise gratification: a state of affairs which will not come about without the intervention of the actor in the course of the event. Such instrumental or goal orientation introduces an element of discipline, the renunciation of certain immediately potential gratifications" (Parsons, 1951, p.48).

With regards to genomic research, immediate gratification is often not an option and therefore individuals can only be motivated by deferred gratification. This is not because actors actively choose to delay gratification for it to be greater, but rather because immediate results are generally impossible. Individuals are aware that the only way to optimise gratification and minimise deprivation is by giving research time to achieve tangible results. Immediate and deferred gratification in this regard are not mutually exclusive, but rather deferred gratification is the only choice.

Time is a key issue within biomedical research, and thus so is the concept of 'solidarity'. Illness and suffering create circumstances which require urgency, a need for an immediate solution, but paradoxically the development of new therapy takes several years and breakthroughs are commonly not made within the life-span of the donors, thus direct benefits for research participants are limited. Solidarity when donating biospecimens is key since direct benefits are not common in the short term. Data sharing is a way of accelerating the process thus allowing for better chances for the donors themselves. "It [usually] takes decades before statistically significant number of diseases arise" (Laurie et al, 2010, p. 330).

Additionally, throughout the long research process, as researchers recruit new participants, attitudes and perceptions of new and existing participants towards medical research and biobanking might change, leading to a collapse is the research structure. Consequently, preserving trust is crucial to the success of genomics and biobanks. "We simply do not know what the privacy challenges will be nor, indeed, what benefits will emerge from biobanks. [...]

The personnel involved suffer from a lack of informedness about the future; they have to trust that the project will be managed appropriately" (Laurie et al, 2010, p.331).

The longitudinal nature of genomic research creates challenges for keeping the donors of biospecimens informed. Carrieri et al (2015) point out that ethical considerations (particularly in consenting) should not be centred on current technology, as the technology we use nowadays will eventually become obsolete. Thus, participants should be informed about the possible use of unforeseen technologies. Oliver et al (2011) claim that 30% of their respondents (individuals taking part in genomic research at a University in Texas) feared the unknown. They were concerned about the risk of future research projects using their data in unpredictable ways, claiming that they fear the repercussions of not being able to know enough about how their genomic data, and other personal information, will be used in the future (Oliver et al, 2011).

In 2010, Laurie et al suggested that one way to combat such risk is by developing a mechanism of consenting which goes beyond a one-time informed consent, and indeed we are nowadays speaking of dynamic consent (Kaye et al, 2014; Mamo, 2019), a model which allows for continuous communication between researchers and researched, therefore providing a solution which combats the challenge of time. The interface, which keeps participants informed and updated, allows for the withdrawal of consent from past projects and for giving consenting to new ones, thus providing the research participants within greater control over their personal data. This process creates a sense of community which harvests the fundamental factor of trust (Laurie et al, 2010).

2.4.5 Domestic versus international research

> "Biomedical research is international in nature and is highly dependent on international networks and collaboration" (Tupasela and Snell, 2012).

Nationalism is a key topic in biobanking, precisely the issue of whether to keep biosamples and data local or go international. Tupasela and Snell (2012) found that when asked about national protectionism, respondents expressed that they were more willing to donate their biospecimens to local researchers as opposed to foreign researcher bodies. This raises a huge concern as genomic research has become significantly dependent on the sharing of data amongst biobanks across nations. The two main reasons for the inevitable need to share data are; a) the requirement for large pools of samples, and b) the scientific need to validate findings within multiple populations before drawing scientific conclusions (Tupasela and Snell, 2012).

A survey assessing the attitudes of the UK public found that data sharing on an international level enjoys significantly less support as opposed to data sharing on a national level (as cited in Laurie et al, 2010). The study conducted by Tupasela and Snell (2012) elaborates on this finding, as it distinguishes between the general public and patients; it appears that patients are significantly less likely to disapprove of international research since they are generally personally invested in the process. Most times members of patient organisations possess greater knowledge about the research process than the general public and tend to become lay experts, thus whether research is conducted nationally or internationally is less likely to matter for this group as they are well aware of the research process and perceive international collaborations as a means for greater hope (Tupasela and Snell, 2012; Boat and Field, 2011).

The benefits which emerge through international data sharing for the research participants, or members within their community, remain unclear, thus some level of hesitance amongst participants persists. The issue of trust is crucial here. It appears that when it comes to international research, most participants, especially non-patient participants, require greater transparency, further information (such as country, purpose and details of the institution), and the possibility for re-consenting (Tupasela and Snell, 2012). "Participating in a national publicly funded research projects produces more direct expectations that the benefits will return to the [local] public through public health care" (Tupasela and Snell, 2012).

2.4.6 Public versus private research

Another key issue in genomic research is the commercialisation of data. Critchley (2008) found that publicly funded scientific researchers enjoy greater trust levels over privately funded researchers. Tupasela and Snell (2012) found that Finns were much less ambivalent to donate to local private enterprises than to foreign public researchers, meaning that the issue of commercialism is less of a concern than the issue of transportation. Patients had optimistic perceptions toward commercial entities; what mattered to them was the end result, which is getting closer to finding novel cures or treatments for disease.

Nonetheless, it still appears that research participants tend to be more trustful of public researchers; they are more likely to give their samples to be re-used without re-consent to public research than to commercial researchers, whereas they commonly would want feedback and updates from the commercial sector to re-consent. Although they would ultimately be willing to donate to the commercial sector due to the dedication towards the ultimate goal, potential participants tend to consider researchers of public health as being more trustworthy than

pharmaceutical researchers. A risk-utility trade-off comes into play here; research participants would give consent to have their data shared with private pharmaceutical companies, but only because their desire for the development of new treatments or cures is more significant than any concern (Tupasela and Snell, 2012).

Research projects are often promoted to participants as a means of achieving scientific progress for the wellbeing of society or the common good, and since the donation of biosamples usually happens within the public sector, the role of the private sector is not always clear for research participants. “People have expectations, hopes and worries about the functioning of tissue economics - who should have access to tissues and research information and what benefits and risks are embedded in different ways of producing biovalue” (Tupasela and Snell, 2012). Research studies conducted by Ipsos Mori (2016) amongst Europeans indicate a sense of scepticism towards data sharing with commercial entities. Transparency throughout the whole process of participation is key for populations to become less suspicious of such collaborations (Goisauf and Durnová, 2019).

Professionals, in general, seem to recognise the importance of Private-Public Partnerships (PPPs), claiming that their key motivation for such collaborations lies in scientific advancements and social wellbeing, superseding economic motivations (Goisauf, 2019). Moreover, the majority of experts who participated in the study conducted by Goisauf et al (2019) expressed the importance of informing participants about such partnerships at the stage of recruitment.

2.4.7 **Incidental findings**

The fear of being exposed to information which is uncalled for about themselves or their family members seems to be common amongst those participating in genomic research (Oliver et al, 2011). Incidental findings happen when through analysing a genome sequence with the hopes of gaining knowledge about a certain disease, the researcher incidentally discovers secondary genetic information such as genetic variations and mutations (Powledge, 2015).

The Council of Europe's Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research (2005), Article 27, states as follows: "If research gives rise to information of relevance to the current or future health or quality of life of research participants, this information must be offered to them. That shall be done within a framework of health care or counselling. In the communication of such information, due care must be taken in order to protect confidentiality and to respect any wish of a participant not to receive such information".

Even though in some countries the law obliges researchers to return incidental findings, this practice seems to be a controversial issue which generates antagonism between genomic researchers. Consensus on the responsibility of researchers to return secondary clinical results to research participants is far from being achieved (Vaught and Lockhart, 2012).

Whilst some argue that researchers must provide research participants with information which emerges through the research process, especially if such information can impact on their health and lifestyle, others advocate that the purpose of medical research is to generate knowledge and come up with macro solutions and not to provide clinical care. Those who oppose the idea of reporting such findings further claim that research laboratories and clinical laboratories operate under distinctive standards, and thus it would be irresponsible and harmful to provide

research participants with results which could be invalid or "of unknown utility" (Vaught and Lockhart, 2012, p.9).

> "[The] variation is often of unknown significance for health and other kinds of well-being. So how should it be handled? […] At this still-early stage of genetic knowledge, it's often impossible to know just what effect a particular genetic variant will have, or if it will have any effect all" (Powledge, 2015).

In December 2013 the US Presidential Commission for the study of bioethical issues presented a report with the title 'Anticipate and Communicate'. The commission claimed that participants should be informed beforehand about the possibility of incidental findings and that the participants themselves should decide in advance whether they would like such additional information or not, should it exist (Powledge, 2015). Critics, such as UK geneticist Caroline Wright (2013), claim that whatever decision is taken by the participant, it is usually a decision made out of inadequate knowledge since ample genomic knowledge is required for participants to truly make an informed choice about whether or not to receive incidental finding information (Wright, 2013).

The Commission further denotes that reporting a finding which has no possibility of preventive action might be irresponsible as it can cause unnecessary stress for the participants without any corresponding medical benefit. In order to explain this, the Commission used as an example the incidental finding of misattributed paternity, claiming that in such cases, revealing such result to the participants would cause distress without direct medical benefit. However, Powledge (2015) begs to differ, claiming that it is incorrect to assume that this result would lead to no medical benefit. She recalls two cases in which she was involved, where through testing for a heritable disease (clinical testing) which the father was suffering from, it was revealed that both men had not biologically "fathered their apparent children", and where neither themselves nor the children knew about this fact. Powledge (2015) claims that such

situations are rather confusing for the clinician; “the docs handled the cases with superb nonchalance. They happily told the families not to worry, that the tests revealed the children were not at risk. They simply skipped explaining the reason why”. Powledge (2015) questions the ethicality of this approach.

Providing research participants with incidental results can be life-changing; however, it is often complex for researchers to confirm the validity of the incidental findings which emerge. Sometimes, or oftentimes, incidental research findings are futile with little to no clinical relevance, thus it might not always be rational to provide participants with the incidental findings which emerge (Carrieri et al, 2015). For instance, cancer marker genes are only an indication of a possible risk of developing cancer. The role of social and physical environmental factors might switch these genes on or off, making the issue even more complex and difficult to predict actual risk (Wilson et al, 2002).

Nonetheless, some ethics committees expect researchers to return a summary of the generated findings to the research participants. Ethics committees usually argue that participants preserve the right to know about all that emerges even if it is not “clinically actionable”. Sometimes participants may wish to know such information as a means to an end; maybe to obtain that “knowledge for its own sake” or to pass on the information to future generations (Carrieri et al, 2015).

According to Powledge (2015), the significance of incidental findings is uncertain. Whilst some experts claim that incidental findings are a certainty, especially when looking at whole genomes and exomes, other experts believe that coming across a significant incidental finding is rather unlikely and they further denote that “biomedical research [...] is not primarily about

[one's] own health but rather about potential health benefits for future generations" (Steinsbekk et al, 2013, p. 900).

Research conducted by Goisauf et al (2019) shows that researchers and other biobank professionals tend to be equivocal of the practice of returning incidental findings. It results that, amongst researchers, the option of providing information about the return (or the non-return) of incidental findings was the least frequently selected option amongst a list of possible topics to be raised on the informed consent form. Furthermore, a separate study conducted amongst biobanking experts in the EU showed that the main obstacles in returning such results were a lack of funds, a lack of organisation and human resources, as well as a lack of guidelines about the legal and ethical obligations of researchers (Budin-Ljøsne et al, 2016).

2.5 Striking a balance between risks and benefits

A constant risk versus benefit analysis exists to achieve goals for the greater good whilst safeguarding individual donors; there is a constant tension between facilitating and promoting the idea of research for the public interest, and responding to challenges also to protect the public interest (Laurie et al, 2010). Therefore, appropriately safeguarding individual donors is "in itself a public interest". Thus, the two factors are "not necessarily mutually exclusive" (Laurie et al, 2010).

One of the biggest challenges with genomic research is to strike a balance between the risks of the "inherent identifiability of DNA data […] with the utility associated with amassing genetic data for analysis. […] Participants are making a deliberate privacy-utility trade-off" (Oliver et al, 2011, p.112).

The right of individuals to be protected through confidentiality measures is in itself a public interest. Furthermore, Laurie et al (2010) claim that "this public interest is not an absolute one and it is often met, ironically, with countervailing public interest claims that seek to limit it" (p.321). Consequently, there might be instances where it would be required to hinder privacy with the aim of protecting the general public. This does not mean that privacy rights aren't protected, but it means that ensuring privacy rights does not imply absolute rights. Ethical scientific research using biobanks is conspicuously aimed towards the public interest, and the benefits which emerge from biobanking might be returned to individual research participants, even though not necessarily (Laurie et al, 2010).

The dialectic between promoting ends which benefits society and preventing outcomes which harm society, or individuals within it, provides a healthy environment for biobanking as it encourages professionals to seek for ways which allow the research process to keep going whilst developing mechanisms to counter risk. Nonetheless, participants must be informed that risk persists, even if it is minimal (Laurie et al, 2010). "The public interest should include acts or conduct which further society's better interests as well as providing adequate protection of the general public from harm" (Laurie et al, 2010, p.324).

A study which displays the trade-off made by participants is that conducted by Oliver et al (2011) with individuals who were at the time participating in genomic research at the Baylor College of Medicine in Texas. This research revealed that there is a significant difference between data sharing preferences and data sharing decisions. Whilst participants were likely to claim that they have concerns about the sharing of their data with third parties, their ultimate decision was highly likely to be positive towards data sharing. The reason for this was that risks were considered by the participants to be "less concrete [...] and were largely outweighed by purported benefits", thus their sensible decision was to go ahead with the sharing of data,

with the most common motivation being for research advancement for the greater good. “Most are willing to share their data despite concerns regarding government oversight, privacy and confidentiality, and profiteering or misuse of data” (Oliver, 2011, p.107).

Laurie et al (2010) suggest three tests which should help researchers maintain public interest whilst ensuring the least harm possible: a) “test of effectiveness”, which is coming to terms about what the ultimate goal of a particular action is; b) “test of necessity and subsidiarity”, which ensures that an action is indispensable to achieve the aim and that no “less intrusive” measure is possible; and c) “test of fair balance”, which calls for a strict balance between the aims achieved and the harm caused through that achievement (p.324).

Research is in itself a means of safeguarding the interests of the public through the provisions of adequate healthcare, yet, the research community must concurrently provide the means to proportionately protect individuals’ rights. A fair balance is achieved through “sound scientific, ethical and legal principles, and committing responsibly to benefit-sharing” (Laurie et al, 2010, p.333).

2.6 Willingness to participate in genomic research and biobanking

Most genomic research is impossible without the donation of human biospecimens and thus, research to observe patterns in willingness to participate in genomic research and biobanking is crucial.

Research conducted in Jordan by Ahram et al (2014) revealed that the major factor which determined a lack of willingness to participate was the practice of not returning personal results. Forty-seven per cent (47%) of the sample claimed that research which lacks to provide

feedback about the personal health of participants would harm their willingness to participate, this finding was especially common amongst females in the sample. Limited or unavailable information about the type of research to be conducted also appeared to be a factor which correlates strongly with a lack of willingness to participate (45%). Some Jordanians who participated in this research also showed distress to donate their samples to biobanks due to access to personal medical information (9.5%) (Ahram et al, 2014).

In the predominantly Islamic country of Jordan, the factor which encouraged most willingness for participation was religious permission, with 61% of the sample considering this to be a determining factor. Permission to withdraw from the research at any point also correlated with willingness to participate in genomic research and biobanking (Ahram et al, 2014). Despite the factors which trigger unwillingness, Ahram et al (2014) concluded that the vast majority of their sample would generally be willing to participate in biobanking.

It has been revealed that respondents are significantly more likely to be willing to donate their own blood, saliva or urine, rather than the organs of their deceased family members. The uncertainties linked to research on embryos (44%) and international research (35%) commonly raised concerns amongst potential donors, alongside the possibility of stem cell research, cloning and genetic engineering. This analysis also shows a pattern of objection towards research projects which deals with sensitive taboo topics such as intelligence, sexuality and mental illness (Domaradzki and Pawlikowski, 2019).

The willingness of respondents to participate is highly dependent on information; it appears significantly that respondents are more likely to accept to participate when they know who is conducting the research, what the purpose of the project is, where it is being done, how the research happens, as well as who has access to the information. The feasibility to withdraw

consent also tends to lead to positive attitudes towards donating (Domaradzki and Pawlikowski, 2019). In contrast, limited research information discouraged participation. Concerns of a data breach, discomfort with the invasive modes of sample donation (such as when needles are required), incidental genetic findings and the fear of discrimination are the key factors which tend to have a negative impact on willingness to participate (Domaradzki and Pawlikowski, 2019).

In conclusion, the literature reveals that willingness to donate biospecimens is significantly higher than unwillingness. This has been confirmed by studies in Europe and beyond, from studies conducted in the UK, to China, to Saudi Arabia, Finland, Sweden and various other countries (Domaradzki and Pawlikowski, 2019).

2.7 Ethically sound genomic research and biobanking

It is imperative that research participants are respected and are granted full protection over their rights and well-being (Vaught and Lockhart, 2012). Thus, ethical and legal considerations are central to genomic research and biobanking, to safeguard research participants, whilst ensure transparency which ultimately perpetuates trust.

Traditionally, participation in biobank research involved a onetime encounter between researchers and research participants, even though samples continue to be used for decades. The idea of one-time consent is highly questioned by critics who claim that communication between researchers and participants must be more frequent, for the benefit of both parties (Mester et al, 2015).

Furthermore, it was not unusual in the past for samples to be taken without consent, particularly from new-born babies; blood samples were taken for clinical testing for specific diseases, which were then banked for further research. This led to court cases in the USA where several parents objected to this being done without informed consent. Explored by Vaught and Lockhart (2012), it appears that most parents were not against the practice, but rather against being uninformed, as one mother who had taken the case to court claimed:

> "To me, this whole thing was about consent. If they had asked me… I probably would have consented. The fact that it was a secret program really made me suspicious of the true motives, there's no way I would consent now" (as cited in Vaught and Lockhart, 2012, p.7).

Furthermore, simply asking research participants for consent is not sufficient; best practice recommendations as to when, where and most importantly how consent is given are indispensable for ethical research. Various studies suggest that participants often have difficulty to respond when asked what the research they are participating in entails. The reason for this could be attributed to the fact that concepts of genomics are not easily grasped and fully understood by the general public, especially when the general scientific knowledge of the public is poor. Furthermore, informed consent documents are usually rather lengthy and include scientific details which are not generally understood by the general public. Thus, one needs to ask questions about how much information research participants can absorb, and how it shall be presented to them, to ensure that they make a valid and conscious informed consent (Robinson et al, 2013).

2.7.1 Informed consent

Informed consent stimulates trust (Ducournau and Strand, 2009). In general, it is an ethical standard which is required for any type of research which involves obtaining data through human interaction and/ or whenever private data about research participants is needed (Vaught and Lockhart, 2012, p. 6).

As defined by Faden, Beauchamp and King (1986) informed consent involves "(1) disclosure of relevant information about the study, (2) in a way that can be understood by potential participants, (3) so that they can make a voluntary and informed decision to participate". Joffe et al (2001) add that informed consent is not only about *feeling* well informed but essentially about *being* well informed. Sometimes research participants may *feel* they have acquired enough information, but this does not necessarily mean that they have truly understood the research process and its benefits and implications (Robinson et al, 2013).

Robison et al (2013) conducted research with participants who were at the time enrolled in an ongoing research project. Sixty-two per cent of their research participants were either not conscious that they were participants of genomic research, or else claimed that they were not sure. 55% of the research participants failed to answer correctly when asked about who has access to their genetic information and were generally overestimating the restrictions of data sharing (Robinson et al, 2013). Robinson et al (2013) claim that such lack of awareness by the research participants either results from the difficulty of the general public to grasp the concept of genomic research which is a rather complex concept or as a result of inadequate communication between researcher and researched at the stage of informed consent. A plausible solution is the development of less complex and more straightforward informed consent documents. Providing research participants with too much information not only

prevents them from grasping that additional information, but worse than that, the most fundamental concepts end up being disregarded too (Robinson et al, 2013).

Several scholars and researchers have questioned the idea of informed consent in the field of genomic research. Giving truly informed consent, when using human biospecimens, is rather impractical due to the unknown future of medical research, the rapid technological developments and consequently the unknown potential future uses of samples, all lead to unknown future risk (Vaught and Lockhart, 2012).

A recent study conducted by Goisauf et al (2019) identified the most and the least common items to be present on informed consent forms, according to professionals. The most common items include information about the right to withdraw or amend consent at any moment, information about the aims and purposes of the research project, general information about the biobank and about those responsible for holding consent data, and contact details of the biobank, respectively. In contrast, details on research projects conducted by the biobank and information about the right to complain about ethical issues with authorities seem to be fairly uncommon (Goisauf et al, 2019).

2.7.2 The evolution of consenting: from broad to dynamic

Traditional consenting was an "all-or-nothing" kind, where participants would agree to participate in genomic research whilst also agreeing to the sharing of their medical and genetic data (Robinson et al, 2013). With broad consent, research participants get to decide whether or not their data is shared with third parties and/ or is reused for future research projects (Robinson et al, 2013).

When this kind of consent is used, research participants are notified at the start about the constantly developing nature of biobanks, and thus about the need for the re-use and sharing of samples for potential future projects, which they agree to as part of the 'one-time' consenting process. Broad consent best practice requires adequately informing the participants that biospecimens shall be utilised for a variety of research projects, most of which cannot yet be foreseen, and that consent will apply to all research projects, without the possibility of selecting which research projects to participate in and which not to (Vaught and Lockhart, 2012).

Tiered consent allows for an added layer of control, although still limited, for participants as it is "a consent model in which participants are given a set of options allowing them to select how they want to participate in the research" (NIH, 2015). Participants get to choose between three main options: a) data sharing amongst third parties and reuse of data whenever required (without re-consent), b) restricted data sharing and reuse of samples or c) no data sharing (Robinson et al, 2013).

In 2011 Oliver et al had suggested the use of tiered consent for small scale studies which are not dependant on data sharing, claiming that tiered consent provides participants with more control over who can access their genetic data when compared to traditional approaches to consent. However, Oliver et al had added that tiered consent is not practical for genomic research studies which are largely dependent on data sharing. "In studies where the primary goal is to create a community resource (e.g. a biobank), data sharing may be a condition of participation and so tiered consent would not be practical" (Oliver et al, 2011, p. 113). Back in 2011, Oliver et al had suggested that future projects should come up with a new way of consent, one which makes giving consent for the latter kind of research more practical.

A more recent form of consent is dynamic consent**.** This is a concept which aims to create better communication between the key stakeholders, namely researchers and researched, and ultimately provides a more feasible, practical and ethical way of giving consent to genomic research, including data sharing and reuse of data.

In a day and age where the world has become digital, with constant IT innovations, mechanisms of paper-based informed consent are simply outdated, especially in fields such as that of biomedical research (Teare et al, 2015).

The field of genomics is constantly increasing its research capabilities as a result of the rapid technological advancements. In recent years, the approach of dynamic consent was introduced as a means to help ease communication between researchers and research participants and thus enabling the latter to feel in control and informed, thus strengthening their motivations (Kaye et al, 2014).

As medical research became more dynamic, the traditional idea of scientific experiments with a start and end date is no longer relevant, nowadays medical research is ongoing, dynamic and constantly evolving. Thus it has been suggested that the social contract between researchers and researched must evolve concurrently in order to safeguard the rights of research participants whilst facilitating the research process (Kaye et al, 2014).

> "In the case of biobanks where there are multiple researchers and research projects, it is difficult to obtain informed consent for all future research uses at the time of recruitment. [...] Re-consenting is costly and time-consuming, and difficulty in locating people can result in high drop-out rates" (Kaye et al, 2014, p.141).

By definition, dynamic consent is an approach which involves research participants in continuous decision making regarding the use of their personal data. It allows for as much

participation from research subjects as they please and allows for alteration to consent choices over time (Teare et al, 2015). Scientific research pleads for 'respect for persons' and dynamic consent reaches this aim to its full potential as it gives "individuals as much choice and control in what is done with their personal information and material as is reasonably achieved" (Kaye et al, 2014, p.142). With dynamic consent, data subjects are placed at the core of decision making; participants are no longer passive, they are consulted about data sharing, they can re-consent for future projects, they can withdraw from a research project at any point in time, and they get to have access to information about any developments which arise from the research projects for which they would have donated their tissues (Teare et al, 2015).

The approach of dynamic consent depends on the use of an IT interface which makes the data subjects more in control, valued, involved and most importantly informed, as it breaks the hierarchy which previously dominated the researcher-researched relationship (Feeney et al, 2018).

This digital means of consent also allows for a practical way in which participants' consent agreement can easily be shared to third parties along with data samples (in the event of data sharing amongst researchers and/or biobanks), whilst having all data encrypted in a manner which allows for full confidentiality (Kaye et al, 2014).

In a study conducted by Robinson et al (2013) it was concluded that "participants expressed a desire to be involved in the decision about data sharing. They felt that the best way to respect research participants was to be transparent, provide them information and offer them choice" (p.8).

Since with a dynamic consent interface data is re-used with the consent of the data provider, the issue of re-consent is dealt with most effectively. Dynamic consent allows consent

preferences to be altered, and if new ethical concerns arise, the researcher would be able to consult with the research participants, giving them the option to desist or update their consent preferences to be aligned with the new situation. Thus, dynamic consent allows for samples and personal medical information to be reused in the most ethical and lawful manner whilst providing optimum research efficiency (Kaye et al, 2014).

It could be argued that dynamic consent is not a replacement for broad consent but rather a developed form of broad consent. Dynamic consent ensures that participants would still be able to choose to give broad consent for all future research without receiving updates, however, they would remain entitled to 'opt-in' for specific studies at any time. Equivalently, if participants, at some point, decide that they want to decrease their activity, or stop interacting completely, they would be able to do so (Teare et al, 2015).

The GDPR gives research participants rights in terms of control over how their personal data is processed. Recital 7 states that "natural persons should have control over their own personal data". The GDPR has raised the bar for best practice genomic research and biobanking, and dynamic consent deals with the new expectations in the most effective way (Mamo et al, 2019). The possibility for communication between researchers and researched allows for a simple, clear way to keep up with changing legal requirements and ethical considerations. With a dynamic approach to consent, there is a shift from a one-time signature scheme to a long-term interactive relationship between researchers and data-subjects, thus ensuring greater transparency in the research process as well as public trust (Robinson et al, 2013).

2.7.3 Criticism of dynamic consent

Steinsbekk et al (2013), are critical of the implementation of dynamic consent claiming that it is not simply a development of broad consent but rather a ploy. They claim that one must not assume that research participants would be motivated and willing to consent dynamically, and remain actively involved in the research process.

A counter-argument to this criticism is that dynamic consent will not oblige research participants to constantly remain active but it will provide them with that option. As stated above, participants could still choose broad consent without receiving any updates, but they would remain entitled to opt-in should they ever change their mind. Likewise, if research participants decide that they want to limit their activity, they would be able to opt-out (Kaye et al, 2014). Furthermore, research shows that donors tend to prefer to remain involved and receive updates about any findings which might emerge from the research project they would have participated in (Mester et al, 2015). A study by Mester et al (2015) even revealed that a significant 97% of the sample stated they would be pleased to provide researchers with personal health updates now and again, should such data be useful for the research process.

However, the criticism of Steinsbekk et al (2013) goes further as they perceive dynamic consent as "morally problematic". As opposed to broad consent, which asks for re-consent only in the case of a particularly controversial or morally-sensitive issue, dynamic consent gives research participants the right to deny consent for any reason. Steinsbekk et al (2013) claim that it is an individualistic approach to consent which in essence goes against the approach of solidarity. Biobank research "carries the potential for important medical breakthroughs and beneficial medical inventions", and thus it can be unethical "if consent procedures unnecessarily reduce or prevent these opportunities" (Steinsbekk et al, 2013, p.901).

This critical perspective suggests that a constant interaction with the participant is not what matters for ethically sound research. Passive research participation can be just as ethical and sometimes even morally superior as it allows for medical research to reach its full potential for the greater good. Biomedical research best practice requires participants to be well aware of the information provided to them; it is not about the frequency of information but about the relevance and understanding of such information. Informed consent is at the core of this debate, however, providing research participants with more information does not necessarily mean they are better informed, it is the relevance of the content provided with constructs informed consent (Steinsbekk et al, 2013). Participant autonomy is not necessarily achieved through dynamic consent. Just like with the common practice of passively accepting the *'terms and conditions'*, research participants might make "autonomous" but "ignorant choices" (Schmietow, 2016, p.204).

This critique of dynamic consent hinges on the fact that whilst dynamic consent largely focuses on individual control, biomedical research relies on numbers and collaboration, thus the idea of putting "private interests" at the core of the research process can be a red herring which diverts the focus from the ultimate goal of genomic research (Steinsbekk et al, 2013).

In sum, the debate of broad versus dynamic consent is rooted in the main point that dynamic consent encourages "bottom-up governance" of data, placing the interests of the individual at a focal point, while on the contrary, broad consent grants "top-down governance" and ensures "genetic solidarity". According to critics of dynamic consent, whilst the former might provide better individual autonomy, the latter allows for greater opportunities for improved health for the research participants themselves, as well as the public in-general and future generations (Schmietow, 2016, p.205).

2.8 The General Data Protection Regulation in genomic research and biobanking

Consent does not safeguard research participants from breaches to their privacy, thus security over participants' information is not automatically ensured through a consent agreement. Rather, informational privacy is ensured through laws aimed at protecting personal data (Laurie et al, 2010); and this leads us on to the General Data Protection Regulation (GDPR). The GDPR is not an ethical guideline but a pan-EU legal means which safeguards the rights, dignity and safety of research participants whilst allowing for medical research to reach its full potential (Bovenberg et al, 2017).

As a consequence of the rapid developments in technology, genomic research has taken a huge leap forward, while lawmakers try to keep up with the risks which emerge concurrently with such developments (Oliver et al, 2011). The greatest risk of biobank participation is the risk of breach of privacy and confidentiality. Since providing ethical considerations for best practice research might not be enough, clear legal obligations aimed at protecting participant information are crucial (Vaught and Lockhart, 2012). Until a few years ago, different countries applied different legal mechanisms with the common aim of protecting "the same kind of privacy interest" (Laurie et al, 2010), however, since May of 2018, the European Union implemented the GDPR. Transparency and accountability lie at the core of this regulation.

The GDPR has had a direct impact on biobanking and genomic research since biobanks store human tissue alongside personal data including medical histories and genetic information. Data subjects hold several rights, including the "right to consent, to information, to access, to rectification, to erasure (the right to be forgotten), to restrict processing, to data portability and

to object" (Bovenberg et al, 2017, p.5). Research bodies are obliged to observe these rights and ensure that research participants are safeguarded. However, the GDPR allows for restrictions on the rights of data subjects as long as their identity is protected. Restrictions on such rights are strictly allowed when the implementation of such rights is impossible or will seriously destroy the objectives of the data processing. Whenever these rights can be fulfilled, they must be fulfilled. The GDPR principle of data minimisation strongly applies to biobank data. This principle states that organisations should only store and process the personal data which is required to fulfil the research purposes. Personal data must be restricted to what is indispensable (Bovenberg et al, 2017).

When it comes to broad consent, the Regulation recognises that research purposes might be bound to change and that establishing an ultimate aim at the time of data collection is not always possible. Thus, the regulation allows for broad consent, as long as data subjects have the right to freely decide in which specified areas of research they are willing to contribute (Bovenberg et al, 2017).

As for minors aged under sixteen, consent from caregivers is expected as the GDPR recognises that children might not be well aware of the risk that could arise from participation (Bovenberg et al, 2017). Although it must not be taken for granted that adults are always well-equipped to think about the possible implications of participation. In their research with genome research participants in Texas, Oliver et al (2011) found that a significant number of participants had serious difficulties to understand certain complex concepts listed on the consent form. This finding seems to be congruent with findings from other similar studies (Oliver et al, 2011). The lack of simple terminology is seriously unethical because it leads participants to sign a supposedly informed consent form without necessarily acquiring enough information. The GDPR establishes regulations on this matter as it states that consenting must be given freely

and from any coercion, and that it should be informed and specific. Thus, consent forms must be clear and unambiguous, using plain language (Bovenberg et al, 2017).

2.9 Concluding the literature review

Sociological literature about the donation of biospecimens for research and biobanking might not be excessive. However, sociological theory has deep roots in areas which relate closely to biobank participation. The theories of risk and trust are key to contemporary sociology and using these theories to understand the mechanisms behind the concern, or the lack of concern, of individuals in donating their biospecimens, sheds light on the reasoning behind their decisions. The key outcome from the literature is that the topic in question must be analysed holistically and in a comprehensive manner through a chain of ethical, legal and social considerations.

3. Research Design and Methodology

3.1 Objectives and research questions

The goal of this research is to gain a broad perspective of the awareness of biobanking amongst the Maltese general public, as well as a deep insight into the attitudes and perspectives of the key stakeholders in genomics and biobanking, namely patients, parents of patients, members of the general public, representatives of a patient support group, research experts and the biobank manager.

The research questions driving this project are as follows: What is the level of biobank awareness of the Maltese general public? What are the benefits of participating in genomic research? What motivates potential participants? What are the risks associated with participating in genomic research and biobanking? What are the attitudes of stakeholders towards issues of risk and trust?

3.2 Research design: mixed methods

The research design for this dissertation consists of mixed methods where public awareness is measured quantitatively via survey data, and attitudes of stakeholders are explored qualitatively using focus groups and in-depth interviews. Quantitative and qualitative methods are not to be considered as two conflicting paradigms (Gunasekare, 2015) but rather as complementary research methods which, when combined, can provide deep and broad data (Schoonenboom and Johnson, 2017).

Research conducted on a pan-European level reveals remarkably low levels of awareness of biobanks; a research project conducted by Gaskell and Gottweis (2011) indicates that a vast majority of 67% of Europeans had not heard of biobanks, let alone were aware of their

functioning. Furthermore, data which focuses specifically on awareness in a Maltese context was unavailable prior to the study conducted for this dissertation. Achieving the main objective of this dissertation, that of looking into attitudes and perceptions, required acquiring background knowledge about the local awareness on a general level.

A questionnaire was used to gather preliminary quantitative data assessing the level of knowledge about biobanking in Malta. The findings were used to develop effective tools for the generation and collection of qualitative data with key stakeholders that followed. These included a set of informative video clips used during the qualitative focus group sessions to better inform participants about the topic in question, and thus achieve a better insight into their attitudes and perceptions.[4]

3.3 Quantitative design: preliminary research exploring awareness of biobanks

The hypothesis prior to data collection was that the vast majority of the Maltese general public is unaware of the existence of biobanks for biomedical research. This hypothesis derived from literature which indicates a clear low level of awareness amongst pan-European citizens (Gaskell and Gottweis, 2011), and also from the observation that whenever the topic of this dissertation was discussed with individuals outside of the Centre for Molecular Medicine and Biobank, the vast majority asked for further information about what biobanking is.

[4] Further information about the content of the clips shall be provided in the section on 'qualitative design'.

3.3.1 Population and sampling technique

The target population for the preliminary quantitative research consisted of citizens of the Maltese islands aged 18 and over. A survey questionnaire was conducted amongst participants (n=387) using a stratified sampling technique based on gender and educational background, with the size of each group being proportionate to the size of the same group within the actual population. The National Statistics Office provided quantified data about the size of each group within the Maltese general public (last updated in 2017). This data was then used to determine the quotas for the two categories; gender and educational background.

The sample size allowed for a confidence interval of 95% with a +/-5% margin of error. Table 1 below shows the quotas which were used to stratify the sample proportionately to the actual population.

59.8% of 387 participants	**231 participants**	Individuals who have completed primary or secondary education. Also includes persons who have never attended an educational institution.
22.4% of 387 participants	**87 participants**	Individuals who completed post-secondary education.
17.8% of 387 participants	**69 participants**	Individuals who graduated from tertiary education.
100% of 387 participants	**387 participants**	**Total**

Table 1: Sample stratification on the basis of education (Desira and Martin, 2018)

3.3.2 **Quantitative data collection and data analysis**

The process of data gathering happened over a span of twenty days and participants (n=387) were recruited from public spaces at Valletta bus terminus, at the University canteen and quadrangle and at Ċirkewwa ferry terminal. Participant recruitment was conducted in satisfaction of the stratification system mentioned above.

Data was collected using a straightforward survey questionnaire composed of 4 questions (see Appendix B). The questionnaire starts with a direct question which asks whether the respondents know what a biobank is or not. Based on the answer of the initial question, a follow-up question was administered asking what a biobank is, in the case of respondent claiming to know what a biobank is. A question which invites respondents to speculate about what a biobank could be followed in the case of respondents who had claimed that they are not aware of what a biobank is. As for those who mentioned biobanks for the purpose of biomedical research in their answer to the follow-up question, a third question which asks for information about the source of their knowledge of biobanks followed.

After the completion of the process of data gathering, the data from each survey questionnaire was inputted into SPSS and then analysed through the generation of descriptive statistics. The statistical data which emerged through this research reveals the number of individuals in Malta who know what a biobank is, as well as the speculations of individuals about what the term represents. The statistical data also reveal relationships between levels of awareness and the two factors of gender and educational background.

3.3.3 Research results published by 'Xjenza'

The results which emerged from this quantitative research have been published by the peer-reviewed journal 'Xjenza' which is the scientific journal of the Malta Chamber of Scientists (Desira and Martin, 2018). The decision to publish these results prior to the submission of this dissertation emerged from the urgency for awareness data within the local context, for the sake of implementing an information strategy accordingly at the earliest opportunity.

3.4 Qualitative design: exploring attitudes towards the risks and benefits

The key aim of this work is to explore the attitudes of local key stakeholders towards participating in genomic research and biobanking. The assumption is that social actors develop meanings and thus attitudes, based on the way they "interact with the world around them". The attempt is to "understand phenomena through accessing the meanings participants assign" to circumstances (Orlikowski and Baroudi, 1991). In contrast to quantitative methods of research, interviews and focus groups tend to provide a deeper understanding of the attitudes and perceptions of a smaller group of people. These methods do not produce data which reflect about the whole population, however, they provide in-depth data about the beliefs, motivations, fears and thoughts of the research participants, opinion and feelings in a raw and intense manner (Neuman, 2006). The idea that 'proper' research is to be conducted using quantitative methodologies, to produce scientific data, excludes the opportunity to analyse social life through a lens which exposes the deeper corners of social phenomena, as lived by people in their daily lives. Qualitative research provides this opportunity (Silverman, 2013).

Opting to ask questions of how participants feel about specific issues, and of what they have experienced, or else how they would react to specific scenarios, has provided the means for generating good data, which I believe would not have emerged had these data been gathered

numerically. The preference was to seek to understand attitudes and perspective towards such a complex issue by providing real-life scenarios and engaging in conversation which results in interpretations which are valid and authentic, as opposed to generating variables and statistics. As for the focus groups, when one considers that the topic of genomics and biobanking does not commonly feature in daily life conversation, due to low levels of awareness (Gaskell and Gottweis, 2011), the need for participants to be provided with the opportunity to engage in an in-depth qualitative discussion allowed for them to dig deeper and develop more profound thought processes. "Focus groups are, then, an ideal method for exploring people's own meanings and understandings of health and illness" (Wilkinson, 1998, p.329).

3.4.1 Population definition and sampling technique

The key stakeholders being considered for the qualitative phase of this research are; a) the research participants or potential research participants namely patients, the parents of patients and members of the general public, b) research experts, specifically researchers and the biobank manager, and c) patient support group representatives.

The inclusion criteria were set after thoroughly reviewing the literature. Population banks are dependent on healthy individuals from the general public, whilst clinical banks store the data of patients, and sometimes of their respective family members. Hence, the inclusion of both groups was essential, since genomic research and biobanking depend on patients and healthy individuals alike (Marko-Varga, 2013). Being a current or past participant in genomic research and biobanking was not a prerequisite for participating in the focus groups since the attitudes of potential participants are just as valuable. The mix of both participants and non-participants in the focus groups resulted in a broader perspective towards the topic. The research experts, including three researchers from the Centre for Molecular Medicine and Biobanking (CMMB)

and the biobank manager at the University of Malta, were interviewed in search of comparing and contrasting the perspectives of the experts with those of non-experts, as well as for an insight about the practices conducted. Representatives of a patient support group were also indispensable for this research as they typically serve as a bridge which connects the researchers and the patients. A total of four focus group sessions and six expert interviews were conducted for this research.

"Focus groups generally utilize convenience sampling. The sample for a focus group has individuals with characteristics of the overall population and can contribute to helping the research gain a greater understanding of the topic" (Nagle and Williams, 2013, p.3). Thus, a combination of quota and convenience sampling is typically used. Each focus group in this study was structured based on specific criteria; one group consisted of patients, another of parents of patients, a third group was made up of members of the public who have attained up to a post-secondary level of education, whereas the final group was composed of members of the general public who have accomplished a tertiary level of education.

The recruitment for the focus group sessions happened through social media platforms and also through the databases of the Malta Health Network and the National Alliance for Rare Diseases. The two organisations were provided with a recruitment letter (see Appendix D) which was distributed to all of their subsequent members. Some of the patients were not recruited through the patient organisations, and like all of the non-patients were recruited through a call on social media. A brief Facebook post was uploaded on a personal Facebook account and several Facebook groups, and individuals who expressed interest were then sent the detailed recruitment letter which provided a deeper insight into what participation entails.

Convenience sampling was also used for selecting participants for the expert interviews, excluding the biobank manager, since the role is occupied by one individual within the local

context. An invitation to participate was offered to researchers at the Centre for Molecular Medicine and Biobanking, and the sample was formed by those who accepted. As for the support group representatives, a key organisation was identified and requested to provide two interviewees for this research. All expert interviewees were invited to participate through a formal email letter (see Appendix H).

3.4.2 Qualitative data collection

Expert interviews

The fact, that at the time of designing this research, I was already working at the CMMB, exposed me to the cultural dynamics which exists amongst research experts. Consequently, I quickly realised that for generating valuable data from the experts, I had to opt for one-to-one interviews to create a safe environment where they would feel at ease to open up freely without having to consider how other experts would react or interpret one's comments.

In fact, one of the research experts was reluctant to be audio recorded. It can be analysed that the expert perceived the recorder as a weapon of power, and thus preferred to have full control by sending texted responses to the interview questions. This reinforces the presumption that experts are more likely to feel at ease, under circumstances of absolute confidentiality, since a sense of mistrust amongst professionals seems to exist. This notion shall be further discussed in the analysis chapter.

After an assessment of the literature, I developed a set of core issues and themes to guide me throughout the data collection process (see Appendix J). Prior knowledge was essential for the formation of an interview guide to be used with the experts. Having no medical background required ample reading to be equipped to carry out such interviews. As they discussed

challenges regarding incidental findings, data sharing, and other technical issues, foreknowledge was essential to making the necessary follow-up questions, as well as for raising key topics for discussion. Nonetheless, prior knowledge was not used to limit the discussions in any way, and instead, all conversations were encouraged to flourish with the aim of introducing new themes and patterns which had not been previously observed in the literature.

Focus groups

While I believed that the experts would have felt more comfortable if they were in a confidential discussion with just the interviewer, I presumed that the other stakeholders would be more at ease in a group setting. Considering that my preliminary research flagged a minimal level of public and patient knowledge regarding the risks and motivations associated with genomics and biobanking, it was a prerequisite to provide an environment where participants felt comfortable about not being well-informed (informational videos were shown to all focus group participants to enhance their knowledge on the topic, as will be described below). As participants realised that fellow participants too were not very knowledgeable, they immediately felt more confident to contribute to the discussion. In a focus group, as opposed to an interview, participants tend to feel empowered because of the minimised hierarchical dynamic (Pini, 2002).

Furthermore, a lack of knowledge on the research topic makes it rather difficult for one person to think of the implications or the benefits associated with genomics and biobanking, and therefore by individually putting their little bit of knowledge or their personal opinion on the table, all participants were contributing to trigger the discussion. In fact, the focus group sessions lasted between two and three hours, where an in-depth discussion flourished in each group, producing highly relevant and valid data.

The aim of the focus groups was not to gather a group of people to conduct five or six interviews at one go, but rather to create a discussion which generates data from the ways the participants interact with one another (Wilkinson, 1998). The intention of focus groups is not to depict objective facts, but rather the value lies in deep recollections and their interpretation (Neuman, 2006). The collective experiences make the "invisible visible", as focus groups provide the opportunity for reflexivity as they facilitate challenging the status quo (Pini, 2002).

Homogeneity within focus groups is typically preferred (Krueger, 2002). People who share similar experiences tend to encourage each other to speak and create less conflict, thus, polarity should generally be avoided. Evidently heterogeneous people, such as patients and non-patients, were separated to avoid distorted and biased responses. Disagreements amongst homogenous groups persist, and that is essential, however, sharing a similar lived experience provides a safer environment for opening up (Corfman, 1995), particularly in the case of patients and the parents of patients.

The four focus groups consisted of 4 to 6 participants per group, with a total of 21 participants. According to Krueger (2002), this is an ideal number of participants per group as it allows for the creation of a discussion with high levels of interaction, whilst ensuring that the moderator manages to retain control.

An approach of combining inductive and deductive reasoning was adopted. The focus group discussions started off using a guide sheet (see Appendix F) which pointed out the key points raised in the literature, and informative video clips which were shown to the participants, nonetheless, these were strictly used as means of enhancing the discussion and placing key topics up for debate. Consequently, focus group participants were encouraged to elaborate and take the discussion wherever they felt was important. In fact, several themes which had not

been encountered in the literature, emerged in the focus group sessions, adding more value to the data. The use of existing theories for analysing the newly generated data allowed for more richness, however, the freedom of formulating theories based on patterns of observation amongst the participants and between them added richness to this research project.

The use of think-back questions (Krueger, 2002) allowed for the participants to share narratives of their lived experiences within the medical system. Furthermore, "what would you do" questions over "what do you think" questions allowed for participants to place themselves in specific scenarios, and thus, even if they had not yet participated in genomic research and biobanking, they still came up with reasonable motivations and concerns.

Throughout the four sessions, as a researcher and moderator, I consciously tried to avoid observer interferences. The initial moments of the session were the most crucial. I consciously created a permissive atmosphere, to improve the likeliness of generating valid data (Krueger, 2002). At the start of the session, participants were immediately informed that there are no right or wrong answer to the questions being administered, and that their opinions would always be fruitful to the research (Krueger, 2002). I immediately sought to make the research participants feel at ease and a good rapport was built, nonetheless, control was maintained to prevent individual participants from dominating the discussion. As a moderator, I mildly held control over the groups, without being obtrusive.

A research diary was used to write reflections and jot down field notes right after the participants left, which was later useful for analysis. "Darwin's work is a testament to the power of two of the most basic tools used in research, the pencil and paper" (Silverman and Patterson, 2014, p.41).

It was decided not to video record the sessions as the use of cameras can make participants feel anxious, inhibiting their participation, and also raises ethical dilemmas as participants are more easily identifiable (Krueger, 2002). Thus, sessions were audio-recorded and immediately transcribed so as to recognise the voices of participants, for the sake of clarity and accuracy.

Video clips for enhancing the process of data collection

The scientific process, despite being "a serious matter", must not be seen as a rigid practice which cannot be creative (Colucci, 2007). Bloor (2001) insists that the use of at least one focusing exercise is indispensable when it comes to focus group research. The use of stimuli to engage research participants does not only make the process of data collection more enjoyable but also more fruitful. Colucci (2007) claims that such stimuli allow for the production of enriched data as they attract greater attention from participants, whilst composing an environment which encourages greater engagement from participants (Colucci, 2007).

Since the data which resulted from the quantitative study indicated that a low level of awareness was expected from the participants of the focus group discussions, three informative video clips were prepared in advance and shown to the participants to stimulate the discussion. One dealt with an overview of the research being carried out at the Centre for Molecular Medicine and biobanking through filmed comments of experts in the field, as well as footage of the biobank and labs to provide a visual aid since the topic is rather abstract for most.[5] The second clip involved a brief interview with a lawyer who explained the impact of the General Data

[5] Video 1: https://www.youtube.com/watch?v=pJztP_ZQlwM&feature=emb_title

Protection Regulation (GDPR) on biobank participation.[6] Since the GDPR had only been put in place for a few months, there surely was a vacuum in knowledge, and therefore brief information was provided to ensure that the focus group participants give informed opinions on the topic of ethics and law in relation to genomics and biobanking. Moreover, for the third and final clip, a storytelling technique was adopted. This video clip included the narration of two genomic research participants who have their sample, or their child's sample, stored at the biobank.[7] The aim was to expose the focus group participants to the process of donation, as the video clip interviewees recounted what the process involved and commented on their experience as donors. This technique is mostly adequate when the moderator needs to understand attitudes towards a particular real-life situation (Colucci, 2007).

3.4.3 **Researcher's reflexivity**

The active involvement of the researcher's self in the process of research must be acknowledged (Eyles and Smith, 1988). Occupying a research support role as a social scientist at the Centre for Molecular Medicine and Biobanking (CMMB) whilst carrying out this research, impacted both ventures mutually. Through working at the CMMB, I managed to gather information about experts within the field, which in turn influenced my research choices. Case in point, as explained above, I decided to opt for interviews as opposed to focus groups based on the perceptions I had formed from encountering the cultural dynamics which exist amongst the research experts.

[6] Video 2: https://www.youtube.com/watch?v=kLyOH5yXrLU&feature=emb_logo
[7] Video 3: https://www.youtube.com/watch?v=g6ld8jFobHY&feature=emb_title

Furthermore, forming a part of the team at the CMMB, meant that focus group participants could perceive me as a part of the research institution in question, which could, in turn, lead to a Hawthorne effect. This led to the possibility for participants to feel uncomfortable to be forthright about their concerns towards the research process, as they "define the context for their behaviour and respond accordingly" (Adair, 1984).

Adopting mechanisms which immediately put the focus group participants at ease helped to alleviate this disadvantage. Attaining an "unobtrusive stance" in the research setting involved "the art of blending in" through "conscious decisions" (Silverman and Patterson, 2014). Dressing down in casual attire, sitting with the participants in the circle, kicking off the session with refreshment whilst dedicating some time to have informal conversations with the participants (Krueger, 2002), and making clear statements of emphasis about the confidentiality of all participants, all helped to limit the impact of my role at the CMMB on this research project. Indeed, I believe that this factor had little to no impact on the data collection process, as throughout the conversation all participants appeared to be comfortable about making challenging remarks.

In accordance with this, during the focus group session with the patients, I found myself thinking on the spot. As patients recounted their personal experiences of living with illness, and how they perceive medical research as a means of hope for breakthroughs, I found myself blending in by revealing aspects about myself to the participants. As I recounted my personal experience of living with an endocrine condition, I further reinforced the rapport which I had built with the participants. This is not something I had initially planned on doing, but I do believe that it positively impacted the data collection process. Nonetheless, I remained committed to keeping a barrier as a researcher and therefore, I restricted my input to the

discussion on attitudes and perspectives towards the issues related to participating in genomics and biobanking.

3.4.4 **Objective empathy**

Qualitative research often calls for the researcher to be mindful of one's emotional response, while still practising compassion. Empathy fosters connection and trust, two factors which no researcher should take for granted (Dickson-Swift et al, 2007). As participants narrated their experiences and expressed their thoughts, they gave a piece of themselves to the research, and as a researcher, I constantly reminded myself that the data subjects are individuals who may become vulnerable as they "pour their hearts out" for the sake of research (Etherington, 1996, p. 347). Therefore, showing empathy and caring for the research participants, is not merely a tool for collecting valuable data, but rather it is a researcher's trait of being human (Morse et al, 1990).

During the focus group sessions, I often encountered moments where I needed to remain objective, whilst genuinely feeling empathetic. Patients, and the parents of patients, in particular, expressed that the focus group session served as the perfect therapy session for them as they had the opportunity to open up about their troubles and worries with fellow patients who are practically encountering the same hurdles. Consequently, patient focus group participants[8] often went off-topic as they recounted their personal experiences. I found myself listening, feeling for them, and after giving them time to open up, I would subtly refocus the conversation. This might have lengthened the sessions, however, in the long term it allowed

[8] Includes both patients and parents of patients.

for generating rich, valuable data, which would not have been achieved had I tried to control the discussions in a manner which restricts participants from raising points which they needed to speak about. I felt that telling participants to avoid going off-topic with their remarks would have led them to feel inadequate and that would have directly impacted their participation. The fact that participants expressed such comfort informs about the level of rapport which was achieved. Allowing for a certain level of participant freedom, whilst expressing genuine empathy, contributed to maintain their sense of tranquillity within the group.

3.4.5 Qualitative data analysis: thematic analysis

The ontological and epistemological narratives of participants shall be the main tools for analysis (Stalker, 2009). Conversations about "storied experiences" stemming from group discussions and one-to-one interviews "obtain rich and free-ranging discourse" (Salkind, 2010, p.869). The meanings that the participants assign to their lived experiences, as well as the impact of their life histories on their attitudes and perceptions, provide a powerful rendition of human interpretation of the social world, vis-à-vis the social phenomenon in question, at a specific time (Stalker, 2009).

I would say that the qualitative data analysis started before data collection. Recruitment was in itself a finding which kept me thinking. Patients were excessively eager to participate, whereas experts seemed to be somewhat reluctant. These instances immediately sparked potential issues for analysis, and I would immediately grab my researcher diary and scribble notes about potential themes. During the data collection process, further analysis was made, and once again I ensured that immediately after the focus group sessions, and after each interview, I would transcribe my thoughts and point out the key arguments and themes which would have emerged from that session.

Nonetheless, the actual process of analysis kicked off after, whilst I spent ample time re-listening to the recordings, reading the transcripts, and marking the most relevant parts whilst taking notes. The combination of inductive and deductive thematic analysis involved a "hybrid approach" where "an a priori template" was used to "organise the data to begin with, but where novel themes [were] also allowed to emerge from the analysis" (Willig, 2013, p.60). As I listened and read, I started to identify patterns, contrasts, feelings, contradictions, and eventually constructed the key themes and subthemes which captivate patterns of meaning in the data. At times I actively constructed the themes for analysis as afterthoughts about the meanings behind the data and possible links between the data would emerge at unplanned times, and where I would immediately take out my researcher diary and jot down the potential theme and sub-theme ideas. This process was crucial for structuring the data and for building stories from the narrations of the participants (Willig, 2013).

Thematic analysis allows for "theoretical flexibility", in the sense that the researcher does adopt an epistemological or a theoretical framework, however, the choice is made at the discretion of the research, as the method in itself does not incline towards toward a particular framework (Willig, 2013, p.58). A theme essentially emerges as the researcher identifies "a pattern of meaning found in the data" (Joffe, 2012, p.209).

Focus group data were not simply analysed as if they are interview data. The aim was to observe the conversation dynamics, the ways in which participants reacted to each other, or influenced each other, and that is where the valuable data emerged from (Wilkinson, 1998). The data from the expert interviews were interpreted taking on board what emerged in the focus groups, and vice versa. The data from the two methods blended impeccably, shedding light on the themes which appeared to be relevant to analysing the phenomenon under investigation.

The key to attaining a valuable analysis was to identify both manifest and latent meanings present in the data, by observing beyond what the participants directly claimed, and looking for "implicit meanings" (Willig, 2013, p.59; Joffe, 2012). Sometimes participants would claim one thing about a specific issue, but as the discussion evolves and whilst discussing a separate issue, they would make claims which shed clearer light on the attitudes and perspective towards an issue or theme which would have been previously discussed. Being observant of such instances was key to analysing the data. One example from this research project is that of a participant who claimed that she would primarily participate for the benefit of future generations, because she does not believe that a breakthrough would happen within her lifetime, yet, at a later stage, when she was challenged by a fellow participant for having excessive trust towards authority, she claimed that it is not essentially a matter of trust but rather because she cannot wait to feel better. Through this idiosyncratic remark, during the discussion on trust, the participant shed light on the issue of motivation as she declared that some level of hope for personal benefit persists.

3.5 Ethical considerations

The process of the preliminary quantitative research task posed no major ethical concerns since no personal data was collected. All respondents gave verbal consent (see Appendix A) after being informed that their participation is completely voluntary and that they have the right to change their mind about participating at any point during the conversation, in which case all data would be immediately discarded. The information provided by the respondents is completely anonymous since identity details were not required for this research. Verbal consent was the ideal consenting procedure here since participants can in no way be identified through the responses they provided, and therefore, it was the preferred option not to keep signed

consent forms of such participants, for data minimisation purposes. Post participating, all respondents were provided with an information sheet about the research project, including the researcher's contact details (see Appendix C).

During the process of gathering the qualitative data, all participants were carefully informed about the purpose of this research and were provided with ample information about their rights as participants. They were all informed that their participation is voluntary and that they reserve the right to withdraw participation at any time. Pseudonyms are used throughout this dissertation to ensure the confidentiality and anonymity of participants. Participants were asked to sign a consent form after thoroughly discussing all points with each participant, which consent form included all the necessary information about this research (see Appendices E and I). The Faculty Research Ethics Committee (FREC) granted approval for this research and its associated documents, such as the consent forms.

Focus group participants were reminded of the importance of maintaining confidentiality about what was said within the group, once the focus group discussion was over. Since the research community at the Centre for Molecular Medicine and Biobanking is small, extra care was taken to safeguard the identities of the research experts who were interviewed for this dissertation. Hence, no gender pronouns will be used whenever referring to the research experts to disassociate them from their respective genders, with the aim of adding a layer of protection to ensure anonymity and confidentiality. To further protect the experts' identities, researchers and the biobank manager will be referred to as Research Expert 1, 2, 3 and 4, in no particular order, to avoid the identification of the biobank manager, since in Malta the role is occupied by one individual only. Moreover, the support group representatives will be referred to as Support Group Representative 1 and Support Group Representative 2.

The intention was to audio record all focus groups and interviews, however, since one expert interviewee declared the preference of not being recorded, the audio recorder was not used for that particular interview, and the participant provided responses in a written format. Follow-ups were made via email. This did not completely restrict the data collection process since the written communication was smooth as the interviewee promptly replied to all follow-up questions. It was ensured that no deception takes place and thus the participants' desires were given priority. All the other interviewees and focus group participants were comfortable with being audio recorded. The recordings were downloaded onto a password-protected computer and the original recordings were immediately deleted from the recorder. All transcriptions are also being stored on a password-protected computer and the actual names of the participants do not appear on these transcripts.

3.6 Research barriers and challenges

The main obstacle of this research was to get the experts to participate. The lack of eagerness to participate by most research experts lengthened the duration of the data collection phase, but ultimately it did not limit this research, as the desired number of research experts to be interviewed was reached.

The lack of awareness and knowledge of the focus group participants on the topic in question was also a hurdle, which was tackled through the production of informative video clips which highlight the key medical, social, ethical and legal aspects of genomic research and biobanking, as explained earlier.

4. Quantitative Data Analysis: biobank awareness amongst the local population

Literature exposes a very low level of awareness amongst pan European citizens. Gaskell and Gottweis (2011) even warned that if biobanks are not better promoted, they could be at risk of failure, since the success of a biobank is utterly dependent on individuals who are aware and willing to actively participate in research. Thus, data about the levels of public awareness in the local context is indispensable to the success of the biobank at the University of Malta.

4.1 The results: too many know too little

The initial question posed to research participants (n=387) asked whether they understand what a biobank is or not.[9] It appears that a striking 87.1% (n=337) of the sample claimed that they do not know what a biobank is, whereas another 5.1% (n=21) stated that they were not sure. A mere 7.5% (n=29) of the sample (n=387) claimed that they know what a biobank is.

[9] Q1: Do you know what a biobank is?

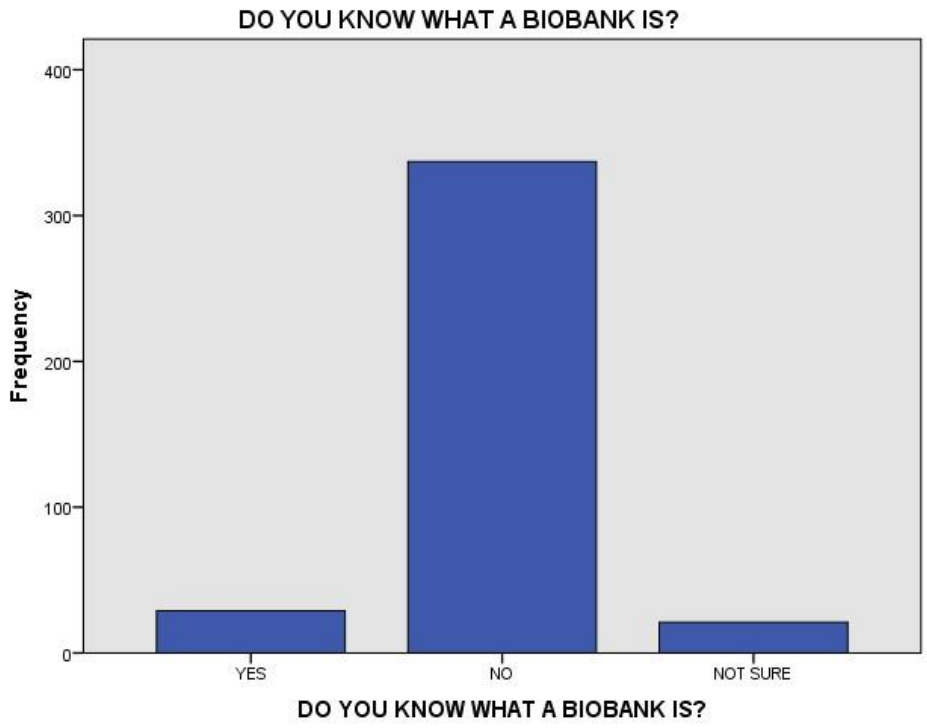

Figure 1: Responses to Q1 (n=387) (Desira and Martin, 2018)

The data which emerged beyond this face value statistic is even more surprising. It was revealed from the responses to the follow-up question[10] that a significant proportion of those who believed that they knew what a biobank is (n=29) was actually unaware of the function of biobanks as repositories of biosamples to be used for medical research. Twelve of these participants gave a legitimate definition of a biobank but failed to mention biobanks which operate for the purpose of biomedical research, and instead named repositories which store embryos or gametes (7 mentions), human organs for donation (4 mentions) and stem cells for future personal use (1 mention). Another 8 participants made speculations which are completely unrelated to biobanking and mentioned financial banks, bitcoin and internet banking amongst other guesses. It was only 9 of the 29 participants who had claimed to know

[10] Q2: What is a biobank?

what a biobank is that went on to mention biomedical research. This statistic is a key outcome which emerges from this study, as it can be concluded that a bare minimum of 2.3% of the population is aware that the term biobank refers to a biorepository which operates for the purpose of biomedical research.

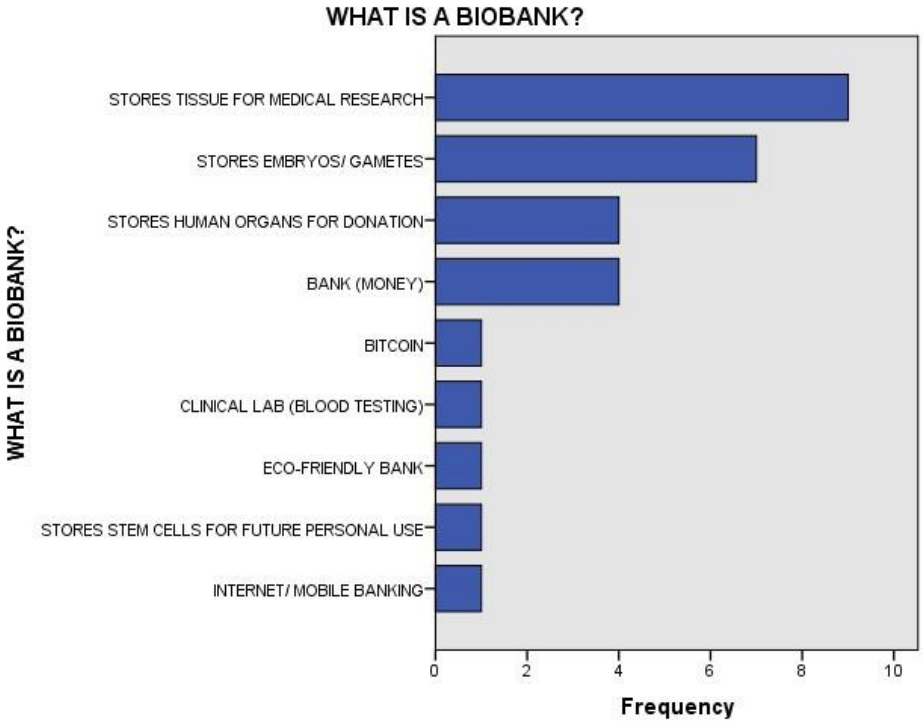

Figure 2: Responses to Q3 (n=29) (Desira and Martin, 2018)

In contrast, when looking at the responses of participants who initially claimed that they did not know, or were not sure, what the term 'biobank' refers to (n=358), it is observed that a portion went on to give a legitimate answer when asked to speculate further about what they think a biobank might be.[11] Specifically, 91 respondents (25.4%) linked the term biobank to a store of biological material. Out of the ninety-one participants, a notable 2.8% (11 participants)

[11] Q4: What do you think a biobank is?

mentioned biobanks which store samples for the purpose of biomedical research, despite initially claiming that they did not know what a biobank is. The remaining 80 participants suggested the following guesses: storage of embryos or gametes for fertility treatment (9.2%), blood banks for blood donation (6.7%), storage of organs for donation (3.9%), storage of stem cells for future self (1.7%), and storage of seeds to be used in case of a catastrophe or extinction (1.1%).

Amongst those who claimed not to know a biobank is when asked the initial question, the suffix 'bank' seems to have had a significant influence as 40% (n=143) went on to give an answer which relates to finance: 126 respondents mentioned 'bank as a financial establishment', 12 participants mentioned 'eco-friendly bank', whereas 5 participants mentioned 'mobile banking'.

When invited to speculate on the meaning of 'biobank' another portion attributed the prefix 'bio' to something 'eco-friendly' (12 responses) or 'organic' (12 responses). Fifteen participants did assume that a biobank is a biological bank but could not presume what the purpose of such a bank could be. Another 3.1% assumed that the term is linked to health but could not specify what it means, whereas 2.6% of the research participants who were invited to speculate about the term 'biobank' linked it to science mentioning different types of scientific labs such as pharmaceutical, clinical and animal testing labs. Some of the most imaginative guesses include 'power bank charger' (4 responses), 'bitcoin' (3 responses) and 'beverage brand' (1 response).

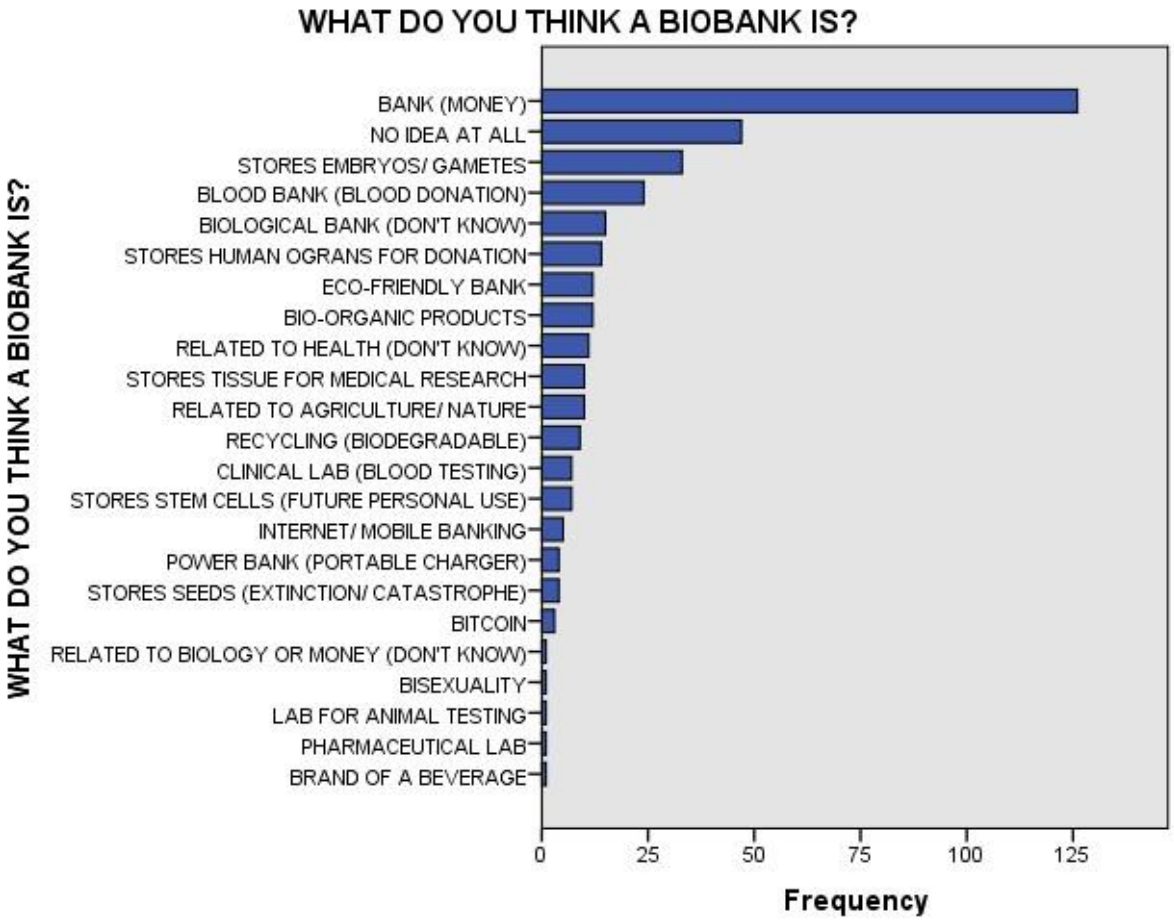

Figure 3: Responses to Q4 (n=358) (Desira and Martin, 2018)

4.2 The few who knew more

An absolute minority of 2.3%, or 9 participants, claimed to know what a biobank is and went on to give an accurate definition which mentions medical research. The absolute majority of these participants (77.7%) are individuals who have completed tertiary education. Thus, it can be said that within the sample there seems to be a relationship between awareness and level of education.

As previously stated, the vast majority of the sample claimed not to know what a biobank is when asked the initial question, however, when invited to speculate on the meaning of

'biobank', 10 of these respondents went on to mention medical research. Therefore, a total of 19 respondents, or 4.9% of the sample, were aware of the process of storing biospecimens for biomedical research, although not all were aware that the premises which serve this purpose are called 'biobanks'. There also is a clear indication that educational achievement is a relevant demographic amongst this group as 100% of these 10 respondents have completed post-secondary or tertiary education.

The 19 participants, who at some point mentioned biobanks which store biospecimens for biomedical research, were also asked about the source through which they learned about the function of biobanks.[12]

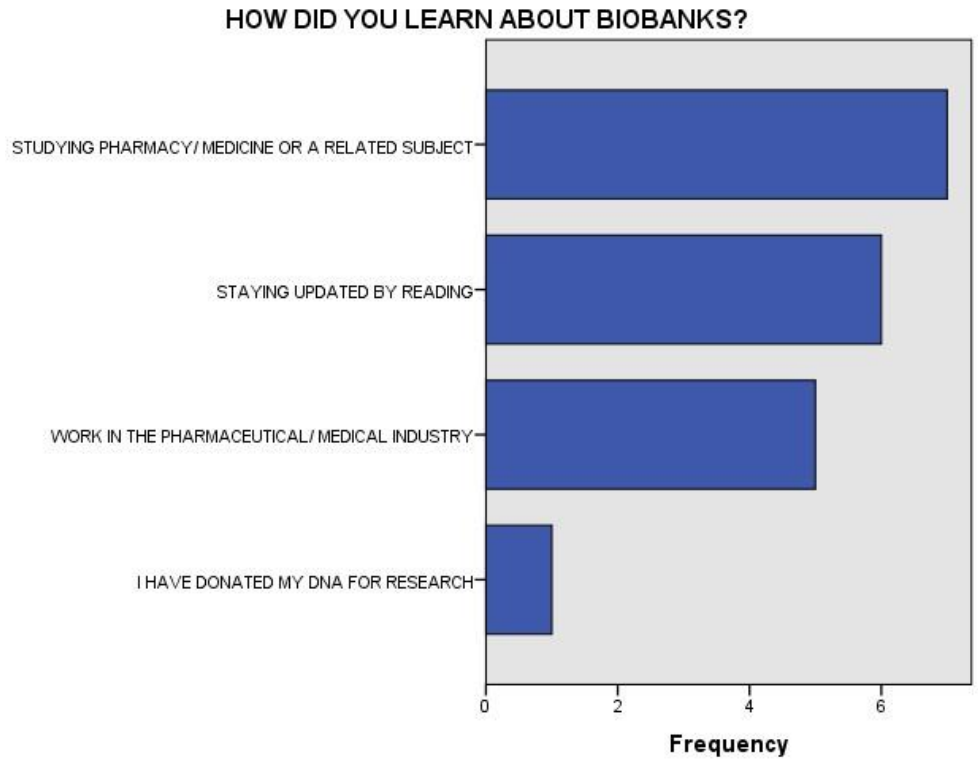

Figure 4: Responses to Q3 (n=19) (Desira and Martin, 2018)

[12] Q3: How did you learn about biobanks?

Seven of the 19 respondents were individuals who are currently studying a subject related to health, medicine or pharmacy, whereas another 5 respondents work in a related industry. Six respondents learnt about biobanks for medical research through independent reading, and one respondent got to be aware after donating biospecimens to the biobank for research. This implies that none of the respondents got informed about biobanks through public outreach campaigns.

4.3 Gender and educational background as predictor variables

The data collected for this dissertation revealed significant correlations between levels of awareness and educational background. As for gender a correlation also exists, however, it is less striking.

Looking at the data of the participants who gave valid answers, it appears that individuals with a primary or secondary level of education who gave a legitimate answer mostly mentioned ‘banks for embryo or gamete freezing’ and ‘blood banks for blood transfusion’, whereas those who completed post-secondary or tertiary education were more likely to mention the other types of biobanks, with a significant positive correlation between the attainment of a tertiary level of education and mentioning biobanks for biomedical research.

When analysing data in relation to gender, it can be observed that there is a marginal correlation between the male gender and those who gave a legitimate answer, with 59.3% of such participants being male and 40.7% being female.

4.4 Discussion: awareness leads to donation and donation allows for research.

With popular responses ranging from a financial institution to stores of blood for transfusion and, with less popular guesses ranging from a brand of a beverage to bitcoin or a power bank, it is safe to say that the level of public awareness of biobanking for genomic research in Malta is remarkably minimal. The most common response amongst participants, after 'financial institution', was related to IVF, specifically to gamete or embryo freezing. This can be attributed to the fact that data collection occurred at a period when the country was undergoing an intense debate on the topic, both in parliament and on the media.

The significantly low count in mentions of biobanks for medical research indicates that the Maltese general public is excessively deprived of information about genomic research and biobanking. When one considers that biobanking has originated over a 100 years ago and that the biobank at the University of Malta was established in the year 1989, such data are rather shocking, especially since biobanks depend entirely on human participation. The need for information aimed at the general public is evident. The publication of the statistics which resulted through this research shed light on the matter for a strategy to be implemented accordingly.

The data which emerged through this preliminary research was crucial for providing contextual information for the next phase of this research project, i.e. the qualitative part.

5. Qualitative Data Analysis: attitudes towards risks and benefits

The qualitative data which emerged through the focus group sessions and in-depth interviews explore the attitudes of patients, the parents of patients, members of the general public and experts towards the key issues related to participating in genomic research and biobanking. Thematic analysis was used to establish the following three overarching themes; a) motivating factors, b) risk perceptions and levels of trust and c) attitudes towards power and control.

5.1 Focus group participants' vignettes: brief outline of lived experiences and key opinions

Focus group session one: patients

Jessica suffers from endometriosis and often feels frustrated that not enough money is being invested in research on her condition. She was not aware of the biobank at the University of Malta and did not know of the research being conducted at the Centre for Molecular Medicine and Biobanking (CMMB). Jessica speaks of a sense of mistrust towards the medical system ever since she was misdiagnosed for approximately ten years and experienced severe repercussions.

Michaela recounts that her mother passed away after battling cancer and that later on she too underwent treatment for cancer. She is well aware of genomic research and biobanking as she has donated her breast tissue for research after undergoing a double mastectomy. Michaela expresses disappointment at being asked to sign the consent form right before entering the operating theatre, claiming that at such a sensitive moment one would not have the courage to

go against medical authorities. Nonetheless, she claims that donating the sample was the right decision since she believes that the research community at large has good intentions.

Elisabeth is diagnosed with a rare disease and has learned about the University of Malta biobank and research at the CMMB through the National Alliance for Rare Diseases. She expresses eagerness to participate in research and feels disappointed that local researchers are not looking into her condition. Elisabeth has donated her sample to a research organisation in Italy which specialises in her condition.

Mariella, who is diagnosed with a rare disease, only had a vague idea of biobanking and genomic research prior to the focus group session. She is one of the most sceptical participants of this research, being particularly concerned with privacy risk and commercialisation.

Focus group session two: parents of patients

Therese's daughter has a rare disease, and both mother and child have donated a sample to the University of Malta biobank. Therese insists that despite any risks associated with the donation of biospecimens for research, the benefits dominate. She raises one major concern about researchers who refuse to share data, claiming that such researchers are motivated by egotism as opposed to altruism.

Frank is the father of a child with a very rare condition, and his only concern regarding participation relates to corporeal risks, such as physical pain. Frank was not aware of the biobank at the University before the focus group session. This father expressed frustration at the fact his son's DNA sample is not being stored at the biobank; considering the rarity of the

condition, Frank believes that the sample should be collected and stored, even if it will not be used for the time being.

Josianne's son has a rare disease, and although they have not been invited to donate samples, they have recently learnt about the CMMB and the biobank through the National Alliance for Rare Diseases. She repeatedly asked whether she would be able to donate a sample, which implies extreme eagerness to participate in genomic research, even though she claimed that she does not believe that a breakthrough will be achieved within her son's lifetime.

Stefania's son has an intellectual disability. She was not aware of the biobank and medical research being carried out at the University of Malta. She recognises that the treatments and therapies which her son receives resulted from past research and therefore, she feels obliged to participate in research for the benefit of future generations.

Dorothy is the mother of two adult sons who are diagnosed with a rare blood condition. She also was unaware of the biobank and the CMMB at the University. She feels that she lacks information and expressed the desire to be better informed about ongoing research, claiming that she would be eager to donate her sample to the research community.

Tina also never participated in genomic research. She lacked awareness of the biobank and the subsequent research being carried out. Tina is adamant about not wanting to receive information about incidental findings, although she believes that participants should have the right to decide. She was not immediately concerned about the risks, but later on during the focus group session she felt rather anxious about the possibility of a data breach, claiming that she is rather secretive about the condition of her daughter.

Focus group session three: the general public[13]

All participants in this group, except for Marica, were unaware of the biobank and medical research being carried out at the University of Malta.

Marica learnt about biobanking and genomic research through her studies, since she is a healthcare worker. She expressed little to no concern about risks, claiming that she would not be concerned if her genetic information was leaked. She, however, had a change of heart later on during the session after another participant spoke about the possible repercussions of a data breach.

Claire did not seem to have major concerns towards the start of the session, however, she later on independently thought of the possibility of data breaches to employers and insurance companies which can in turn lead to genetic discrimination. Towards the end of the session, she became more anxious about donating genetic data.

Robert started to lose trust in medical experts after they were ineffective in treating his recurring migraine, whereas alternative medicine managed to heal him. He claims that the medical system is controlled by Big Pharma and believes that the industry does not have the best intentions as it is driven by profitability. Nonetheless, he understands that Big Pharma requires funds to produce novel medications and thus argues that it is inevitable for the industry to make business out of illness.

Luke trusts the medical system, and is not concerned about risks, stating that he does not feel that genetic data is sensitive data. He is one of the few participants who would not expect

[13] All participants in this group have completed tertiary education.

information about any incidental findings, arguing that research participation should be selfless.

Adriana believes that patients are helpless beings who must trust medical professionals because they have no other option. She recognises the risk of participation and believes that data leaks are especially common in close-knit communities such as the Maltese community. Nonetheless, she believes that donating biospecimens for research is worth the risk, as the benefits supersede the possible dangers.

Focus group session four: The general public[14]

All participants in this group, except for Peter, were unaware of the biobank and genomic research being carried out at the University of Malta.

Mario believes that rigorous regulations and ethical standards are preventing genomic research from reaching its full potential. He is concerned about commercialisation and is a strong believer in the conspiracy theory that the effort to develop treatments is higher than the effort to develop cures, because treatment to chronic illness creates life-long customers for Big Pharma, whereas a cure would generate less profit.

Lina is sceptical of the trustworthiness of the medical system as a whole. She recognises that risk exists and believes that a breach of data is a likely possibility, yet, she is well motivated to donate her biospecimens for the greater good.

[14] All participants in this group have completed primary, secondary or post-secondary education.

Jesmond expressed a high level of trust towards the system, including the pharmaceutical industry. He recognises the implications that can emerge from human error but believes that the vast majority of medical professionals aim towards the best outcomes for the common good.

Mark is highly motivated to participate as he believes that genomic research would be beneficial for future generations. However, he expresses scepticism towards commercial entities, claiming that they are solely motivated by profit and do not share the values of the rest of the research community.

Grace expressed eagerness to participate in genomic research, claiming that she would love to be a part of the solution to combat illnesses. Moreover, she sometimes fears innovation arguing that what we perceive as progress may unknowingly be causing us harm.

Peter recounts a personal experience of how an incidental finding affected his mental wellbeing. He learnt about biobanking from his son who works in a related field, and has also donated his sample for research a few years back. He expressed average motivation and little concern about risks.

5.2 Motivating factors

5.2.1 Beyond motivation: enthusiasm

The data collected through the focus group sessions indicate high levels of motivation amongst all participants, however, there seems to be a clear discrepancy in levels of enthusiasm amongst patient groups and non-patient groups. Participants from the patients' focus groups[15] not only

[15] Includes both patients and parents of patients.

expressed the motivation to participate but also expressed high levels of eagerness and enthusiasm.

> "It would be a good idea if samples were taken immediately after diagnosis. Even for conditions which are not being researched as yet." – **Therese**

> "I would say that samples should be taken from all new-born babies. That is the easiest way to collect as many samples as possible." – **Frank**

Throughout the discussions, patient focus group participants repeatedly asked for more information about how to participate. Despite clarifying that the focus group session was aimed at gathering data about their attitudes and perspectives, and not to collect their samples, they ensured to emphasise their interest in participating at the end of the session.

> "Can the people at the biobank make contact with us? Are we on their list?" – **Dorothy**

> "I would really appreciate it if they called me so I'd come back to give my sample." – **Josianne**

This tallies with the literature which shows that biobanking and genomic research instil a deep sense of hope in those battling illnesses or conditions which are incurable or untreatable (Boat and Field, 2011). Moreover, even though most of the participants in the patients' focus groups were aware that their condition, or that of their child, was not being researched at the time of the focus group, the idea of donating their biospecimens still seemed to inspire a sense of hope.

5.2.2 **Improved healthcare. But when?**

The most common motivation for participation was that of providing better healthcare, mainly for future generations. Most genomic research involves a longitudinal process, and that is why

biobanks are indispensable (Laurie et al, 2010). It is clear that the vast majority of participants recognise that expecting overnight results is unreasonable.

> "We have to understand that the treatments that we give to our children today are the result of past research. It is therefore our obligation as parents to donate our samples and those of our children for research and to help future children who might have the same condition. We all know how much suffering we have to endure… knowing that I can help individuals who will go through this in the future motivates me, even if I am certain that we won't benefit ourselves. Future patients could be our own grandchildren after all." – **Stefania**

> "I am certain that research won't benefit my son, neither today nor tomorrow, and not in his lifetime, yet I still want to participate. I want to help those who are still to be born with the condition." – **Josianne**

The majority of patient participants were in agreement on this issue, however, both patients' groups had individual participants who would beg to differ. Frank, who is the parent of a patient, argued that he is confident that as for illnesses which are being researched, breakthroughs are more imminent than one might presume.

> **Frank**: "I cannot say I completely agree with you. You are wrong to say that only future generations will benefit. Why can't we benefit too?
>
> **Josianne**: "I wish that was possible of course, I would want my son to benefit first and foremost…"
>
> **Frank**: "Technological and scientific progress is happening at a very fast pace. I don't just dream, but I believe that we will get there in the very near future. You're exaggerating the number of years it takes for novel discoveries."
>
> **Josianne**: "I think of the worst-case scenario so I do not get disappointed."
>
> **Maria (author)**: "Frank, but what if you were told that it is certain that for a breakthrough to be found it would take longer than your son's lifetime?"
>
> **Frank**: I would still participate because I am sure that it would not take so long. I am certain, as I know that conclusions have derived even from illegal

experimentation, and sometimes they are leaked to the media, and then they try to cover it up."

As indicated by the European Organisation for Rare Diseases, two of the main concerns for individuals who struggle with rare diseases are a lack of the knowledge on diseases and a lack of treatment options, thus, it is no surprise that the possibility for research inspires a sense of hope. These findings tally with research by Ducournau and Strand (2009) which reveals that in the case of patients and their family members, the hope for a breakthrough which would potentially improve their health, or that of their children, or even their descendants, remains a key motivator to participating in research.

Even though the majority of the patient-participants[16] of this research claimed to be motivated to participate for the good of future generations, their level of enthusiasm, when compared to the non-patient groups, indicates that the possibility of self-benefit is certainly a motivator. Stefania indicated this clearly when she said:

"Future patients could be our own grandchildren after all." - **Stefania**

Whilst discussing the issue of commercialisation and the motivation of the pharmaceutical industry to develop new treatment and cures for the sake of profit, Michaela expressed how she experienced first-hand the advancements brought about by research, claiming that she is convinced that in the long term the donation of a sample can truly impact lives. Thus, her personal experience of medical progress was the key motivation behind her decision to donate her breast tissue for genomic research, after going through a double mastectomy.

[16] Includes both patients and parents of patients.

> "I have witnessed the development of cancer treatment. My mum was diagnosed 18 years ago – and we truly saw a difference from 18 years ago, to 11 years ago when she died. Every year the treatment improved. Had there not been any developments, as a patient she would have suffered as well… it not just about the business. […] I know that my sample donation can make a difference." – **Michaela**

Some research expert interviewees share a similar motivating factor.

> "I do not like the attitude of people who think that our research will not help anyone. […] I am not interested in doing research which will help no one." – **Research Expert 3**

Despite recognising the limits of research, they nonetheless claim that their drive stems from a desire, an ultimate goal, of changing lives for the better, and preferably witnessing that improvement. Whilst non-patients are the stakeholders who expressed the least eagerness to receive feedback about the outcomes of research, patients see donation beyond a one-time event, and therefore seek to remain involved.

Nowadays, researchers are moving towards the idea of considering research participants as part of the research community, often referring to them as research partners as opposed to mere participants. The development of innovative tools, such as dynamic consent portals, encourage the active involvement of participants, by giving them greater control, and providing them with research feedback (Mamo et al, 2019).

My data show that patients are evidently more interested in frequent feedback, as opposed to non-patients, which is another finding which supports the argument that patients have added motivation which results from a deep desire to receive news about breakthroughs, even within their lifetime. Non-patients also participate in research with the hopes of positive outcomes, however, they are less interested to learn about what emerges from research, and perceive

participation as a one-time encounter, rather than a process of active participation. Claire, a non-patient, outlines this finding through the following statement:

> "If it becomes a frequent thing, I'd end up not even opening them, because I would consider them spam." – **Claire**

5.2.3 **Altruism versus personal gain**

Altruism plays a key role in motivation amongst all groups and all participants. At the start of the focus group discussions, all twenty-one participants claimed to be motivated towards participating in genomic research and biobanking, and towards the end of the discussion, the same question was posed to all the participants, and without exception and irrespective of any concerns they might have developed, they all claimed that they would still be willing to participate. The most common reason for motivation was the possibility of generating benefits for the greater good.

> "Even if it means saving the life of just one person, I would do it. I don't care about anything else." – **Michaela**

The altruism of patient-participants is conveyed through their enthusiasm to participate in research which would most likely only be fruitful beyond their lifetime (Ducournau and Strand, 2009). The issue of time is interlinked with solidarity, especially in the case of patients who participate with a sense of urgency, as they seek for solutions to their respective illnesses. Although patient participants seem to be well aware that the development of new cures and treatments takes time (Laurie et al, 2010), their response to this limitation is typically positive, as they often responded with a sense of solidarity.

In 2016, Maltese philanthropist and ALS patient Bjorn Formosa was awarded the European Citizenship Award for 'Best Volunteer' for his "pure expression of altruism" through actively

campaigning to collect funds for motor-neuron disease research. His altruism was described as "absolutely remarkable" for the reason that "[he is] statistically almost certain that a cure will not be found in his lifespan", yet he has dedicated his remaining time and energy to provide a better situation for those who shall be diagnosed in the future (European Civic Forum, 2016). The fact that participants from the patients' groups expressed eagerness to participate in genomic research, even if they believed that they would not get to make any personal gain, shows that ultimately patients too are motivated by altruistic elements.

Potential illness is a preoccupation for healthy individuals as well. Whether the future self will be healthy or not is uncertain as no person is immune to illness, and the possibility of going from healthy to ill overnight persists. However, the motivation level of those who perceive illness as simply a future possibility is not at par with the motivation of those currently experiencing illness. The majority of the participants from the general public focus groups were convinced that they would participate, however, it could be noted that the enthusiasm that they communicated did not correspond to that expressed by the participants in the patients' groups. Towards the beginning of the session, one non-patient participant claimed that:

> "I can only think of benefits. It is probably more for the future generations rather than myself, but there is nothing to lose so I would surely participate."
> - **Robert**

The motivation is there, yet the enthusiasm is not as high as that of the patient groups. Robert later reinforced this argument when he claimed that his only concern is that of finding the time to do the donation:

> "I donate blood regularly, can't they take the sample on the same day. I am willing to participate in research but it is usually not easy to find two hours in my tight schedule to fit this." – **Robert**

It is interesting to note that whilst blood donation is a regular errand for Robert, finding time for a one-time appointment for donation to the biobank seems to be more of a concern. The issue of time came up in both non-patient group discussions. Blood donation involves an immediate sense of gratification, individuals are invited to donate blood to save the life of a patient there and then. The deferred gratification of donation for research could in itself manifest as a demotivating factor for potential participants. Talcott Parsons (1951) speaks of social actors as beings who are motivated by the "optimisation of gratification". When it comes to genomics, immediate gratification is generally not possible and thus social actors, or potential participants, can only be motivated by deferred gratification because it is typically the only option. Nonetheless, it seems that deferred gratification has an impact on motivation levels and enthusiasm.

One of the non-patient focus group participants attributed the lack of awareness about the biobank to deferred gratification, or quite frankly to no gratification at all. Marica's interpretation was that since the biobank cannot make an immediate impact on the lives of individuals, outreach is more difficult as the outcome which emerges from the action of donation is not as motivating as it is for other forms of donation such as blood donation for transfusion. Even if in both cases there are zero personal benefits, the intensity of satisfaction from the altruistic act is greater in the latter case, since the donor knows that an impact has been made there and then.

> **Adriana**: "When did the biobank is Malta start operating?"
>
> **Maria**: "In the 1980s."
>
> *All participants make gestures which indicate that they are stunned.*
>
> **Marica**: "Because participating does not give you a direct benefit. I think that is why people do not know about it".

Video clip which features an interview with a patient-donor and the mother of a patient-donor.

Marica: "Those who are ill, or their families, will obviously participate. But how do you get a sample from someone who is old, or not interested? It is an extra hassle from which they would not benefit. Ok, you can say that other people will be benefit – but when will they benefit? And can you guarantee it? I believe it is a complex concept which is not easily understood by those who are not directly affected by some form of illness".

Luke: "Can't the biobank pay individuals to motivate them?"

Robert: "As if! People would still donate voluntarily. I'm sure you wouldn't need to pay them to donate."

Marica: "But not for science! They would donate if they can help someone there and then and save a life, but not for something so slow and abstract."

Adriana: "For those who are sick there surely is a greater motivator, which is hope for a cure."

Robert: "I disagree. To me, seeing that mother speaking[17] about how she truly believes that research can help her child is more than enough for me to donate a sample. I don't need to hear anything else!"

The disagreement amongst members of this group indicates that whilst some may perceive the concept of genomics and biobanking as abstract and therefore hard to be conveyed to the general public, others quickly changed this perspective once shown visuals of potential beneficiaries. It is thought-provoking that just a little bit earlier Robert had commented that he is concerned about finding time to donate, however, after putting a face to the situation by showing the video clip of a mother of a patient who is participating in genomic research, Robert's motivation to participate spiked and he was immediately convinced that he would

[17] Referring to one of the video clips shown at the beginning of the session.

donate his sample no matter what. This pattern was also observed in other participants from the non-patient groups, but not amongst the patient groups.

The discrepancy in motivation between groups can be interpreted through Maslow's (1943) theory of human motivation. The concept of biobank participation for genomic research is interpreted by patients as a means to satisfy physiological needs or the need for safety, whereas non-patients perceive the same concept as a means of satisfying the need for self-actualisation. Even though both patients and non-patients expressed motivation to participate in genomic research and biobanking, there is a clear disparity in the levels of motivation. Whereas non-patients are motivated by esteem needs, which they acquire from being moral and altruistic, alternatively, patients are also motivated by more primitive needs, specifically a physiological need and the need to feel safe and secure. Almost all participants in the patients' groups expressed a desire to help future generations, hence their sense of altruism is clear, yet it can be concluded that such individuals are further motivated by a more basic need, which is the physiological need of personal health, or the health of their dependents, and thus, according to Maslow (1943) the first and most basic need of all human beings. The instinct of motivation which originates from the need for physical wellbeing and security of the body was underlined by Elisabeth when challenged by a fellow participant about trust towards medical professionals:

> **Mariella**: "You trust authority figures too much."
>
> **Elisabeth**: "It's not that only... I cannot wait to feel better. I want to do anything it takes, everything, to make it at least 1% better."

Although Elisabeth had declared that her motivation lies in altruism for the benefit of future generations, when asked about a separate issue, that of trust, she made a claim which indicates that ultimately a sense of hope for personal benefit persists.

5.2.4 **Actions speak louder than words**

The superior level of enthusiasm of patient participants of this research when compared to non-patient participants, could be identified even prior to data collection. The enthusiasm expressed by patients and the parents of patients during the recruitment process was a revelation in itself. It appeared also from the focus group discussions that these individuals are constantly seeking information; they seek for solutions, and they feel the need to learn and do more. Those battling rare diseases often feel helpless (Boat & Field, 2011), and this could immediately be sensed from their excessive eagerness to participate in this research as they displayed their impulse to grasp on to anything which might be available.

> "I enjoy searching for information because I enjoy searching for hope." – **Dorothy**

Whilst patient groups expressed an intense desire to be contacted to participate in genomic, non-patients were much more likely to mention issues of inconvenience such as finding the time to visit the biobank or place of donation, and they were also quicker to mention risks which concern them and to open up on such a discussion. All of this implies that although at face value all 21 focus group participants claimed to be motivated and interested to participate, their actions and their subsequent contributions to the discussion, as they discussed attitudes towards specific issues related to genomic research and biobanking, imply a clear disparity in enthusiasm and thus in levels of motivation.

Moreover, the fact that most of the participants in the patients' groups had not been asked to give a sample, made them realise that there are no local research projects aimed at the conditions or diseases which concerns them, and this in itself further decreased their hope and instilled a sense of frustration.

> "I know that research is being carried out, but not on my son's disease. He has a rare disease and I was never asked to give a sample. That must mean that nothing is being done to find a cure or treatment for his disease." - **Josianne**

> "It is heartbreaking. I knew very little about my condition and I needed to gather more information so I would be able to lead a better life. However, knowing that my disorder was not being researched locally meant that I would not be able to move forward. Then I learned about a hospital in Italy which specialises in my condition. The knowledge which has emerged from their research has truly improved my life. I went from spending 70% of my time in the hospital, to spending 30% of my time in hospital. I would do anything to help those who will be diagnosed in the future… I don't want them to go through what I am going through. However, it hurts that nothing is being done about it in Malta. I am ready to do anything to help out the researchers, yet they are not doing anything about my disorder. Is my life not as valuable as that of persons with diabetes or whatever?" – **Elisabeth**

5.2.5 A sense of belonging

Amongst participants from the patients' groups also emerged the concept of community. Participating in research meant they formed part of a greater community which can possibly provide a sense of belonging. Those experiencing rare diseases tend to feel isolated, and being a part of the research community through the donation of a sample seems to be an aspiration for most patients and parents of patients.

> "As humans, we have the need to relate to others, and not feel like we are alone in this." – **Tina**

> "After giving my sample to the researchers in Italy, I often received feedback about the progress of their research, and they often publish about their findings. They have also provided me with information about the websites that I should use – websites which have reliable information about my condition which results from research from America, Turkey, Italy and from all over the world. At least I no longer feel alone." - **Elisabeth**

The concept of dynamic consent (Kaye et al, 2014) comes into play here. It is evident that participants, particularly patient-participants, are not satisfied with merely donating a sample, but rather they expect communication, they expect to be considered a part of the research community. Platforms which allow for communication amongst the key stakeholders, whilst providing participants with continuous information about the progress of research, are key to harvesting a sense of community which in the process nurtures a sense of trust within research participants (Laurie et al, 2010).

5.3 Risk perceptions and levels of trust

> "*It-tobba jsalvawk u huma joqtluk*." [Doctors are the ones who can save you, but also the ones who can get you killed.] - **Claire**

As determined in the previous section, the majority of participants from the patients' focus groups seemed to be more passive in discussing the risks associated with research and declared high levels of trust towards the medical system in general, including the medical research hub. This excludes one patient-participant, Mariella, who is one of the participants who conveyed deep concern, and who probably was the most sceptical participant from all the groups.

As for the non-patient groups, despite consistently expressing willingness to participate, several participants were immediately explicit about their scepticism towards the medical

system as a whole and expressed serious doubts about the intentions of Big Pharma. This particularly stood out amongst the second non-patient group.[18]

The distinction between the two group types (patients and non-patients) can be interpreted as a form of 'stratified reflexivity' (Ward and Coates, 2006). The data reveals that the interpretation of the information which is available depends on the lived experience of each individual. Patient-participants of this research were consistently prioritising and making trade-offs; at instances where the discussion on risk escalated to real-life scenarios of possible negative repercussions, all patient-participants, except for one, chose to dismiss such information and constantly sought to refocus the discussion towards the favourable outcomes of biomedical research. In contrast, non-patients were more comfortable with elaborating on such discussion. It appears that reflexivity and the interpretation of information are not merely influenced by socio-economic factors, as claimed by Ward and Coates (2006), but are also reliant on various other variables, depending on the topic in question. As for the donation of biospecimens for genomic research and biobanking, there is a clear stratification in attitudes based on health status, and this can be attributed to the distinctive way in which the two groups perceive the outcomes of research and biobanking; for one group the outcome is about improved health for the good of the community, whereas for the other group the hope for an outcome is more intimate and personal. This finding chimes with the literature which reveals that participants who are also patients are more likely to perceive researchers as trustworthy,

[18] All participants in this group have completed primary, secondary or post-secondary education, but none have completed tertiary education.

which in turn results in a higher probability of giving full consent for the use of their genetic data and biosamples (Robinson et al, 2013).

Nonetheless, the patient groups still engaged in an interesting discussion regarding possible risks, however, they consistently appeared to be less concerned than their non-patient counterparts. The burdens which instigated fear for the parents of patients, and the patients themselves, were more concrete and imminent, and were mostly related to the physical aspect of donating biospecimens. The initial concern that the parents of patients raised was associated with the fear of physical pain for their children, other than that, parents did not seem to be concerned about any other risks, particularly towards the beginning of the session.

> "Is it a blood sample that they would need? Or it is something else? His behaviour is what worries me because as soon as he feels the needle he gets really angry. Saliva would be much easier. My only worry is that he would not stay [still]… other than that there's nothing I would be concerned about." – **Frank**

This pattern was also flagged in data from interviews with researchers, particularly when Research Expert 1 confirmed that patients are typically more concerned with physical discomfort or situations of inconvenience, rather than other long-term issues of risk.

> "Patients typically make the decision of whether to participate or not based on the amount of discomfort… that is what they consider first… sometimes they ask about the number of times they would have to attend. […] I don't think I ever encountered a patient who did not participate because they did not trust us or because they were concerned about what might happen with the sample… no, never." **– Research Expert 1**

A representative of a local patient support group further confirmed this:

> "No, they never spoke to us about being worried about participating in research. Their fears are centred on their condition and not being able to get better. For example, they often mention that they fear not being able to access very expensive medications. […] Participating in research is surely not

> amongst their worries, in fact, we have around 200 members and 199 of them have ticked the box on the application which means that they would like to participate in research. […] They look at research as a form of empowerment, more than anything else." – **Support Group Representative 1**

In contrast to patients, the majority of participants in the non-patient groups were preoccupied with issues related to the storing of personal data, and thus they immediately delved into issues of privacy risk, amongst other concerns. Within the first few minutes of the sessions, the non-patient groups was engaged in in-depth conversations about trust towards the medical system and other systems within society. Whilst some were critical and outspoken in expressing their deep concerns and mistrust towards the system, others took a more pragmatic approach, one that dovetails with a functionalist approach (Luhmann, 1979) and insisted that trusting in professionals is a basic requirement to be able to function within society.

> **Lina**: "I cannot think of a reason why anyone would have a problem to participate."
>
> **Jesmond**: "Because the sample is traceable to your identity and to information about any condition which you might have."
>
> **Lina**: "But if I can be of help to others, I wouldn't even worry if I knew that my personal information was to be leaked."
>
> **Mario**: "I do worry about having my blood sample stored. Say there is a huge crime investigation going on, and my sample is used for a frame-up on myself. It is farfetched, but it is not impossible. You cannot say that there are no risks."
>
> **Lina**: "If you worry that much you end up doing nothing."
>
> **Mario**: "We've taken it too far. We can now alter human DNA. There is a doctor in China who fixes the genetics of babies before they are born. Does he know what repercussions there will be? He claims that he did it for the best, to eliminate certain conditions, but he cannot be sure that no problems will come out from what he did."

While participants such as Mario recognise the benefits which emerge from progress, they concurrently anticipate the possibility of new risks and challenges which may arise from the same progress. Uncertainty is characteristic of a risk society. What one makes up of the present has an impact on how they perceive the future (Beck, 1992). Sceptics recognise that the challenges we face nowadays are possibly past risks which had negative outcomes. As societies become better aware of such possibilities, they become more reflexive and start to challenge the status quo accordingly (Giddens, 1999). Mario's concern, which was also raised by other participants, stems from a fear of the possibility for unprecedented, undesirable outcomes as a result of innovation. Circumstances such as climate change have revealed that as the world seeks for progression, new manufactured risks with unknown consequences emerge, causing increased levels of uncertainty amongst social actors (Giddens, 1999).

Mistrust can be detrimental for genomic research and biobanking (Laurie et al, 2010), and this was a topic that was given much attention during interviews with researchers, where they claimed that trust must not be taken for granted. They described how they consistently seek to maintain the trust of research participants since a lack of trust can directly and severely impact the success of genomic research and biobanking.

> "Oh, trust is very important because if people don't trust the system they wouldn't participate and research would have to stop, full stop. Bottom line, you don't have participants, you stop! It can have a big impact yes." – **Research Expert 2**

Practically all focus group participants spoke of episodes, or moments of interaction within the healthcare system, at which their trust towards the medical system was either strengthened or hindered; it is at these "access points" (Giddens, 1990) that participants build and reconstruct their trust, based on what they make-up of the experiences they encounter.

The recollection of experiences in the data shall be analysed throughout this section in order to make sense of how such episodes construct and recreate the attitudes and perceptions of participants towards issues of risk and trust. Despite the consistency in motivation and eagerness within the patient groups towards participation in medical research, several participants recounted experiences which made them question the trustworthiness of medical professionals or the healthcare system.

The vast majority of participants in all groups had not yet engaged with medical research professionals at the time of the focus group sessions, and thus the stories they narrated were not necessarily directly related to genomic research and biobanking, but to their attitudes towards the medical and research professionals in general. In spite of this, such data is nonetheless extremely relevant to this research project since the lived experiences of participants within the medical system, and even the greater system, seem to have a direct influence on their attitudes and perceptions towards the research community, which in itself forms a part of the wider medical system.

5.3.1 **The roots of mistrust: the establishment**

"You trust authority figures too much!" – **Mariella**

Participants who conveyed attitudes of mistrust were probed to elaborate further, and the data indicate that mistrust goes beyond the medical or healthcare system for such participants. During a focus group with non-patients, after Mario expressed scepticism towards medical innovations due to feared repercussions, he was encouraged to elaborate on his perceptions of the trustworthiness of the medical system:

> **Maria (author):** "But do you trust that medical professionals, and the system, have the best intentions?"
>
> **Mario**: "As with everything else, I think there are some who have good intentions and some who do not. The problem is that nowadays bad intentions have become the norm in everything."

Mario responded by extending his argument to society as a whole, and not solely based on the medical system, implying that issues of trust or mistrust towards health systems might be rooted in a sense of mistrust towards the greater system. As a matter of fact, throughout all the sessions, even though participants were aware of the topic in question, they often referred to experiences beyond the medical system to prove their point.

> "I think we need to leave it to the professionals. We're entrusting professionals all the time… when we go to the bank, to the notary, to the doctor." – **Therese**

This implies that social actors do not focus on systems one at a time, but rather holistically develop their attitudes and perspectives based on how they interpret what occurs within society at large (Luhmann, 1979).

As already stated, patient focus group participants were significantly less sceptical, however, one participant from the patients' group was evidently not on the same wavelength as her counterparts. Mariella claimed that she feels troubled by the idea of donating samples to a biobank. Although she ultimately claimed willingness to participate, she emphasised her concerns about the whole process. Once again, her hesitation is rooted in a lack of trust towards authority figures in general, and not particularly towards medical professionals. Mariella even challenged fellow participants as they tried to convince her that what she was saying was incoherent when she was urging them not to trust authority figures so much. Her mistrust in the greater system prompts a sense of mistrust towards the research community.

Amongst her group category, Mariella stood out as being the one who is the most adamant about not excluding the possibility of misuse of data, claiming that she feels that her privacy is only safeguarded as long as third parties do not require information about her. Mariella further argued that she believes that should the need for such information arise, a breach is surely possible, as authorities have the power to get away with anything. Such a statement is indicative of a sense of mistrust towards authorities in general due to a perception of a culture of impunity for powerful figures.

> "What if it is ordered by the government? Nothing will happen to the person responsible. […] We're trusting a whole organisation which might decide to give in if the government asks for information." – **Mariella**

Giddens' theory of trust is primarily focused on a relationship between facework and faceless commitments, through which individuals build or hinder trust towards systems, however, Luhmann (1979) explores a broader perspective of trust, claiming that understanding the development of trust or mistrust is more complex than this two-way approach. The findings of this research are congruent with the findings of Ward's (2006) study, which was informed by Luhmann's theory, and which revealed that participants' mistrust towards the medical system was not merely a result of personal unpleasant experiences from their general practitioners (GPs), other healthcare professionals or the healthcare system, but rather the result of a general sense of mistrust towards the wider system, the establishment. The general feeling of mistrust expressed by such participants is attributed to a sense of mistrust towards authority figures in general.

5.3.2 **Risk related to progress**

Participants who were more sceptical than the others did not always impact the other participants' level of motivation, and their willingness to participate, however, their sceptical comments undoubtedly stirred up interesting discussions on risk and trust and encouraged other participants to speculate more, especially in the case of the non-patient groups.

> **Lina**: "Mario's argument hadn't even crossed my mind. I was like wow this is great and everybody should do it. I did not think about any negative outcomes which can emerge."
>
> **Mario**: "There is a lot of good which can come out, such as improved medicines, but the samples can be used in a wrongful manner as well."

Trust is the result of a combination of emotion and cognition, and the data which emerged from this dissertation exposes clear scenarios of what Giddens (1991) refers to as 'ontological insecurity', where social actors become increasingly informed and hence develop the reflexivity to recognise the risks and start questioning the knowledge and trustworthiness of professionals and authorities.

> "They prescribe us medications, and after years of taking them, they tell us that they can cause something which is even worse. So to avoid getting heartburn I end up risking getting something which is more serious. [...] We had moved away from glass bottles and started to use plastic, and now they tell us that plastic can be harmful and that we have to go back to using glass bottles." – **Mario**
>
> "I feel that we are moving forward and forward until we realise that we have to go backwards, and that scares me because it means that we trust and obey people who are not completely sure of what they are saying and doing." - **Grace**

These data demonstrate how, as reflexive individuals comprehend the consequences of the past on the present, they consistently make anticipations about the future as they attempt to mitigate risks whilst challenging the trustworthiness of experts (Giddens, 1991; Beck et al, 1992).

Data from research expert interviews show how, although the experts themselves also recognise and point out the risks, they emphasise the benefits which can result from progress, as opposed to the possible dangers. Preventing innovation to avoid possible risks, can be a risk in itself (Giddens, 1999). Research Expert 3 insisted that awareness of the impact of progress is essential for patients and the general public to build informed opinions and perceptions. Case in point, studies conducted on a Pan-European level indicate that both policymakers and the general public are concerned about the impact of Genetically Modified Organisms (GMOs) on human wellbeing and the environment (Drott et al, 2013), yet Research Expert 3 insisted that GMOs have been crucial for healthcare in Malta.

> "There are risks, but there are also great benefits. One of the things that the Maltese public has to realise is that our country has benefitted greatly from GMOs. People think that this is bizarre, but it is the truth. In Malta, we have an enormous amount of diabetics. Diabetics use insulin, and insulin is made from GMOs. People do not realise this. Before the technology of GMOs existed, people used insulin made from pigs. Since the protein did not match perfectly with humans, once the immune system starts to recognise the foreign insulin from pigs, the body would refuse it. Therefore patients were only offered a temporary treatment. Thanks to GMOs the treatment we have nowadays is much better. […] I am not saying that we do not need to be cautious… we need to be ultra-careful… but at the same time, we have to recognise that most modern medicines are made from GMOs. We have people suffering from severe arthritis, or severe asthma, or certain types of cancer, and they are all being treated thanks to GMOs." – **Research Expert 3**

The attitude of fearing innovation is not congruous amongst all stakeholders; patient focus group participants were less eager to elaborate on the mistrust expressed by the few, and thus the conversation on risk amongst the patient-groups was less contentious. The vast majority in these groups were outspoken about defending the cause for research participation, and even though at some points during the debate they started to recognise the potential risks, the vast majority spoke about trusting medical and research professionals, some because 'they feel they

should', and others because they essentially feel that they have no other choice. The fear of innovation was less marked within the patient groups than in the non-patients. Their desire to change the current circumstances and their sense of hope for a better future instil an attitude of perceiving progress through a more positive lens, whilst focusing primarily on the beneficial outcomes which can emerge if research is allowed to reach its full potential.

> **Frank**: "When someone holds your information, you can never be 100% safe."
>
> **Josianne**: "But what can we do? Otherwise, we can just lock ourselves inside and not leave the house, to avoid risk. You cannot overthink everything, especially when you are looking forward to something good to come out of this."

As the discussion on risk escalated amongst the parents of patients, two sub-groups with contrasting views emerged: a) those who believed that the biobank has the best intentions and therefore expressed no worries whatsoever, and b) those who believed the biobank had the best intentions but still worried about the possibility of a default.

> **Stefania**: "I truly do not worry at all about all of this. They can literally do whatever they want the blood sample. I don't believe that it is alarming if I am not informed. [My child] cannot be directly harmed [by donating]."
>
> **Josianne**: "No, I wouldn't want anyone to use the sample as they please. I would want to know about how and when it is being used. It's like I am giving them a piece of my son, even though it is simply a blood sample, it contains all information about my son."

Notwithstanding, none of the participants in this group (parents of patients) questioned the idea of participating in genomic research and biobanking, and their levels of motivation and enthusiasm remained constant. This implies that patients' parents deeply strive for more information and hope, and that no risk can demotivate them. A similar perspective was outlined by the majority of the participants in the other patient group. Once again, the general idea of this group, excluding Mariella's opinion which as described earlier revealed a high level of

scepticism, was that the donation of biospecimens for research was not of any concern whatsoever:

> **Jessica**: "I would participate no matter what. […] I'm tired of giving blood for tests. What's another bottle of blood?"
>
> **Maria (author):** "But this is different because it will be stored."
>
> **Jessica**: "Even better, at least it does not go to waste like the others. This is a zero risk for me."
>
> **Michaela**: "I totally agree."
>
> **Elisabeth**: "At least this blood sample can be used for something useful!"

It is essential to point out that this conversation happened towards the very end of a two-hour focus group session, meaning that these participants did not change their perception towards risk, despite an in-depth conversation about the various issues of risk and trust in relation to donating biosamples for genomic research and biobanking.

Although the concept of risk is more frequently associated with negative connotations, risk is not the same as danger and therefore it is completely reasonable for participants to have more positive attitudes towards risk, since without risks progress would be hindered. Whilst risk may have adverse effects, it is typically behind most successes (Giddens, 1999). Additionally, if safeguarding research participants is a must for the sake of public interest, research too is conducted in the interest of the public (Laurie et al, 2010). When compared to their non-patient counterparts, participants in the patient groups were more likely to highlight the benefits which can emerge from innovation, as opposed to the dangers.

> "I look at it from a more positive lens. I focus on the good that can come out of it. I close my eyes and trust the professionals… I believe that researchers are trustworthy. God forbid that they end up doing something wrong. I don't believe they would." – **Elisabeth**

5.3.3 Insufficient knowledge and human error within the medical system

Insufficient knowledge is key to perceptions of risk and trust vis-à-vis the healthcare and medical system. Individuals seek professional advice for comfort and more importantly for solutions, therefore, when encountering episodes which reveal a lack of knowledge, the sense of hope of such individuals diminishes as they realise that even professionals are sometimes oblivious, and consequently realise that entrusting in professionals is in itself a risk, as they too are susceptible to fail (Luhmann, 1979).

The experts recognise that nowadays the public is richer in terms of information and that this leads to higher levels of mistrust. Research Expert 3 argued that better-informed patients keep professionals accountable, yet, optimistic presumptions by patients about their own level of knowledge can be detrimental if patients decide that their google search is equivalent to the expertise of medical professionals.

> "In the past, we used to believe whatever the doctor says. Nowadays we search on the internet and we often find ourselves doubting what the doctor said. The internet has obviously lead to mistrust. […] I have friends who have mugs that read 'your google search is not the same as my medical degree', and it obviously is not the same. But there is a tendency… patients double-check their doctor these days." – **Research Expert 3**

A key point raised during the expert interviews with representatives of the local patient support group, is that one of the most common statements of concern declared by the patients within their network is about feeling that they are better informed than their doctors, and how this creates frustration and a lack of trust in professionals.

> "They sometimes know more than the doctor following their condition. […] With my hand on my heart, I can say that in some cases the patients are the best doctors to themselves." – **Support Group Representative 2**

This is what Giddens (1999) refers to as "lay re-skilling", where non-professionals start to gain knowledge and consequently express mistrust by challenging or rejecting the advice of experts to regain control over their health.

> "I know where to look for accurate information. I don't use google but I look for papers published by authorised hospitals or other research institutions." – **Michaela**

The day-to-day experiences which the patient support group encounters affect the way the representatives themselves perceive the system and the medical professionals, in fact, at certain instances during the interview the representatives indirectly expressed concerns about the trustworthiness of professionals.

> "We have also had parents who carried out their own child's diagnosis. Parents with no medical background who keep searching until they get to the bottom of things. They insist that they want to know what their kid is suffering from, and with their consistency they get there. […] Sometimes what they suspect is actually there, and no doctor would have noticed it. People are no longer ignorant. Which is good. And that is where we come in, because we help the patients to look up for information through the right sources and not through a simple google search." – **Support Group Representative 2**

Participants, especially those from the patients' focus groups, spoke of the fear which emerges when professionals appear to lack knowledge and recalled disappointing experiences where they encountered professionals who did not know how to tackle their health problems, or dealt with them inadequately. Such episodes have a direct impact on levels of trust. The father of a child with an incurable and untreatable condition recalled when the medical staff taking care of his son asked them to stop visiting the outpatient department as there was nothing else they could do.

> "I was surprised when the genetics consultant drew a line and told us to stop visiting because there was nothing else they could monitor. I thought it was

> not appropriate because at least you keep seeing him to learn more about his condition – so that if someone develops his condition in the future they would be able to guide him or her better… they'd know what the stages are. Even if they simply collect information, we wouldn't be visiting for nothing." – **Frank**

He interpreted such an encounter as the way through which the medical system informed him that they had given up on curing his son, and on curing those who will develop the same illness in the future. Frank feels that even if they did not require to keep following his child, they should have still followed-up on the development of his illness for the sake of gaining knowledge. The majority of participants from the patient groups indicated that not only would they be eager to participate in research, but also that they would be willing to do beyond the one-time visit for donation and would accept to engage in reoccurring visits, even if it is for the benefit of third parties; Frank's rendition above depicts this desire.

A common feeling amongst participants from the patient groups is the disappointment with medical professionals who do not seek to gain further knowledge from the information which is available to them, and simply work for a means to an end. Patients expressed the desire for professionals who are ambitious enough to gather as much information as possible for the sake of generating new knowledge and allowing for medical advancements. The concern of participants in this regards has a direct impact on the way they perceive the trustworthiness of professionals and the system, yet they exclude entirely the researchers from the equation, and the reason is clear and fair. The researchers seek to fill the gaps which originate from a lack of knowledge, as they constantly aim at gaining new knowledge in the hopes of improved healthcare.

First-hand experiences which reveal a lack of knowledge or misinformation hinder trust towards professionals, as well as the system as a whole. Participants from the patient groups recounted experiences which made them feel that they have no choice but to trust in individuals

and systems that are not necessarily well-prepared and well-informed to fully support them. It almost seems that through their journey of seeking for medical treatments they are consistently embarking on risky endeavours.

> "My condition is endometriosis. The lack of awareness and information is shocking. I am basically better informed than my doctors. My diagnosis took around 15 years to happen. I get excruciating period pains and they used to tell me that it's normal… they used to tone down what I feel. My very long process of diagnosis led to a lot of damage, I have stage 4 endometriosis and that means that I have serious infertility problems. I basically cannot have children." - **Jessica**

The lived experience of Jessica is not only a representation of how patients might feel when they are about to leap into the unknown by entrusting professionals, but it also exposes the repercussions which might result when such a leap ends in a turbulent landing. Due to trusting in professionals who assured her that what she felt was normal menstrual discomfort, Jessica lost her fertility after spending fifteen years of not getting treated for her condition.

> "I actually had to self-diagnose… then they opened me up to see, and when they did they found a mess." – **Jessica**

These data show the way that reflexivity brings together risk and trust; as individuals are exposed to further information they become increasingly aware of the world around them and consequently seek for ways to deal with risk whilst questioning the trustworthiness of individuals and institutions (Beck, Giddens and Lash, 1994).

As a result of her unpleasant experience, Jessica seems to have developed acute mistrust towards doctors and other healthcare professionals, however, ironically, this only strengthened her desire to participate in genomic research, as she feels that only through research can knowledge be generated, and only through improved knowledge can patients start receiving

improved healthcare. In fact, right after narrating her unfortunate personal experience, Jessica maintained that:

> "That's why I believe that research is extremely important!" – **Jessica**

This scenario came up amongst various participants from the patients' groups; episodes which seem to have somewhat hindered trust towards professionals and the system only further enhanced their eagerness to participate in genomic research. Participants perceive research as the solution to the lack of knowledge and the adversity this brings about, such as misdiagnosis or the administration of inadequate treatment. Hence, patient-participants in this study argue that lacking research is what poses risks for them, and not vice versa. The vast majority are not concerned about risks which might emerge from research and biobanking, yet, they are excessively concerned about the risk which would emerge if research does not take place or does not reach its full potential. Furthermore, through generating new knowledge, the system instils a new sense of hope and consequently restores the trust of patients.

> "It has now emerged from research that my condition is directly related to chronic fatigue. At least I now know that I am not crazy, there is a reason behind everything that I've been feeling for the past years. I used to think that I am a horrible wife; one day I'm fine and next day I would feel like doing nothing. I used to feel so guilty. Thank God for research." – **Jessica**

This was a theme that also emerged within data from expert interviews. Research Expert 2 declared that the motivation to get involved in rare diseases and genomic research derived from a revealing moment which showed the drastic outcomes which can result from a lack of knowledge due to lack of research.

> "I remember when I started my Masters, in the late 90s, we were working on this group of patients with a rare disease, and it was a treatable disease, but when they were born they were all [wrongly] diagnosed with cerebral palsy, and cerebral palsy is not treatable. So they spent their life stuck in a wheelchair essentially when all they needed was a dopamine tablet twice a day. Giving them dopamine meant they could walk, talk, and slowly they

> started regaining facilities that they had lost or not acquired. Of course, you don't regain everything because there is some damage which cannot be reversed, but had they been diagnosed at birth they would probably have gone to lead a normal life, and yes that's what got me into this field… because yes, you could actually make a change. [...] Developing new treatments is the cherry on the cake, but most times a diagnostic result is also a relief." **– Research Expert 2**

This narration shows that even if the development of new cures or treatments might sometimes seem unattainable, the generation of new knowledge for its own sake can truly have a major impact on the lived experiences of patients.

> "We can benefit from any little bit of crude knowledge." – **Research Expert 2**

Participants across all focus groups recounted experiences which reveal a lack of knowledge or misinformation on macro and micro levels. Whilst Jessica spoke about her experience related to her missed diagnosis by professionals on a micro level, other patients narrated incidents where they questioned trustworthiness on a macro level. This resonates with Giddens' (1991) concepts of facework and faceless commitments. Jessica's misdiagnosis, as well as her experience with GPs who did not seem to understand the level of pain she experiences, resulted from physical interaction, and thus it can be concluded that the facework commitment that she speaks about impacted her trust levels toward both the professionals she encountered and the medical system in general. In contrast, Tina, who is the mother of someone who has autism, recounts a situation where her trust towards the system, in this case including the research community, was hindered as a result of a system-based relation, or faceless commitment, and not through an interpersonal relation. Tina recites as follows:

> "It's important that when researchers make a discovery they ensure that what they are saying is accurate. For example, the research which had revealed that the MMR vaccine is related to autism was later turned down because we now know that there isn't enough proof about this. But in the process, a lot

> of harm was caused. Illnesses which had diminished are resurfacing because of such fake claims." – **Tina**

Human error is inevitable and participants are aware of this, yet this does not mean that they think it is acceptable, not in the case of healthcare workers or medical professionals.

> "Two out of my four sons have haemophilia. My eldest son went in for a biopsy, and the nurse asked him since when had he been suffering from haemophilia! You don't expect that from a nurse.[19] I've been through so much that you learn to build a barrier and not let such things hurt you." – **Dorothy**

Here too the data highlight a discrepancy in the ways patients and non-patients interpret professional errors. As indicated from Dorothy's narrative, who is the mother of a patient, patient-participants speak of such experiences with high levels of concern, whereas non-patients were more likely to use a humorous tone as they recounted their own experiences, even if the human error which they encountered could have had serious negative outcomes. Below are two narratives of two non-patient participants of this research which reveal a more mellow perspective to situations of omission on a micro-level by medical professionals.

> "I was at a private hospital to do a nose operation and instead they almost ended up doing an appendix operation on me. They called my name, I went in and he told me 'we'll soon have your appendix out, Madame'. I told him no, I am not doing my appendix, I'm doing my nose. There was another person with the same name." *laughs* – **Lina**

> "Something funny happened to me recently. I received a paper which says 'Mrs [surname], you are being invited for a mammogram to test for breast cancer'. No wonder I never received the paper to go and do the prostate test... I am listed as a woman on their records." – **Peter**

[19] Haemophilia results from a genetic defect; those who suffer from haemophilia are born with the condition (Peyvandi et al, 2006).

The major discrepancy between the patients and non-patients in this regard boils down to what the two groups regard as a major concern. It emerges vividly that the risks which concern the vast majority of patients were directly related to their physical well-being and their health, whilst in the case of the non-patient focus group participants, the risks which topped the list of concerns were more likely to be related to privacy and issues related to power and control.[20] For patients, research participation is in itself interpreted as a venture which barely entails any risk, whereas a lack of investment in research which leads to a lack of knowledge is interpreted as the major issue of concern. They prioritise the physical over the incorporeal; they are fully focused on attaining the healthiest-self possible, and thus, if there is no risk of physical harm, then there is no risk at all. They disallow incorporeal concerns from limiting their motivation towards the ultimate aim of achieving improved health.

> "To me, this [participation in genomic research and biobanking] is not risky at all. They really have to dig deep to be able to find out something about me. There is nothing which can be done which can harm me physically". – **Jessica**

> "I want to do anything it takes, everything, to make it at least 1% better. […] Even if just 10% of the outcome of the research is positive, it would still be worth it." - **Elisabeth**

As opposed to patients, participants from the non-patient groups did not commonly express concerns related to a lack of knowledge, or a lack of financing for research, and were more likely to speak of the risk directly related to donating biospecimens for research. In summary, patients struggling with the challenges of living with chronic disease fear the risk of restricting research, whereas non-patients were less likely to recognise this limitation and immediately

[20] Theme to be analysed and discussed subsequently.

focused their discussion on risk towards the potential harm which might emerge from genomic research and biobanking.

5.3.4 Privacy risk: data breach or data leak

Privacy risk can be considered as one of the key risks associated with participation in genomic research and biobanking. The donation of biospecimens, in conjunction with other sensitive personal data, requires entrusting an institution with resources which have the potential to reveal significant information about oneself. Hence, genomic research and biobanking are associated with various ethical, legal and social implications (Kaye et al, 2015). Despite ensuring their commitment to professionalism and best practices in research, the majority of the research professionals interviewed acknowledged the risks, especially since not even experts can predict where technology will take research in the future.

> "There are issues and some of them arise from the fact that we don't know where the technology will go, so you will have your sample collected under some agreement and then the technology moves on and it becomes difficult to re-contact these people. […] There is the risk of mismanagement of samples. There are ethics protocols in place and one should always abide by those protocols of course, but there is the risk. There is always the risk of hacking data." **– Research Expert 2**

Innovation is indispensable for progress, yet the risks which emerge through technological advancements are typically unpredictable (Schmilden, 2016). As technology continues to develop, the possibility of identification increases, and privacy risks increase (Balaji and Terry, 2015). Nonetheless, the interviewed researchers described their utmost efforts to safeguard the privacy of research participants, with the majority of the experts claiming that they choose to store data on "offline servers" to provide an added layer of protection. They also declared that

all genomic data is pseudonymised and that access is restricted to specified individuals, as indicated on the consent forms for their respective studies.

> "If a hack is large enough, possibly people can be identified. Which is the reason why we have decided not to put data freely available on online servers and added a layer of protection for the participants." **– Research Expert 2**

The issue of privacy risk immediately sparked interesting discussions during the focus group sessions, especially within the non-patient groups. The mechanisms mentioned by the experts which are aimed at protecting research participants from privacy breaches are not necessarily enough for gaining their trust. The discussion within the non-patient groups revealed that participants are particularly concerned with individual data leaks, as opposed to systematic breaches into the data by hackers. Most participants were less concerned about the possibility of a data breach as a result of a coordinated hack of data systems, claiming that such a possibility is rather far-fetched. However, they did seriously consider the possibility of data leaks and argued that close-knit communities, such as the Maltese community, make data leaks even more probable than in other cultures. This argument was raised by several participants during both of the non-patient focus group sessions:

> "Everyone knows each other in Malta. I know people who work at the department of health, others who work at the department of education. If someone wants to know something, they just ask for information and get it. That's how things work in Malta." - **Adriana**

> "I have friends who work at the hospital... they all gossip a bit about what they see, and then they tell you to keep it a secret." - **Mark**

Lina supported the argument raised by the other participants by narrating her personal experience of working within an institution which holds sensitive personal data:

> "Back when I worked at the bank, the most important rule was not to speak about clients outside of work. We recently were a party and there was this ex-colleague who was speaking about one of his clients, he was saying that

> this man used to come to our branch and that he had that much money and this and that. I left the bank a long time ago and for me it is still a huge deal to do that. He was exposing all that he knows about him... in front of everyone." - **Lina**

During the focus group discussions, participants indicated adequate levels of awareness about the objectives of the General Data Protection Regulation at safeguarding research participants from misconduct (Bovenberg et al, 2017), and they were also shown a video clip which further explained their data protection rights. Nonetheless, awareness of laws and regulations did not seem to have much of an impact on the perceptions and attitudes of the participants within the non-patient groups.

> **Maria (author)**: "May I remind you that there are laws and regulations against the breach of data or misuse of samples… such as the GDPR."
>
> **Mario**: "But people break the law all the time!"

Participants proved their point by giving accounts of incidents of data leaks which occurred in Malta and which had adverse consequences. Once again participants recalled and narrated episodes which were unrelated to health and which happened outside of the system in question. This further reinforces the argument that social actors do not merely develop attitudes towards a system based on the experiences they encounter within that system, but ultimately they reflect the general attitudes and perceptions which they develop onto individual experiences within specific systems (Luhmann, 1979; Ward, 2006).

> "Some years ago there was a case with a telecommunications company in Malta. A man had a friend who worked with the company and he would give him information about the whereabouts of his ex-girlfriend. With that information, he followed her and killed her." - **Mario**
>
> "To me confidentiality is just a word... it doesn't really exist in Malta. I surely cannot say that I am 100% sure that the biobank would not expose information about myself if asked to do so, especially if the police are asking for it, even though I don't believe that they should." - **Lina**

The three main reasons behind the possibility of data leaks which were raised by the participants are: a) for gossiping, b) for doing favours for relatives or friends by providing information about a third party, and c) for law enforcement.

In the case of law enforcement, there was a disagreement amongst participants on whether such a leak would be legitimate or not. Approximately half of the participants in the non-patient groups argued that solely in this case, the provision of personal data to third parties is reasonable, however, the other half were adamant that such action would set a dangerous precedent which allows for the unauthorised sharing of data with third parties.

> "So I give my sample out of goodwill, and all of a sudden I make a mistake, I become involved in a crime, and they use the information which I would have given against me. It does not make any sense. Irrespective of what I've done, I participated for the sake of research and information about me should be used for that purpose only." – **Peter**

Expert interview data highlight how the information held by the biobank at CMMB is not passed on to third parties, and this includes authorities.

> "Personal results are not made available to third parties such as employers, governmental organisations, insurance companies or educational institutions, unless they provide specific authorisation. This also applies to spouses, partners, family members or family doctors." – **Research Expert 4**

This information was passed on the participants during the sessions to clarify the practices and procedures put in place by the University of Malta biobank. In spite of this, participants who had expressed scepticism towards the greater system did not seem to be convinced with such a statement.

> "They say the data that you put online can come back to harm you. You give a lot of 'likes' to this or that- then they make a profile of you and target you politically or for advertising. We can get targeted with DNA as well. What if the government really wants to get access to it? A totalitarian government

> that wants to get rid of people with disabilities to clean up the race would do anything to get this information. I mean that is extreme... but well these things have happened. Maybe they find out about who is defective and disallow them from reproducing. […] Imagine they do research on your DNA and they learn to identify your illness before birth, and they create a chemical which aborts the unborn child with your illness. If they see that a baby has the characteristics they give them this chemical and the baby is aborted. They do research on your DNA to turn against people like you. This is already happening in other countries with babies who have Down syndrome." – **Mariella**

Mariella's comment is, by her own admission, extreme, however interesting as it highlights the extent of the insecurities in the public imaginary. Varying levels of concern in relation to privacy risks not only occurred amongst groups but also within them. Despite a somewhat clear consensus amongst most patient-participants, Mariella was one particular patient who demonstrated a distinctive attitude towards privacy risk and was sceptical of the practice of donating biospecimens for research throughout the session. It is interesting to note that her perspective of why privacy risk was a major concern differed from what non-patients had declared. Mariella was less concerned about a breach for gossiping purposes, or law enforcement, but instead linked privacy risk directly to her condition, and gave an example of a concrete negative outcome, as she presumed the risk of eugenic potential through the deprivation from the right to reproduce or through abortion. Mariella's lack of trust in the government prevents her from perceiving the law as a means of protection and argues that if there were to be a breach on a macro level with the intent of eugenics, such act would be structured and organised in a way to avoid legal action. Mariella insists that irrespective of whether such an act is legal or not, anyone would get away with it if the breach were to be ordered by the government.

> "…but what if it is ordered from the government? Nothing will happen to the person responsible." – **Mariella**

Mariella's perspective of privacy risk stems from her fear of data breach with the intent of directly causing harm to patient-participants or the patient-community. Discrimination on the basis of genetics exists, and whilst concerns are typically related to disadvantages in employment opportunities or health insurance (Gostin, 1991), Mariella's concern with eugenics is not surprising as there is evidence in the literature of the impact the spectre of eugenics may have on trust towards research professionals. Research by Domaradzki and Pawlikowski (2019) shows that levels of trust towards the field of medical research tend to be lower amongst ethnic minorities who have previously experienced eugenics.

Focus group data also highlight the way that a participant[21] was singled out by the other participants when she claimed her deep concern about the dangers of data breaches. As the other parents discussed their lack of worry about the risks, Tina kept quiet throughout, however, she later expressed her concerns claiming that she was trying to keep the condition of her child undisclosed.

> "What you are all saying worries me. Many people in my family do not know of my child's condition, and I want to keep it private. The possibility of a breach worries me. […] Everyone has a right to privacy." - **Tina**

The fears of Mariella and Tina highlight two of the core privacy concerns associated with genomic research and biobanking; Mariella speaks of a risk which is rooted in fear from direct harm towards oneself by their community, whereas Tina perceives a data leak or breach as a means of intrusion into her private life which can, in turn, lead to stigma.

These two sceptical interventions described above both caused reactions of disapproval by the other patient-participants, reinforcing the finding that on a general level, patients were not

[21] The mother of a patient.

concerned with privacy risk, which contrasts with the findings in the non-patient groups. They might have recognised the risk, but they chose not to be concerned about it and expressed full trust in the medical research system and those who make it up.

> "I don't worry at all. My duty is to help out so they would be able to carry out studies which could help other people, or maybe myself who knows. After all, if they do not use the sample right it cannot really affect me personally. I would have done my duty and my conscience would be clear."
> – **Michaela**

Michaela represents the attitudes of the vast majority of the patient focus group participants, whereas Mariella was an exception within her category. In contrast, within the non-patient groups, perceptions about privacy risks were more balanced:

> **Luke:** "I believe that the chances for a breach are very small. [...] To me this is not sensitive information, it's not like financial information."
>
> **Robert:** "I disagree. The possibility for a breach worries me deeply."

Even though a few non-patients, such as Luke, were adamant about not being concerned, the reasoning behind their perception is not attributed to a lack of fear of the possible repercussions but rather a result of not recognising what negative outcomes can emerge from a breach. They spoke of genetic data as data which is not sensitive, and thus their lack of fear of a breach is indicative of a lack of knowledge of the possible, though unlikely, negative outcomes. The varying levels of health literacy and knowledge create a stratification amongst potential participants, those who are better aware are better equipped to make informed decisions, whereas those who lack the knowledge are more likely to take leaps into the dark without even realising.

In conclusion, it appears that when encountering a situation where they have to make a health-related decision which requires an evaluation of risk and trust, patients are most likely to react as what Giddens (1990) refers to as "sustained optimists", who perceive the medical field as a

source of positive outcomes which operates under the best of intentions. Nonetheless, this cannot be stated for all patients, as Mariella expressed an opposing perception and adopted an adaptive reaction which Giddens (1990) refers to as "cynical pessimism". Furthermore, non-patients are split into two groups, those who, like Mariella, react as "cynical pessimists", and are therefore are critical and not optimistic about the intentions of the medical system, and those who are "pragmatic acceptors" (Giddens, 1990), who choose to ignore the risks and limitations because they would rather avoid challenging the system. Even amongst the optimistic participants, there is a disparity between patients and non-patients; whilst the latter group might not have been critical towards the system, at the same time they did not express excessive enthusiasm towards the way the system operates. In contrast, patients who spoke with optimism, expressed a quasi-religious attitude towards medical research institutions, as though their hope for a miracle suddenly depended on researchers, and they somehow are confident that the aim will be reached, maybe not within their lifetime but eventually.

5.3.5 Repercussions of a data breach: the risk of genetic discrimination

The theme of concerns about the risk of data breaches was also one that emerged in the data from expert interviews. The biomedical researchers interviewed for this dissertation emphasised that the intentions of the research community at large are genuine, and that research based on sound ethical practice is the norm amongst the significant majority of researchers. Yet, they did acknowledge the risks as they claimed that the donation of biospecimens for research is not entirely safe, and that the possibility for a breach persists.

> "It is personal data, it is genetic data, and the reality is that with enough effort, if you sit down and look at a person's data and compare it to other databases and other data, you can actually identify individuals. […] It can be used by insurance companies to decide who is insurable and who is not

> insurable. So with enough resources and effort, data can be hacked. So yes these are the real risks." **– Research Expert 2**

In relation to this issue, the participants in the non-patient focus groups were roughly split into two groups with contrasting views; around half of the participants believed that a breach was unlikely, yet a possibility, and thus were somewhat concerned, and the half who believed that a breach was extremely far-fetched and that even if it were to happen the consequences were not too troublesome, and therefore expressed no concern whatsoever.

The risk of genetic discrimination, which is a direct consequence which can emerge from a breach of personal genetic data which is leaked to third parties, was not one of the key issues raised in the focus group discussions. In fact, only Mariella had mentioned an example of how a data breach can lead to eugenics and thus discrimination based on genetics. However, after some time of being quiet and listening to the others discussing their levels of concern in relation breaches of data, Claire asked a question which changed the dynamic of the discussion of one of the groups.

> "May I ask you something? Could somehow insurance companies get access to such information?" – **Claire**

Suddenly there was a concrete example of how a data breach, or a data leak, can cause actual harm.

> **Marica**: "Ah no! No one has the authority to make such findings about me."
>
> **Claire**: "But didn't you say that you didn't care if a breach was to happen?"
>
> **Marica**: [laughs] "Hmm… I hadn't imagined how it can affect me."
>
> **Adriana**: "Hmm… that can lead to discrimination."
>
> **Claire**: "Even employers might be able to use such information to check about any illnesses."

Marica: “But would they bother?!”

Claire: “Of course they might bother. They can use such information to employ healthier individuals to avoid employees who might need more sick leave. That would be extremely discriminatory.”

Marica: “But that’s totally illegal!”

Robert: “So what! People break the law. […] These things barely cross our minds, but there can be repercussions.”

Claire: “I know that the possibility for this to happen is very small, but it still worries me a lot. I feel that only I should have access to such information, not even family members need to know such information, and strangers definitely shouldn’t.”

Maria (author): “But what is the possibility of this actually happening?”

Claire: “Of course there is a possibility because we are humans. If the database contains details of thousands of people than the possibility is even bigger. It does happen in Malta that people ask friends or family members for information about others as a favour… why wouldn’t it happen in this case?”

Luke: “But they simply cannot do that!”

Claire: “But it happens anyway!”

Robert: “There is a huge difference between what should happen and what really happens.”

Luke: “Ok there is a possibility, but at the end of the day if I participate, I would be doing it for the best, for research. Worrying too much leads to no good and we end doing nothing. We must be realistic here.”

Claire: “Well I was with you on this at the beginning, but the more I think about it the more I think of things that can go wrong.”

Participants who had previously spoken with indifference about privacy risk were suddenly questioning the possibility of this happening and expressed higher levels of concern. This reconfirms that not being concerned about potential repercussions is not typically the result of a lack of fear of the repercussions but it primarily results from a lack of information and knowledge of the same repercussions. Nonetheless, even after acquiring such information,

various participants were quick to shake off such concerns, claiming that such concerns are far-fetched.

> "I still cannot even imagine that a professional employer or insurance company would try to make a find out about the genetic makeup of other people, and I cannot imagine medical professionals sharing such data. I think you are being unreasonable." – **Luke**

Whilst Luke, who is a non-patient, attributed his lack of concern to the implausibility of negative outcomes, most patients decided to ignore concerns, irrespective of whether they felt they were plausible or not, and expressed a desire for progress not to be hindered, even if it incurred high probability risks.

> "I would still donate even if I knew that there were great risks. Even if I was sure that there would be a breach. We have to trust so much in our everyday lives, that it does not make sense to try and avoid risks, because it is impossible. At the end of the day, if it involves people, there is always the possibility of default. [...] What matters is that something good will come out." – **Therese**

5.3.6 Participant responsibility

As focus group participants discussed the issue of risk vis-à-vis participating in genomic research and biobanking, an interesting conversation about who it is that should hold responsibility came up. This discussion mainly cropped up amongst non-patient focus groups, and this can be attributed to the fact that patients were less likely to be concerned about the risks.

> **Marica:** "When a person takes a medication or is participating in research, or whatever, they must make sure to do their own personal research to check about the trustworthiness of the medication or the institution doing research."
>
> **Adriana:** "But do you really think that the general public does that? They just obey their doctor's orders and do as they're asked, that's it!"

> **Marica**: "But as a patient or participant you have an obligation to do that."
>
> **Adriana**: "That's surely not what happens... especially when it comes to the elderly or those who cannot read and understand. It is the responsibility of the professional to ensure that the other person is well-informed."

These data demonstrate the concept of 'stratified reflexivity' (Ward and Coates, 2006). Whilst Giddens (1990) speaks of reflexivity as a characteristic of modern societies where social actors develop the capabilities to recognise risks and henceforth start to question the trustworthiness of social systems and authority figures, Ward and Coates (2006) elaborate further and distinguish between varying levels of reflexivity amongst members of society. There is empirical research in the literature that shows a correlation between reflexive action and socio-economic status; disadvantaged individuals within the social hierarchy tend to have less access to information as opposed to those who with a higher socioeconomic status (Lupton, 2012), thus health literacy and levels of reflexivity are not to be considered constant amongst all members of modern societies. This is reflected in my data where some participants argued that one should not assume that research participants are capable of informing themselves and that therefore it is the responsibility of the research institution to ensure that the relevant information reaches the research participants, to avoid giving privilege to the "information-rich" over the "information-poor" (Elliot, 2002).

> "Ok, some do question what the professionals say and do their independent research, but it is not very common. Not everyone is capable of doing that, and not everyone feels the need to question the professionals... I would say most trust in them. We cannot really doubt the professionals if we are not knowledgeable of the subject... that's why we simply trust in them." - **Claire**

Risk is directly linked to responsibility (Baker, 2002). As claimed by Beck during his interview with Joshua Yates (2003), risk is becoming more prominent, as it is becoming increasingly difficult to identify the holder of responsibility for outcomes, particularly when they are

negative. As modernity is faced with a responsibility crisis, social actors find it increasingly difficult to attribute responsibility to one entity or individual (Giddens, 1999).

Awareness of risks associated with participating in genomic research and biobanking was generally very low within the focus groups. With some individual exceptions, the vast majority of participants showed no knowledge whatsoever with regards to such risks, particularly towards the beginning of the focus groups session. As the sessions progressed, through exercises of brainstorming, all groups managed to come up with, and discuss, some of the risks associated with the donation of biospecimens for research. Consequently, it seems unreasonable to expect genomic research participants to be responsible for making all the necessary inquiries before participating in research and biobanking. Rather, that it should be the responsibility of researchers and research institutions to ensure that participants are adequately informed about the risks and benefits of participation. For instance, the GDPR expects researchers to provide all participants with the necessary information using simplified terminology (Bovenberg et al, 2017).

The need for researchers to adequately inform research participants came out clearly in the data from the expert interviews, where one described how not even individuals who are expected to be knowledgeable of genomics tend to be aware of any possible risks. When asked about whether they think that participants think about the risks or not, Research Expert 2 responded as follows:

> "You find all sorts, there are those who are not aware of risks, and there are those who are a bit paranoid about their sample. What will you do with my sample, where will it end up? And it's fine, it is their sample after all. However, the vast majority haven't thought about it enough, including people who are following science courses, and who would know about DNA and all that. I do ask students sometimes… they're studying the sciences... every year there is this black Friday deal to get your genome data sequenced for 300 dollars and they give you reports about the risk for 200 diseases and

> your intolerances for 500 things and a good workup for a disease of your choice. And I always do this session where I ask the students 'would you do it?', and they're all like 'ah 300 dollars isn't too much for all that'. Most say that they would do it without thinking twice. Then I go through the whole thing with them, and when they think more about it, in a number of cases they change their mind – some don't, but some do. I think the reason is that people haven't thought of the repercussions in most cases, and so we have to explain what it is all about." **– Research Expert 2**

5.3.7 Impact of risk on willingness to participate in research

> "I wish I could trust blindly but I don't trust much. However, I still believe that there is a huge need for more research, and so I'm not discouraged to participate." - **Adriana**

The risk-benefit analysis which Adriana makes here was similarly made by all participants in all focus groups. Irrespective of the level of concern which certain participants might have expressed, they ultimately all remained motivated to participate in genomic research and biobanking. Regardless of their attitudes of mistrust towards the broader system and their negative perceptions towards the risks associated with participating, especially in the non-patient groups, they ultimately seemed to express faith towards the research community.

This research reveals that concerns of risk and trust and the motivation towards participating in genomic research and biobanking are not mutually exclusive. Motivation typically allays any worries which might arise as one thinks deeper about possible risks. Irrespective of single episodes which might have hindered trust and irrespective of perceptions of mistrust towards the greater system, all focus group participants remained committed to accepting an invitation to participate in genomic research, should they be invited. The focus-group conversation surely instigated the participants to think more deeply than one normally would about issues of risk and trust.

The general belief that emerged from the data is that the research community is trustworthy and that their intentions are generally benign, nonetheless, most participants managed to think of at least one concern when pressed to discuss the issue of risk. Some participants did claim to be preoccupied with possible abuse, although not to the extent to demotivate participation.

Even participants who spoke of high levels of mistrust and who were consequently highly concerned of privacy risks, such as Mariella, all made a deliberate privacy versus utility trade-off (Oliver et al, 2011), and ultimately showed willingness to participate in genomic research, albeit under specific conditions, such as a consenting procedure which is up to standard. All participants spoke of the need for genomic research and biobanking, and all agreed that the conceivable benefits out-weigh the concerns.

It is clear that whilst certain participants were more sceptical and expressed deep concerns, other participants, in both the patient and the non-patient groups, did not even feel the need to make a trade-off between the beneficial outcomes and the risks as they insisted that any scepticism towards medical professionals is baseless and declared high levels of trust towards the research community. As described by Luke:

> "I trust in them with my eyes closed. Sometimes people expect too much… doctors have their limitations, research has its limitations, and people must be aware of this. I believe that doctors and researchers have the best intentions and are genuine… those who aren't are the exception and are the very few." – **Luke**

Despite the varying levels of concern amongst participants, the data which emerged from the focus groups show a clear consensus amongst all groups with regards to willingness to participate; non-patients, patients, those who expressed scepticism and those who did not, all claimed that they would choose to participate if they were to be invited. Moreover, the focus group discussion, in itself, seems to have had an impact on the attitudes and perceptions of

those who started with blind trust in the research process and were previously unaware of possible risks, yet, this did not seem to have an impact on their willingness to participate.

> **Maria (author)**: "Would you mind participating if it means that they would need to store your personal information at the biobank?"
>
> **Grace**: "No I wouldn't mind. Well, there are risks... what the others said did make me think... at the start I would have given you an easier yes. An invitation would get me thinking but ultimately I would surely say yes."
>
> **Mark**: "I think everyone should do it. There is more good than bad that can come out of it. The probability of something going wrong is small!"

This non-patient group was undoubtedly the group which generated the most fevered discussion and flagged various issues of fear. Nonetheless, when towards the end of the discussion they were asked about whether they would participate or not, a consensus was clear:

> **Grace**: "For sure!"
>
> **Lina**: "100%!"
>
> **Peter**: "Me too."
>
> **Mario**: "Of course I too would participate. Still, I cannot understand how people can be so naive and not think of how things can go wrong... I do know that things can go wrong but as long as I know that some good can come out I would still choose to participate."
>
> **Mark**: "We can go give the sample now if you want to. (laughs) All I'd ask for is to be informed about what the study is about."
>
> **Jesmond**: "Obviously yes... it would be great to contribute to the healing of certain illnesses."

Similar to what is discussed in the literature (Ducournau and Strand, 2009), whilst some participants portrayed "natural trust", others required more deliberation and challenged the trustworthiness of the institution before coming to a conclusion and making a decision. Some expressed major concerns, and others argued that they feel that the donation of biospecimens does not incur any risks. Some feared the possible dangers of passing on their genetic data for

research, and others feared restrictions which prevent research from reaching its full potential. Yet, irrespective of the varying attitudes and perceptions of risk and trust, all participants in all focus groups agreed that they would accept to participate in genomic research and biobanking if they were to be invited; some simply claimed their willingness to participate, whilst others enthusiastically stated that they desire to be invited.

This finding in itself shows that there is a strong consensus amongst focus group participants that even if risks do exist, this venture is worth the risk, because the plausible positive outcomes are greater. The final choice of participants reflected their hopes and not their fears. This finding resonates with studies conducted on a global level, where willingness to participate in genomic research is drastically higher than unwillingness, across the board, around the world (Domaradzki and Pawlikowski, 2019). "Most are willing to share their data despite concerns regarding government oversight, privacy and confidentiality, and profiteering or misuse of data" (Oliver, 2011, p.107).

Despite their willingness to participate, the majority of participants of this research expect to have some form of control over how their personal and genetic data is processed. Their willingness to participate is not unconditional, as several focus group participants declared that their contribution might depend on the particular project's approach towards specific issues such as the consenting process, data sharing and incidental findings, as shall be discussed in the next theme.

> "I would want some control over it. After all it is my life essence. It is me."
> - **Mariella**

5.4 Attitudes towards power and control

A theme which emerged through the focus group discussions and the interviews was that of power and control, as participants typically referred to issues which highlight the power dynamics associated with the involvement in genomic research and biobanking. It has been established in the previous chapter that knowledge is a determining factor in terms of building trust. The work of Michel Foucault reveals how knowledge and power are interrelated, in the sense that power operates through knowledge, and knowledge generates more power. The two concepts occur mutually through each other, and as a result one group, or individual, gains control over others (Foucault, 1976). In the previous theme, it was revealed that when professionals appear to lack knowledge, or rather, are inconsistent in their knowledge, non-experts develop concerns, which in turn demean the power of professionals. Consequently, non-experts start to regain control over their personal health and other life matters (Giddens, 1999).

Despite the fact that all the participants of this research remained motivated to participate in genomic research throughout the focus group sessions, there were several instances where participants reclaimed control and argued that they would prefer to set certain conditions for the researchers in the case of specific scenarios. Even participants who expressed a quasi-religious attitude of trust towards health professionals and researchers, still felt that they needed to attain some power and control in the process of research, and they disliked the idea of being passive participants who are completely detached from the research process. Whilst some demanded some sort of general feedback on the progress of the project from the research institution, others claimed that they expect researchers to come back with any incidental findings, whereas some even insisted that they would be hesitant to donate their biosamples to researchers who choose not share data with other researchers.

The latter concern was the least expected, as the literature predominantly points out issues of data-sharing in terms of privacy risk, whilst my findings show that the only prevalent concern of participants with regards to data-sharing was expressed towards researchers who refuse to share data. The participants perceived such a decision as resulting from an attitude of selfishness and personal attainment, as opposed to achieving the best results for the greater good in the shortest amount of time.

Experts possess the powerful tool of knowledge (Ward, 2006), nonetheless, the level of control they exercise is largely dependent on the level of knowledge and trust of the other group (Luhmann, 1979), in this case, those donating their samples.

It was clear at the start of the focus group sessions that the participants had very little knowledge about the process of biobanking. The vast majority of the issues raised during the sessions were put forward by the moderator, as the participants declared that they had barely ever thought about the practice of genomic research and biobanking, let alone about the issues which are associated with such venture. This knowledge vacuum creates a discrepancy between the level of control which the participants of this research request, and the level of control which potential genomic research participants would claim when they are about to give consent for their participation. To be able to make an informed decision, one must have some form of knowledge. Thus, the information-poor are always at a disadvantage in taking control of the situation, thus being more susceptible to accepting whatever is advised and giving full power and control to the professionals (Domaradzki and Pawlikowski, 2019; Elliot, 2002).

Modern societies are reflexive and tend to be better aware of the risks and other issues related to health and medicine, yet, awareness about the issues related to genomic research and biobanking remains rather low (Domaradzki and Pawlikowski, 2019). Additionally, one must consider that “knowledge does not empower to the same extent everyone who possesses it”

(Fuller, 1999, p.28) and therefore, even when researchers or biobank managers take the time to point out the risks and benefits of research, participants might not all process the information in the same manner.

> "Of course it's part of the informed consent that you explain both the benefits and the risks to people. […] At the end of the day I would say most listen and sign, then there are a few who ask questions and who try to understand more." – **Research Expert 2**

This excerpt indicates that even if individuals' desire is to have a certain level of power and control, under the real circumstances most individuals choose to abdicate the power that they hold as participants, and often choose to act passively and trust in the professionals. The key reason for this is perhaps because most individuals struggle to recognise the risks unless probed to think deeper about the issues, and thus they do not recognise the implications related to genomic research and biobanking unless provided with the opportunity to engage in an in-depth discussion about the topic. This was underlined by Therese, who is a patient participant who had already given her sample for medical research prior to the focus group session, and also by others who had not yet participated in genomic research or biobanking.

> "When I gave my sample, these things did not even cross my mind. I am now starting to realise what it really involves, through this conversation." – **Therese**

> "It is difficult for participants to consider all the risks that we spoke about after an hour and a half of discussing… it requires a lot of thinking." – **Claire**

The focus groups sparked intense discussions on the power dynamics which come into play when donating biospecimens for genomic research and biobanking. The key issues which provoked discussions of power and control were: commercialisation, incidental findings and issues of ethics, law and consenting, such as data sharing.

5.4.1 **Commercialisation: the involvement of Big Pharma**

The issue of the involvement of Big Pharma in the research process sparked concerns amongst the vast majority of participants. A key finding in my data is the evident sense of mistrust towards the pharmaceutical industry, by both patients and non-patients, however, Big Pharma is also considered crucial for providing a sense of hope, particularly amongst patients.

> "I think that pharmaceutical companies benefit the most. They aim to make money out of this, but ultimately they give hope to the patients, and I believe that if you take away hope from patients, you take away everything." – **Support Group Representative 2**

The participants versus Big Pharma

> "It depends on who the researchers are... I have less trust in research financed by pharmaceutical companies because all they care about is profit." – **Adriana**

Participants make clear distinctions between Big Pharma and the researchers and were surely more sceptical of the intentions of the former group, however, they nonetheless recognise the importance of the involvement of the pharmaceutical industry in the development of novel medications.

> "I think that pharma people do not really care about us, but the researchers are totally different people. I would donate irrespectively. At the end of the day, with no research and without the pharmaceutical industry we will never get there. I see some negatives and some positives, but I try to focus on the positives." – **Elisabeth**

Elisabeth here declares that the people, the participants, have no control. This statement triggered a critical reaction from some of the other participants in this patient group, who declared that they are not willing to allow Big Pharma to take full control. Participants here felt

the need to reclaim power and control and started to speak of conditions or precautions which they would expect to be implemented. Therese recalled how some patients struggle to survive because the cost of medications is sometimes beyond what they can afford. She argued that if pharmaceutical companies are to claim all the profit which emerges from the development of novel medications, they should at least ensure that medications are affordable for the patients, who often would be contributing to further research through their participation.

> "You should speak with people who have to pay thousands of euros just to stay alive. I think that there needs to be some form of regulation on the prices of medicines. I wouldn't expect any profit from the breakthrough, but at least I should be able to afford it. After all, there were patients behind that breakthrough." – **Therese**

At the introduction of the issue of commercialisation into the discussion, the majority of participants in the patients' focus groups began to manifest scepticism or critique. The fear of lack of access to medications impacts attitudes of potential participants as they start to view the medical system as a capitalist institution which solely aims for profit, as opposed to humanitarian action. Yet, the general attitude of the patient focus groups participants towards Big Pharma seems to be bittersweet. On the one hand, they perceive it as an entity which is merely motivated by profit, but on the other hand, they perceive it as the entity which can make the attainment of the ultimate goal possible. Even if they all spoke of Big Pharma through a sceptical lens, they ultimately all hoped for its involvement.

> "Pharmaceutical companies are after business and that's how business works. I'd rather have the option for a very expensive pill, then no option at all." – **Jessica**

Some were more accepting of the profit motive of the industry, whilst others were more resistant and insisted that they believe that Big Pharma is motivated by greed as opposed to profitability. Two key points emerged behind the latter reasoning. First of all, as mentioned by

Therese, there is a concern that some medications are beyond affordable and therefore pharmaceutical companies create inequalities in health risks amongst patients, who are also potential genomic research participants. Secondly, that the pharmaceutical industry chooses to develop novel medication depending on costings over anything else. Participants argued that this ultimately implies that the development of a cure, treatment or diagnostic test depends entirely on a cost-profit analysis, hence why rare disease patients often end up with no treatment options whatsoever (Agius, 2018). This argument was further reinforced by the research experts.

> "The industry benefits in a number of ways. First of all to start designing a drug they need to know what they have to target, and through genetic research we inform them about which enzyme protein is malfunctioning. The patients will then potentially benefit from the drugs which they might develop. Knowing the number of patients who would benefit helps them do their costings and this is why rare disease patients tend to be left behind because if they can design a drug which will target thousands, they would go for that rather than one that will target 10 people." – **Research Expert 2**

It can be argued that whilst patients are willing to do their utmost for the sake of research and the greater good, with minimal hope for self-gain, they also feel that whether they receive something back or not depends merely on costings. Once again, even though participants express scepticism and the desire to take ownership and have control over the situation, they quickly realise that they lack the resources and ergo that Big Pharma holds the power to control the future of millions of patients around the globe, including themselves.

Non-patient focus group participants likewise perceive pharmaceutical companies as driven by profit, and frequently argued that although this might be illegitimate it is expected. Participants in these groups, further claimed that GPs and other health care professionals are also controlled by the industry. Participants also commented on the power relations between Big Pharma and medical professionals, with comments that the former gains power over the others through

economic means, particularly through providing them with commissions. The absolute majority of non-patients, from both groups, seemed to be in agreement on this rhetoric.

> "It's amazing how the doctor has to write two whole pages of prescriptions whenever I go to him. […] There is the ideal world and the real world. Ideally, doctors would care for the people, but the reality is that would rather earn some extra cash. […] But you must also understand why the pharmaceutical industry does that. If they don't charge certain prices they wouldn't be able to do more research and improve medications." – **Robert**

Robert here justifies overprescribing and overcharging and put forward a similar assertion to that made by the patient focus group participants. This perspective implies that whether the actions of the industry are equitable or not is irrelevant, as participants recognise that the fate of medical progress depends on it.

The discussion on Big Pharma was characterised by several conspiracy theories, and this, in itself, is a finding which highlights the suspicion of participants towards the commercial entities involved in research. My data show that the general feeling is that the pharmaceutical industry perceives patients as customers over anything else.

> "What have they cured in the past years? They give you treatment for a lifetime but not a cure. Having a common condition such as high blood pressure means taking medication until death, and for pharma, it means a customer for life. […] People can afford a few euros a month but might not afford to pay a large sum at one go for a cure. So if they provide a one-time cure it would not be as profitable as a treatment. They only consider profitability and they provide medications accordingly. Hundred per cent they'd rather provide treatment over a cure, irrespective of what would be most beneficial for patients." – **Mario**

Most of the other participants in Mario's group agreed with him claiming that money dominates over anything else. Despite attributing the success of available treatments to pharmaceutical companies, participants argued that the industry actively chooses not to provide cures, intending to perpetuate profits. There was one, however, who held a different view. Jesmond

rejected this conspiracy theory and argued against the other participants, claiming that society must be grateful for the medications which are available for they have improved the lives of millions. Jesmond contested the idea that novel cures are intentionally disallowed from being developed.

> "There is no cure for most illnesses, for the simple reason that they have not found it yet. […] They have eradicated polio and other illnesses. To me it's obvious that they try to find cures, but it is not as easy as you think. I would never believe that they've found the cure to cancer and kept it hidden, for example." - **Jesmond**

This remark triggered an immediate negative reaction from the other participants, who all believe that Big Pharma holds back certain research, directing it towards outcomes which are more profitable.

> "I am convinced that research has found the cure for cancer, but they will never make it available because it wouldn't benefit them." – **Grace**

To gain further insight about the 'cure to cancer' conspiracy theory that emerged in the focus groups, the issue was raised during an interview with one of the research experts, who is involved in cancer research.

> "Cancer changes so quickly. […] It is really difficult to find something which works on it, especially since drug companies work for a single drug. […] Many people believe the conspiracy that the cure has been found and is being kept secret. It is not true that there is someone who is controlling everything, that there is a cure and is being suppressed." – **Research Expert 3**

The response of Research Expert 3 does put an end to the 'cure to cancer' conspiracy, however, to a certain extent, it also reinforces the argument made by Mario that cures are developed or not depending on profit. Here the research expert explains that since cancer is constantly evolving, drug companies have not yet created a cure because they are not able to target the various forms of cancer with a single drug, and thus the profitability of the drug would be low

as it would not target all types of cancer. At various instances, the research experts implied that they feel that the motivations of Big Pharma are not the same as their own; it is as though research experts consistently felt the need to disassociate themselves from the commercial entities. This could result from the need for research experts to prove to the public that they are not after profit, but are genuinely after providing a better tomorrow within the medical parameters. This can be interpreted as a means through which professionals seek to build and perpetuate relations of trust with potential participants.

> "I believe that in the context of medical research, the main benefactor is the industry, pharma. We, as researchers, do our best so that patients benefit as well, but that usually takes several years." – **Research Expert 1**

The focus group participants, both patients and non-patients, often clarified that they too disassociate the researchers and other health care workers or medical professionals from Big Pharma. They frequently conveyed the belief that these entities are to a certain extent independent as they share different motives, however, they still operate hand in hand, where the pharmaceutical industry holds power over the other parts. There was a clear agreement amongst the absolute majority of the participants from all groups that whilst doctors, researchers and other healthcare professionals are typically motivated by providing improved healthcare, Big Pharma is solely driven by capital and therefore has entirely distinct intentions. This conforms to the literature where it is often outlined that research participants are significantly more likely to perceive public researchers to be more trustworthy than commercial researchers (Tupasela and Snell, 2012).

> **Maria (author):** "So, all in all, do you trust the system?"
>
> **Mario**: "I trust doctors, but not the pharmaceutical companies, no 100%."
>
> **Lina**: "Me too. I believe that research is very important, but I don't trust the companies. Wherever money is involved, there cannot be much trust."

The title of this subsection implies a tension between the research participants and Big Pharma, furthermore, the discussion often refers to Big Pharma and the research network as two separate entities. Yet, for the sake of progress, it is a prerequisite that all of these stakeholders collaborate and form a research hub which links and connects the contributions of all parts. The literature (Goisauf et al, 2019) reveals that research professionals emphasise the importance of collaborating with the private sector, with the key aims being the attainment of scientific advancement and the improvement of social wellbeing. Also flagged in the literature is the importance that participants are informed of the possibility of commercial collaborations at the stage of recruitment, to ensure transparency (Goisauf et al, 2019).

Profitable participation?

> "If there is any possibility that industry may or will become involved, that should be part of the informed consent. One must add what happens with any money that comes out of this. So alright, yes, the data may be developed, and companies may make money off this, and you will get nothing, or you will get 1%. That information must be provided." – **Research Expert 2**

A debate on whether participants are to be paid for their contribution to drug development or not emerged in all focus group sessions and the dynamic of the conversation was similar in all groups. In each group some argued in favour and others against.

> **Elisabeth**: "It is true that they make a lot of money but you need to consider all the years of studying, and all the scientists they would have paid, and the money they would have paid for labs and equipment."
>
> **Mariella**: "So you are saying that it's ok for them to make money out of my free sample."
>
> **Jessica**: "Nobody does anything for free."
>
> **Mariella**: "Except for us." *laughs*

The reasons behind the binary of opinions depended on the way the participants perceived the donation of biospecimens. Whilst some argue that a sample donation is giving a part of oneself, and thus ownership remains of the donor, others perceive donation a self-less act and believe that once a sample is donated, it becomes a part of the research infrastructure. Luke perceives donation as a simple altruistic act which should provide the participants with no profit whatsoever, because the sample on its own is of no value, and requires the expertise of researchers and industry to be materialised into something useful.

> **Claire**: "If out of research they manage to make millions, I see nothing wrong with giving the participants a small share of the money."
>
> **Adriana**: "It's fair enough."
>
> **Luke**: "This does not make any sense. Ok, my blood or whatever is there, but my blood sample by itself is literally nothing. My sample together with the work of the researchers is valuable… on its own it isn't."
>
> **Claire**: "If the sample on its own means nothing, neither does the researcher without the samples."
>
> **Luke**: "But the researchers are actively involved… it's their job and it takes a lot of their time."
>
> **Adriana**: "They still remain dependent on each other."

This dialectic stems from a perceived power struggle between those participating in genomic research and those conducting the research or those developing the outcomes of research into a tangible product. Whilst some perceive participants as powerless, others believe that participants should hold all of the power, since research would not be a possibility without their contribution.

Alternative medicine: a means of regaining control

A way through which non-expert participants convey the regained control over their personal health is through rejecting traditional medications and turning to alternative medicine for treatment.

> "The pills they used to give me for migraines used to help me with the pain but would create other problems. When I discovered cannabis oil, I started to get migraines every 3 months, instead of every week. […] It's much more effective than the medications which the doctors prescribed." – **Robert**

As society becomes reflexive and starts to recognise that experts are fallible, they start to attain control, and turning down expert systems becomes inevitable. As non-professionals gain more knowledge, they increasingly develop the skills to challenge tradition and engage in alternative ways of life, to combat risks and create a more favourable present and future (Giddens, 1999; Elliot, 2002). However, it is interesting to note that research experts interpret this finding in an opposing manner. When asked to express their personal opinions about encountering individuals who choose alternative medicine over traditional medicine, Research Expert 2 claimed as follows:

> "Many people are falling for that gimmick, generally older people who are not very well informed about science and who are used to believing that what they hear on the news is a fact, so they struggle to be critical of what they read on social media. […] Science communication is extremely important." – **Research Expert 2**

The expert here interprets this novel way of treatment as a result of misinformation as opposed to resulting from better-informed non-professionals, attributing this finding to a lack of scientific communication which as a result leads to a less critical general public, which often believes what is said on the media. There is a clear antagonism in the interpretations of this finding. Engaging in alternative medication practices can either be seen as a naive act which

results from a lack of scientific knowledge, or else as a novel attitude of the "information-rich" who consciously chose to become increasingly critical of expert systems (Elliot, 2002). Giddens (1990) refers to individuals such as Mark, Robert, and others with similar perceptions, as "radical engagers", who aim to mitigate the weaknesses of traditional medicine by eradicating or overstepping them.

Nonetheless, none of these participants changed their attitude towards participating in genomic research, and they all remained consistently motivated throughout the focus group session. This suggests that the critical interpretation of several participants towards specific matters did not impact their willingness to participate, and hence, irrespective of the way they perceive traditional medicine, they are all willing to contribute to its further development. This finding reveals the power of the medical system, traditional medicine and Big Pharma. Irrespective of any concerns towards pharmaceuticals, and any desire to engage in natural therapeutics, the essence of everything boils down to the final decision, which reveals that all participants in this research ultimately feel that society at large cannot do without traditional medicine.

My data also revealed antagonism between the opinions of patients and non-patients towards this issue. The distinction in attitudes can probably be attributed to the level of threat experienced due to illness. Patient focus group participants who are experiencing life-threatening conditions, expressed a significantly higher level of trust towards traditional medications over alternative medicine, as opposed to their non-patient counterparts.

5.4.2 **Incidental findings: a deal breaker?**

The expert interviews revealed that the likelihood of encountering incidental discoveries is relatively high when it comes to genomic research. Despite agreeing on

this fact, expert opinions varied in terms of whether such findings are to be returned or not. The two contrasting views outlined by the interviewed research experts are the following:

> "When we do whole exome and come across a risk for a particular illness, we would go back to the patient, yes. Then we refer them to the appropriate healthcare physician." – **Research Expert 1**

> "The way we deal with it is that we do not report back incidental findings." – **Research Expert 2**

Research Expert 1 insisted that reporting secondary information is imperative if researchers are to convey the message that they genuinely care for the wellbeing of patients. The expert explained how genomic research can reveal information which can considerably impact the lives of participants, and since their sample and personal data remain their property, they should hold the right to access such information.

> "When we do whole exome sequencing, we sequence every, or almost every, gene. So we can have access directly to information about paternity, the risk for cancer, risk for mental problems, and many other illnesses. The participant wouldn't have consented for us to look at that, but sometimes we have no option but to see such information. UREC[22] obliges us to look at the genes which are relevant for our project, but the rest of the data is there, and it is extremely valuable to us as researchers, and also to the patients." – **Research Expert 1**

Nonetheless, Research Expert 1 clarified that not all findings are necessarily equally valuable; whilst some variants only indicate the possibility of secondary illness, others provide more accurate information, and thus, the expert was particularly adamant about returning the latter form of results.

[22] University Research Ethics Committee

> "Sometimes we find VUS, variants of uncertain significance, and international guidelines such as the rules of ACMG[23] say that we are not obligated to inform the patients about them since they should be interpreted with caution. […] Other variants are very specific, for example, if you find BRCA1 and BRCA2 which are highly penetrant breast cancer genes then you should counsel the patient accordingly. It would be highly unethical not to." – **Research Expert 1**

Other research experts made similar claims about the possibility of inaccuracy or lack of clarity with certain incidental findings:

> "For instance, there was this research group from Israel and they tracked a group of patients with cardiopathies, so these people could drop down dead essentially. All the people who had a variant were told it was pathogenic, and what they needed to do. Then suddenly they found out that it was benign, and they had to call them again to say it's nothing. […] Findings are not static in genomics, because we are still in the phase of discovery. You can create confusion." – **Research Expert 2**

Historically, the reconsideration of scientific facts led society to start developing a dubious perspective towards science, consequently lowering the levels of trust of the public towards experts (Giddens, 1999). Therefore, it is legitimate that experts fear providing inaccurate information, and might choose to be cautious and not report back any findings, to avoid damaging their reputation which can, in turn, minimize trust levels. Nonetheless, it must be pointed out, that some of the research experts claimed that they would not commit to providing participants with information about incidental findings, even when the data is highly likely to be accurate, due to reasons of lack of expertise for follow-up genetic counselling and resources. Research Expert 2 even claimed that at times they choose to completely anonymise the data to provide an added layer of protection to the participants, despite knowing that such practice cuts

[23] American College of Medical Genetics

the link between the sample and the personal data, and thus excludes the possibility of returning any findings.

> "If your expertise is on disease A, you can keep the participants up to date on findings about disease A, […] but what do you do about the thousands of other diseases that there are. That's what incidental findings are... when you find information which is relevant to a different disease, a disease which you are not an expert on. How do you report on that? […] You cannot do that without lots of personnel and resources, as well as the necessary expertise. If my expertise is on these five diseases, I cannot go tell the patients that I found a variant which is related to a disease outside of those five, and telling them to do something about it, because I am not expert on that disease." – **Research Expert 2**

Similarly, Research Expert 3 underlined that importance of research experts being precautious in reporting back incidental findings:

> "First we have to be aware of the resources that there are. If you tell someone that they have the gene for [a particular condition] without providing the necessary support, it can turn bad. All they did was give a sample for research, and their life would turn upside down overnight." – **Research Expert 3**

According to Research Experts 2 and 3, best practice calls for being responsible and cautious with incidental findings, even when findings appear to be accurate and significant, whereas, Research Expert 1 believes in passing on the responsibility to research participants and allowing them to make an autonomous decision about whether they would be prepared for such information or not.

> "We ask the patients whether they would want to know about any incidental findings which we might come across and which could be directly relevant to them. If they say no, we don't inform them, but if they say yes, we do. […] The departure point must be the well-being of the patient, and not whether we have enough resources or not." – **Research Expert 1**

In contrast, it is often argued that making a truly rational and informed choice about receiving such information requires participants to have a wide knowledge of genomics (Wright, 2013). Research Expert 2 contests the idea of simply accepting the decision of participants at face-value.

> "There is the issue of added burden. A person can come to the study because they have a problem with their kidneys, and we go back to them saying they have the variant for Huntington's disease, or breast cancer, or anything else. […] You cannot just trust that they say that they would want to know, we must be sure that they have the right support. I don't think it's fully responsible otherwise." – **Research Expert 2**

The literature refers to antagonism between two perspectives with regards to the matter of incidental findings. Case in point, the US Presidential Commission for the study of bioethics (2013) has declared that participants should be informed about the possibility of incidental findings in advance, and should be allowed to make their own decisions. Such a claim was contested by those who argue that participants might not always be capable of making an informed decision, as they often lack the required knowledge (Wright, 2013). Yet, the same Commission (2013) also denoted that in the case of findings which disallow for the possibility for preventive action, the returning of such findings may be considered irresponsible, and here the commission was contested was ones again, but this time by those on the other end of the opinion spectrum, who argue that researchers should not have the power to presume about which findings might have a medical benefit, and which do not (Powledge, 2015).

The opposing views amongst researchers are not necessarily troublesome, as the research experts who participated in this research all agreed that they deal with this dilemma by being thorough and transparent in their respective consent forms, and ensuring that all participants are well informed about the incidental finding practices of the particular research project. However, a complex power struggle comes into play between researchers and research

participants. In totality, the majority of the focus group participants, patients or not, agreed on an ideal which presumes that genomic research participants should always have the option to access information about incidental findings. With the exception of one non-patient participant, all focus group participants shared this common belief.

> "It is really unethical [if they don't]. They're not supposed to just take and not give." – **Mariella**

> "We participate for the good of society in general, but also for our personal satisfaction. The outcome of research should be on two levels, on a global level, for everyone to benefit, and on a personal level. If they don't give us such information then we really are not going to gain anything from this. […] I shouldn't even have to say it… of course, they should inform us about such findings." – **Frank**

> "This is the least they can do to reimburse us as participants." – **Jesmond**

Possessing adequate knowledge is crucial for participants to make an informed decision on the matter (Wright, 2013). The discussion consisted of very clear opinions by the focus group participants, as they perceived the issue at face-value and typically failed to consider the complexity of secondary genetic findings.

Irrespective of whether participants possess enough knowledge or not, and whether they understand the whole picture or not, these findings are extremely relevant as the perspective of potential participants regarding incidental findings can have a significant impact on their willingness to participate. In fact, the discussion on incidental findings led to a peak in demotivation levels, as participants clearly expressed disappointment towards the possibility of researchers keeping any secondary findings from participants. In the literature, it is revealed that a disagreement on incidental finding protocols is often declared to be the main factor in determining unwillingness to participate (Ahram et al, 2014). Focus group participants who

had previously claimed full trust towards professionals, suddenly started to express doubts, claiming that researchers who fail to pass on such information are contradictory in their actions.

> "If they truly are doing research for the sake of helping others, then it does not make sense to have such information and keep it from the patient… it defeats the purpose." – **Elisabeth**

> "Unfortunately some people use you. I mean I know the ultimate aim is to get more research on a disease to find a cure to help people but practically the person doing the research is trying to get a PhD or an advancement at work. Really we're just objects that they are using. At least they could give something back to the participant." – **Mariella**

The message is clear in my data. Participants expect to be aware, and if they are not provided with such an option, they automatically start to question the trustworthiness and the real ambition behind the work of researchers. The majority of participants perceive researchers as the only hope for improved healthcare, and patients, in particular, look at the research community as their only gateway for a possible breakthrough which can have a direct impact of their lives. Consequently, they fail to understand how researchers can hold data which might directly impact the lives of individuals. Participants agreed that such practice is unacceptable, and as a result, some participants started to seriously question the intentions of research authorities and started developing doubts about participation. Moreover, Luke, from one of the non-patient focus groups, raised a completely opposing perspective on the matter.

> "This happens with all research projects. If I take part in research by filling up a survey questionnaire and I write the wrong answers, they won't get back to me to inform me that I got them wrong, because I would have participated for the sake of research and not for myself. The aim is not to learn more about yourself." – **Luke**

Luke's interpretation is that individuals must distinguish between personal health care and participation in research; whilst the first is about personal well-being, the second is completely selfless and must be looked at as a contribution which goes beyond any personal gain. The

discussion on incidental findings is revealing on the issue of altruism. Although participants claimed that their motivation to participate primarily derives from a sense of altruism, when faced with real-life scenarios they typically claimed that they do expect some form of return. They do not base their decision to participate on the possibility of personal gain, however, they expect to receive something back when the circumstances provide the possibility. Furthermore, the research experts interviewed who declared that they do not report such findings, also claimed that they do so responsibly and for the sake of participants' wellbeing. As underlined by Research Expert 2:

> "You cannot just trust that they say that they would want to know, we must be sure that they have the right support. I don't think it's fully responsible otherwise." – **Research Expert 2**

This intriguing remark was perfectly reflected in the narration of one participant who recounted his personal experience with receiving information regarding an incidental finding which emerged during clinical testing. Although he generally felt that receiving incidental finding information was a positive thing, when he experienced it, he realised that one does not immediately consider the implications that such news can have on mental wellbeing. Peter narrated his personal experience as follows:

> "Once I fainted for a long time and ended up in the hospital, and one of the tests they did was an MRI, and they accidentally discovered a cyst in my brain. It's benign, but since they discovered it my life has changed because I constantly fear that something might change and it starts to grow. Since I can do nothing about it, I think I would have preferred not to know about it. […] If this discussion took place a few years back, I would have definitely said that I would want to know, but now I must say that I am not sure." - **Peter**

Despite, a general agreement amongst the vast majority of participants about the importance of returning incidental findings, a good number of participants from the

non-patient groups claimed that although they believe that they should have the option to decide, they would not opt for receiving such information.

> "If it is incurable I wouldn't want to know, because it would damage me psychologically. If there is a cure, I would consider it." – **Adriana**

> "I wouldn't want to receive such information. If it would impact my lifestyle, then no. I'd rather live day by day." – **Marica**

Patients were more likely to say that they would want to know, when compared to non-patients, irrespective of what the condition might be, and whether it is curable or treatable, or not. Since these individuals are already living the experience of illness, they are better equipped to look beyond the face value of the situation, and thus managed to think of specific reasons why they would want to have access to such information, under any circumstance. One of the key reasons being future decisions, such as family planning.

> "Any information can be useful, even if it won't directly affect my health. It's up to us to see whether the information is useful or not, we should be the ones to decide. Maybe if I knew that my genes could have led to my son's condition, I might have decided to adopt instead of conceiving a child." – **Stefania**

This issue of incidental findings triggered intense emotions amongst the participants. Whilst some expressed fear of the risk of learning about potential future uphill battles, others conveyed anger at the possibility of research professionals not returning such findings. The general agreement was that the participants should be making the decision and that researchers should always provide the option of returning secondary findings. Participants generally expressed negative perceptions towards researchers who fail to provide the possibility for returning such findings, claiming that these experts would declare that their true intentions are inspired from an egotistical desire to succeed and not from a motivation to provide better healthcare experiences for patients.

The majority of the focus group participants perceive the issue of research on a micro-level. They might recognise that participation in genomic research and biobanking is not a personal venture, but at the same time they expect to remain involved and have a certain level of control, and consequently, they expect researchers to look out for them and be in communication with them. In contrast, the research experts typically perceive research within a macro context, and whilst they all ensure that they follow ethical and legal protocols, not all experts feel obliged to report incidental findings which may crop up. The antagonism in perspectives, between researchers and participants, and even amongst researchers, classifies the issue of incidental findings as one of the key subjects of debate within the field of genomics and biobanking (Vaught and Lockhart, 2012).

5.4.3 Consenting, ethics and law: safeguard or hindrance?

Attitudes towards ethical and legal standards in genomic research and biobanking vary; some participants perceive them as a setback for medical progress, whereas others consider them to be protective tools against potential hazards. Following ethical protocols such as informed consent not only grants research participants more power and control but also serves as protection for research bodies. As outlined by Research Expert 2:

> "There is the issue of protection of the scientists, for example with the issue of commercialisation or incidental findings. There have been problems. For instance, in Germany, a group of researchers were looking at brain scans for something in particular and there was a patient who had some anomaly in the brain scan, in another area, not in the area that they were looking at, and they didn't notice it because their focus was on a particular area. That person actually took them to court saying you 'should have been on the lookout for me and noticed this and warned me about it.'" – **Research Expert 2**

This chimes with the findings of the study conducted by Goisauf et al (2019) which revealed that the vast majority of research experts seem to prefer the ideal of transparent informed consent and therefore adopt adequate practices because they consider them to be valuable, and not because they are obliged by the law. This can be attributed to the idea raised by Expert 2 that transparent consenting aims at thoroughly informing participants about the process of research, and specific issues related to research, whilst providing research professionals with peace of mind. Furthermore, informed consent disallows researchers from using samples and data autonomously, thus, granting an adequate level of protection for participants.

> "Informed consent is important because, whichever way you look at it, the sample is part of a person. You need permission, and permission isn't vague. If you go and ask somebody to borrow their car to drive to Mellieħa and they say yes, they said yes for you to use their car to drive to Mellieħa not to borrow their car and on the way to Mellieħa rob two shops. You're doing something with people's sample so you must inform them exactly of what you will do with that sample." – **Research Expert 2**

Research Expert 2 constantly emphasised that the sample remains the property of the donor, hence, accentuated the importance of consent forms not being vague, as participants reserve the right to be fully aware of who gets access to the samples and data, for what reason, where, why and how. The General Data Protection Regulation (GDPR), not only expects researchers to provide all the necessary information but furthermore to do so clearly and unambiguously (Bovenberg et al, 2017).

Despite the efforts of ethical committees and regulation bodies at establishing a framework for best practice research, reality often reflects mishaps in the system. Focus group participants recounted experiences which reveal that the ideal and the real are not always at par. It appears that sometimes experts may follow legal guidelines, but ignore the ethical standards which are

expected for best practice consenting procedures. Michaela's recollection of her personal experience of consenting is a clear example:

> "There was something which bothered me when I did the operation to remove my breasts. As I was about to go in for the operation a nurse asked me if I was interested in giving my breasts for research… she asked me to sign a consent form. I wasn't informed about it. I immediately said yes, because I know that research is important. […] I think that it is something that should have been discussed before and not on the day of the operation. It's better if they give you some time to sleep on it instead of making a decision there and then." – **Michaela**

This is a scenario where the distinction between research and clinical practice becomes obscure and the two are diffused into one, thus creating ethical concerns (Carrieri et al, 2015). Although interpersonal relations between researchers and participants are not always necessary, sometimes they are inevitable, particularly when researchers are also clinicians.

> "I know the participants, and they know me. Most times I would know their whole family, including their siblings and everyone. In fact, they often email me to ask about the progress of research." **– Research Expert 1**

Professionals who occupy a dual role are expected to be extra cautious to avoid influencing the decisions of potential participants and must allow them to make an independent choice, free from coercion (Carrieri et al, 2015). Those invited to participate in research must be provided with an environment which allows them to feel emancipated and free to give a negative response. This can be achieved first and foremost if researchers and clinicians ensure that the participants recognise the distinction between clinical practice and research (Carrieri et al, 2015). Consenting procedures which happen at inappropriate moments are a means through which researchers can take advantage of the situation to attain a goal without opposition. This is an abuse of power, which immediately places potential participants at a disadvantaged position.

> "You get the feeling that you must say yes. You cannot go against them at that moment." – **Mariella**

When recruiting participants who also happen to be patients, the chances for abuse might be higher. The recruitment phase for this dissertation was a revelation in itself, as the excessive enthusiasm shown by patients and the parents of patients to attend for the focus group sessions was incomparable to that of non-patients. In fact, the recruitment for the patients' focus groups lasted a few days, whereas the recruitment for the non-patient groups took over a month. The excessive enthusiasm of patients makes them more susceptible to be "pragmatic acceptors" (Giddens, 1990), who tolerate more and accept without much deliberation, as they consistently hope for more research directed at their condition or illness. The vulnerability and helplessness of patients are not to be used as weapons against them (Boat and Field, 2011), as best practices call for researchers to recruit in a manner which is free from coercion, to adequately inform potential participants, and to prioritise the best interests of the individual, whilst aiming towards the best interest of society (Laurie et al, 2010; Carrieri, 2015; Bovenberg et al, 2017).

Data Sharing

> "It is important to inform participants that their data can be shared in an aggregate form with international networks. This is particularly important with rare diseases as sometimes there will be only one patient in any one country. Minimum information sharing is important for these cases." – **Research Expert 4**

The issue of data sharing is often discussed through the lens of privacy. When discussing the issue with experts, they particularly directed their narrative towards the importance of being transparent about data sharing, and of safeguarding participants from privacy risk. Data sharing increases the exposure of personal sensitive data, as more individuals gain access (Laurie et al,

2010), hence why experts immediately declared the importance of providing sufficient information about the possibility of data sharing.

> "We need to be specific about the people who might get access to the data. Are they researchers, academics, scientists, hospital?" – **Research Expert 2**

Overall, focus group participants conveyed a distinctive attitude towards data sharing practices. Interestingly, the concern about risk did not increase with the introduction of the possibility of data sharing, but rather the participants focused their attention on the collaborative nature of data sharing which increases the chance for breakthroughs.

Irrespective of whether they were previously troubled about privacy risk or not, all participants kept their level of concern constant and did not feel that data sharing escalates the possibility for privacy hazards. Participants with contrasting views about privacy risk all agreed that data sharing should be encouraged, and furthermore, they communicated doubts about research bodies that fail to share data.

> "Researchers who refuse to do this are limiting my sample from reaching its full potential."– **Marica**

As with other issues, such as when dealing with incidental findings, once again the participants felt the need to reclaim a certain level of authority and control. Yet again, they started to question the intentions of the research community, arguing that those who prevent collaboration are motivated by personal attainment, as opposed to the greater good. Some scholars argue that not only is data sharing considered to be ethical, when done in compliance with standards of best practice, but it might, in fact, be unethical for researchers to refuse to share data, as resistance can be interpreted as a way of going against the commitment of aiming to attain the best outcomes for the common good (Laurie et al, 2010).

Participants, like Therese, who had previously expressed full trust and a quasi-religious attitude towards the research community, suddenly felt the need to challenge the researchers, and claim greater control. She swiftly started to reconsider her willingness to participate in research, admitting that the issue of data sharing is a deal-breaker for her, as she would refuse to contribute to researchers who inhibit collaboration.

> "I wouldn't give the sample to a researcher who is obviously selfish. I know that these things happen all the time, and that is the risk that I would be taking when donating the sample, but if I get to know beforehand I would have greater control and would choose not to participate." – **Therese**

It has been established in this research that patients generally desire to be part of a community, and that collaborating to genomic research helps in partially satisfying this need. Therefore, they seem to feel frustrated at encountering scenarios where individualism reigns over collectivism. Biobanking largely depends on collaboration and solidarity amongst research experts, research participants, patients and the general public. Data is nowadays being shared beyond a national level, as best practice biomedical research calls for compliant data sharing practices aimed at achieving optimum results for the greater good (Laurie et al, 2010). Nonetheless, a few participants seemed to accept individualism from researchers and argued that even if it seems to be unfair, such behaviour is reasonable.

> "We have to be realistic… there is competition in everything." – **Jessica**

Furthermore, most participants felt uneasy about the situation and insisted that researchers who are motivated by personal achievement defeat the purpose. Since participants are expected to contribute to research for the greater good, and with an altruistic motive, the same participants tend do expect researchers to share the same ambition.

> "I wouldn't give the sample to someone who refuses to share the data. I don't mind about breaches and the other stuff, but this issue bothers me because I believe it is selfish." – **Luke**

The literature shows that only through collaboration can data be exploited in the most effective way (Robinson et al, 2013) and therefore it is legitimate for participants to have this attitude, since their ultimate goal is that of attaining the best outcomes, in the shortest time possible. A few participants claimed that they would be unwilling to donate biospecimens to researchers who claim that they would not share data, however, the majority would choose to give the sample either way, even if they feel that the researcher is not incorporating best practices.

> "I believe that such researchers would be working for their personal achievement only. I think they should share data, because if research grows, the chances for a breakthrough would be greater. […] But still, if I get a chance to participate and the researcher has this condition, I would not miss it for sure… I would still participate." – **Elisabeth**

Despite the ever-increasing sense of individualism in modern societies (Beck, 1992), individuals seek for circumstances where they feel part of a community and where they share goals collectively (Davey, 2019). The belief that one shares similar goals with others creates social connections, however, sharing the same goal does not necessarily mean having the same intentions towards attaining that goal (Tuomela, 2004). While patients and experts might share the same goal of medical breakthroughs, the intentions behind the need for that attainment differ. Patients hope for a breakthrough for their personal health, or that of those who shall be diagnosed after them, whereas, experts perceive the attainment of the same goal as a professional accomplishment which in turn provides better health opportunities.

Professionals' resistance to sharing data might result from a power struggle amongst experts. Whilst conducting this research, a sense of mistrust between experts was felt. Case in point, one research expert refused to be audio-recorded and requested to answer the interview questions via email. This in itself reveals a sense of insecurity in collaborating with other researchers. Furthermore, one of the research experts declared this bluntly:

> "I don't believe that the samples are personally mine, but the biobank is basically a framework for sample sharing… [this leads to] a sense of loss of ownership and loss of control of the samples." – **Research Expert 1**

Researchers feel the need to have control over their data. Ultimately the samples are the tools which make their work possible, and therefore fearing the loss of control is expected. At one point, Research Expert 1 declared that for one of the studies they are not using the biobank and decided to store the samples in their own freezers. Here this researcher explains what the impact of a loss of ownership can result in.

> "The biggest challenge that we face in genetics is getting access to the samples, therefore giving the samples to someone else is not always easy. To be honest, whenever we used the biobank we did have a problem with getting the samples back, it was a bureaucratic process, and I had spent four to five years of my life working hard to get those samples." – **Research Expert 1**

The personal experience of the expert sheds some light on the issue of resistance towards data sharing. The expert here points out that opting against the sharing of data is not necessarily the result of greed, but it can result from a fear of inhibiting the research process due to issues of red tape. The same expert has declared that they do engage in data sharing, but particularly under circumstances which would not impact the progress of their research.

> "We share samples with other researchers all the time, mostly with students. Most theses are done using the sample which we collect. We also collaborate with a foreign centre, we don't share personal data, and we only share coded data." – **Research Expert 1**

Once again the level of genomic knowledge of research participants impacts the way they perceive specific issues and ultimately impacts their motivation and willingness to participate in research. Looking at the issue of data sharing from the outside does not expose potential participants to the real hurdles which researchers might encounter. A lack of clarity about what the practice entails has resulted in participants doubting the intentions of members of the

research community, based on their data sharing preferences. This is why the consenting process must involve an in-depth discussion between researchers and potential participants, where the latter would feel at ease about trashing any concerns which they might have. Dynamic consent portals were designed with these issues in mind – to facilitate communication between stakeholders after consenting, which in turn allows participants to clarify matters, and perchance even alter consenting preferences as desired (Kaye et al, 2014).

Dynamic Consent and Participant Control

> "I might have an issue with the unknown but I would want to have specific information about how my sample will be used." – **Adriana**

The opinion of focus group participants with regards to the different types of consent varied. One of the key arguments with regards to broad consent was that participants should be provided with information about potential ways in which the sample might be used, and where the researchers would be obliged not to utilise the data beyond those parameters. In contrast, others stated that the requirement for professionals to consistently inform participants might hold research back, and thus argued that researchers should be free to utilise the sample as they deem necessary.

Dynamic consent allows for continual communication between researchers and researched, in the interest of both parts (Mester et al, 2015). Due to rapid technological advancements, one-time broad consenting procedures are insufficient for providing adequate information, because researchers are not capable of predicting where innovation will take research (Robinson et al, 2013; Teare et al, 2015). A dynamic consent interface tackles this limitation as it creates a hub where stakeholders remain in communication, and where participants are given the opportunity to re-evaluate their consent preferences (Kaye et al, 2014).

Biobank based research at the University of Malta will shortly have the means to provide research participants with greater autonomy and control over their samples and data. The development of a dynamic consent portal named 'Dwarna'[24] shall empower participants and provide them with greater control over their personal data. The portal shall facilitate the consenting process and will allow for participants to withdraw their consent at any moment through the online tool, thus guaranteeing greater autonomy and freedom (Mamo et al, 2019). When introduced to this innovative form of consenting, some participants claimed that such active involvement can be inconvenient and insisted that researchers should opt for broad consent which would allow for greater flexibility amongst the research community.

> "It's still an inconvenience. Isn't this for medicine after all? I might not trust the process blindly, but whoever wants to do bad would still do it, and I think that that from such research we can truly see positive results." – **Mario**

However, Mario, and others with a similar perspective, were immediately challenged by fellow participants who desire to have greater control over the use of their sample and who believe that broad consent should be an option but definitely not the rule.

> **Mark**: "No way! If I gave my sample for research on a type of cancer it should only be used for that and thrown away if the study is over, or else they should ask me for permission should they decide that they need it for something else."
>
> **Mario**: "That's why we always remain a hundred years behind!"
>
> **Mark**: "But that is my blood, how can someone use it in a way which I am not informed about? No way!"

Although dynamic consent empowers research participants, by giving them increased control over their consenting preferences, some participants might perceive it as burdensome, meaning

[24] https://dwarna.mt/

that they would rather have limited power and attribute full control to the researchers. The majority of the participants in this research shared Mario's perspective in this regard and commonly believed that the researchers should have a certain level of freedom, to facilitate the research process and create better chances for a breakthrough.

> "I trust that it will be used in a good way and therefore I would prefer to allow for the researchers to use the samples as they want." – **Jesmond**

Despite the lenient perceptions of several participants towards consenting processes, the GDPR disallows research institutions from claiming absolute power and control over samples and data, and therefore dynamic consent offers an effective way to ensure compliant consenting practices, whilst effectively facilitating the research process. Nonetheless, it is also crucial that participants are provided with the option to opt-out of dynamic consent, as they ultimately hold the liberty to not be actively involved in research (Kaye et al, 2014; Teare et al, 2015).

> "People should still have the option to give consent to a list of projects instead of giving consent to each specific one. The option for broad consent should remain for those who would get annoyed with such notifications." – **Claire**

Patient focus group participants were also highly likely to argue in favour of giving researchers the power to use samples and personal data as required, however, they also perceived dynamic consent as a means of empowerment, through which they would be able to keep track of their involvement in research whilst acquiring feedback from researchers.

> "A portal of the sort would be taken very positively… I'm sure. It's a sort of empowerment for them." - **Support Group Representative 1**

Legal and Ethical Standards: Are they enough or are they too much?

The findings which emerged through the discussion on ethics and law were somewhat surprising, and at first glance, seemed to be inconsistent with what was revealed in the previous sections. Individuals who had previously claimed to be concerned about breaches of data, and who had expressed fears related to the donation of biospecimen, were suddenly vouching for tolerating practices that encourage abuse, such as the liberty for researchers to utilise the acquired samples without restriction.

> "I have feeling that this thing involves a lot of ethics and stuff. All I know is that because of ethics and political correctness, we have wasted 70 years when it comes to medicine. […] That's why we never move forward in Europe. If there are a thousand ongoing studies they would probably expect me to give consent a thousand times." – **Mario**

Mario, and other participants, had declared a lack of confidence in the trustworthiness of the medical system. They did claim they would accept a request to participate in genomic research, however, they also claimed not to be highly confident in terms of risk and privacy safeguards.

> "I do worry about having my blood sample stored. […] You cannot say that there are no risks." - **Mario**

Yet, when discussing ethics and law, Mario, and others, expressed a desire for ethical and legal standards not to serve as hindrances to medical progress and declared the need for medical professionals to be allowed to work more freely.

> "When you give a gift… do you instruct the person receiving the gift of how and when they should use it? No! You simply give it and allow for them to use it as they please." – **Lina**

> "We no longer even experiment on mice... too many regulations and ethics are delaying progress I believe." – **Grace**

The contradiction which is observed here is that participants who had previously raised concerns about the misuse of samples suddenly started vouching for lenient regulations for the

sake of progress. Various participants had argued that they do not necessarily feel protected by the regulations in place, claiming that laws are frequently violated. The discussion often referred to a culture of impunity for authority figures, which in turn makes the people feel less secure and protected. Yet, all of a sudden they expressed drastically lower levels of concern with regards to consenting procedures, and preferred to pass on the power to the researchers.

This pattern was observed amongst a number of participants. Initially, this seems to be contradictory, however, it emerges that participants who had flagged various concerns, often claimed that they would not allow their fears to stop them from contributing, because ultimately they believe that medical progress is a necessity which cannot happen without taking risks. Several participants, such as Mario and Lina, also argued that illegitimate researchers who intend to misuse samples and data would not ask for consent to do so, and thus, consenting procedures are insufficient at preventing abuse. Thus, it can be concluded that the participants are not necessarily being inconsistent, but rather they feel powerless in controlling abuse, and therefore they perceive ethical and legal standards as means of giving the impression that participants hold some form of power and control, when in reality they do not. Furthermore, they also believe that the contribution of participating is indispensable for progress and therefore they subconsciously make a privacy versus data utility trade-off which results in a pro-research attitude.

> "I personally believe that participants do not understand the implications of what they sign up for. Most times they barely care about what's on the consent form. They just sign. We are obliged to do consent forms which are unambiguous, and we do try to explain, for instance, that they won't be getting paid, about incidental findings etcetera, but they are not always bothered." – **Research Expert 1**

This expert comment further reinforces the argument that the absolute majority of participants do not recognise the significance of the consenting process. Separate studies show that a

significant number of participants commonly struggle to respond to questions about the studies they enrolled to, so much so that one particular study revealed that 62% of the respondents were not aware that their sample was being held at a biobank, despite previously giving consent (Robinson et al, 2013).

Moreover, the opinion of Mario, Lina and others, is not representative of the opinions of all the participants in all focus groups but rather represents a specific group of individuals. This perspective was particularly common amongst participants within the first non-patient focus group, which was composed of individuals with a secondary or post-secondary level of education. The focus group discussions overall revealed a dichotomy in opinions regarding ethics and law, such as the GDPR. Some participants perceived laws and high ethical standards as a hindrance, as previously discussed, whereas others argued that laws provide them with a sense of security, especially laws such as the GDPR which provide supervision over professionals on a European level, as opposed to local laws and regulations.

Interestingly, the perception that strict legal and ethical controls in research can hinder progress, was also pointed out by one professional.

> "The GDPR has caused us added stress. Students are wasting a lot of their time getting ethics clearance." – **Research Expert 3**

The expert also argued that ethical standards have truly made the achievement of medical breakthroughs tougher.

> "Nowadays, even though there are good intentions, the idea of ethics has become too much. I always say that the person who has saved the most lives around the world is the person who did the most unethical experiment in the world. […] He had observed that [cow milkers] were not getting ill from smallpox… they had a pox on their hands, which is a virus, called vaccinia, which they were getting from the cows. He injected the cowpox into a young boy and then injected smallpox as well, and the boy never got ill from the

> second virus. That is how vaccines were invented. He saved billions of lives around the world. That's why nowadays, it is much more difficult to put a new drug on the market. Nowadays you need two billion euros to get that done." – **Research Expert 3**

Although the expert recognises that there would be chaos if regulations were not in place, it was acknowledged that ethical and legal considerations do hinder progress. Other experts were keener on discussing the benefits which emerge through ethical procedures, and this implies that the dichotomy of ethics and law as a safeguard versus and a hindrance exists amongst professionals and the general public alike.

> "The GDPR has improved the transparency of what data about participants is collected." – **Research Expert 4**

Although ethical and legal restrictions might impact advancements, they ultimately allow for greater transparency, which in turn leads to higher levels of trust. If ethical standards were to be more lenient, the possibility for public disappointment would increase, leading to a decrease in trust. Since genomic research and biobanking are entirely dependent on the donation of human biospecimens, damaging the trust of the public can be catastrophic (Vaught and Lockhart, 2012).

This research has revealed that ultimately even those participants who desire optimum medical progress and therefore would grant researchers as much freedom as possible, still do expect to have some control. This can be concluded since when such participants were asked about specific issues, such as data sharing, commercialisation and incidental findings, they generally preferred to have control in determining how their samples are to be used. Therefore, they do prefer to be better informed and more aware. Literature shows that sometimes participants might not be too keen about ethical considerations, however, in cases where they eventually

learn that their personal data was processed without their knowledge, they tend to feel uneasy and develop a sense of mistrust towards the system (Vaught and Lockhart, 2012).

> "If they had asked me, I probably would have consented. The fact that it was a secret program made me suspicious […] there's no way I would consent now" (Vaught and Lockhart, 2012, p.7).

Ethics and law may restrict research to a certain extent, however, in the longer term, they allow for research to progress sustainably, by attributing some control to all stakeholders, and minimising the risk for abuse. Ultimately, best practices allow for the protection of research participants, in ways which, as much as possible, do not hinder the research process (Laurie et al, 2010).

6. Conclusion

This research was aimed at generating broad quantitative data about the level of biobank awareness within the local context, as well as deep qualitative data about the perceptions and attitudes of the key stakeholders (patients, the parents of patients, the general public, researchers, the biobank manager and representatives of a patient support group) towards the benefits and risks associated with genomic research and biobanking.

The quantitative data that emerged from this research expose the fact that an absolute majority of the local population does not know what a 'biobank' is, with a mere 4.9% (19 participants) of the sample mentioning the process of storing biospecimens for the purpose of biomedical research at some point during the survey, although only 2.3% (9 participants) knew that the premises which serves this purpose is called a 'biobank'.

The suffix 'bank' had a major influence on the interpretation of the term, as the most common response was associated with finance. There were also a good number of other creative guesses which are completely unrelated to biobanking or the medical system, ranging from a power bank to charge mobile phones, to the brand of an energy drink, to internet banking – these are evidence of how far outside the public imaginary the genomic research that takes place within the biobanking process lies.

These findings which emerged from the preliminary quantitative research informed the development of research instruments used to generate rich and in-depth qualitative data. Audio-visual tools were produced which were then used to provide an overview of the process, and the rights and implications of participating in genomic research, in order to set the scene during the focus group sessions with patients, parents of patients, and members of the general public.

The qualitative data gathered reveal disparities between the patient[25] and the non-patient focus groups. Patient-participants generally expressed enthusiasm as opposed to mere motivation, as they consistently emphasised their eagerness to participate in biomedical research, claiming that having their sample stored at the biobanking instilled hope and a sense of belonging. Non-patient focus group participants questioned privacy, and elaborated on privacy risks such as the breach of personal data, whereas patient focus group participants were more commonly concerned about the risk of research not reaching its full potential, either due to a lack of investment, stringent legal and ethical procedures or due to researchers who might prioritise personal success over the greater good, such as through choosing not to share data with fellow researchers. Patients constantly gave priority to the corporeal over the incorporeal, as they discounted possible non-physical repercussions from limiting their motivation towards the ultimate aim of achieving improved health.

This, however, was not always the case, as one patient who participated in this research was highly concerned about the storage of biospecimens and associated personal data at a biobank, claiming mistrust towards the medical system. Nonetheless, as a general rule, this research reveals a form of 'stratified reflexivity' (Ward and Coates, 2006) on the basis of health status. Participants consistently made risk-benefit trade-offs, and in the case of patients, a quasi-religious attitude towards research experts was observed, as they perceived researchers as the key agents who are active towards attaining the goal of enhancing medical knowledge and thus improving their chances for breakthroughs. Even if at face value, patients focus group participants declared that their motivation lies in altruism, when challenged about other issues,

[25] Includes both patients and parents of patients.

such as the issue of trust towards authorities, they sometimes indicated that hope for personal gain persists, as they declared that their optimistic attitude does not necessarily stem from absolute trust but rather from their willingness to try anything in order to get better soon. The distinctive way in which patients and non-patients perceive the outcomes of research and biobanking shows that whilst for non-patients participation is about self-fulfilment and the greater good, in the case of patients, the eagerness to participate, derives from a more basic need for optimising their health or that of their kin.

The general feeling of mistrust expressed by various non-patient participants, and by a few of the patient-participants, goes beyond their experiences within the health system, as their mistrust towards the health system and those who make it up appears to be attributed to a general sense of mistrust towards authority figures. It was established that social actors do not solely develop attitudes and perspectives towards a system based on what they experience within that system, but also through their interpretations of the broader system, which attitudes are then reflected in social action within specific systems and processes (Luhmann, 1976; Ward and Coates, 2006), such as the donation of biospecimens for research. Reflexivity entangles risk and trust; as individuals enhance their knowledge they become increasingly aware of the world around them and consequently seek for ways to deal with risk whilst questioning the trustworthiness of individuals and institutions (Beck, Giddens and Lash, 1994).

The issue of the involvement of Big Pharma in the genomic research process sparked concerns amongst the vast majority of participants. A key conclusion is that there is a sense of mistrust towards the pharmaceutical industry, amongst patients and non-patients alike, as it is perceived as being exclusively motivated by profit. Nonetheless, Big Pharma is at the same time considered crucial for providing a sense of hope, particularly amongst patients.

Despite expressing high levels of trust towards genomic researchers and biobankers, various patient focus group participants spoke of the need to reclaim power and control when it comes to certain specific circumstances. The issues of incidental findings and data sharing sparked critical feedback, and a desire for power and control, from both patient and non-patient focus group participants. There was a general consensus that researchers who fail to report back secondary findings, as well as researchers who do not collaborate with other experts, would be indicating that their real motivation is not rooted in a desire for better health for the greater good, but rather from an egotistical desire for professional success.

The research experts interviewed clarified the issues of incidental findings and data sharing, claiming that both processes can, under certain circumstances, limit research as they sometimes lead to complex bureaucratic processes. Participants might not always be capable of visualising the real-life issues which might emerge from such practices, however, their perceptions towards these issues, whether they are informed perceptions or not, remain crucial as they can significantly impact potential participants' willingness to contribute to research.

Looking at the issue of consenting vis-à-vis power and control, it appears that whilst some participants perceive stringent ethical and legal standards of consenting as a hindrance to innovation and progress, and therefore as a threat to future health, others believe that ethics and laws give participants control, and provide security and protection.

The research experts also highlighted both facets of ethics and law, and despite conveying the message that preventing innovation is a risk in itself, their general conclusion was that even if high ethical standards and strict regulations may restrict research to a certain extent, ultimately they are indispensable for research to progress sustainably, as they attribute some level of control to all stakeholders, and thus curtail the risk for abuse.

For most patient focus group participants and especially for the patient support group representatives, the idea of dynamic consent came across as a balanced approach which allows the participants to be active, or not, depending on their preference, whilst attributing them with greater control and keeping them informed. An opposing perspective expressed by some of the non-patient focus group participants was that dynamic consent limits research, as research progress might end up being dependent on the involvement of participants who at times might prefer to be passive donors as opposed to active participants.

Although both patient and non-patient focus group participants expressed the need to retain some level of power and control throughout the process of participation in genomic research and biobanking, it appears that what occurs in practice does not reflect this ideal. The research experts declared that the vast majority of those who participate express little to no interest in the consenting process. This reveals that in real circumstances participants often abdicate any power that they hold as participants and choose to act passively by trusting in the professionals, especially when researchers are also clinicians and the distinction between the two roles becomes obscure. The lack of concern of genomic research participants could be attributed to the fact that most individuals would fail to recognise the implications of participation unless they are provided with an opportunity to discuss the issue deeply. In fact, various focus group participants felt that the focus group discussion was revealing and enriching as they had never thought of the implications which participation can involve. It can be concluded that the lack of concern amongst some participants, regarding the repercussions associated with genomics and biobanking, is not always the result of not fearing the negative outcomes, but mainly the result of a lack of knowledge about what the repercussions might be.

A key conclusion of this research is that participants' interpretations of issues of risk and trust are not straightforward; the two concepts are undoubtedly intertwined and dependent on each

other, but distinctions lie in perspectives and attitudes based on health status amongst other factors, where participants in the patients' focus groups were generally more trustful and less concerned about the risks. Nonetheless, the process of reflexivity as a result of enhanced knowledge was observed during all focus group sessions, as participants went through a process of "lay re-skilling" (Giddens, 1999). Whilst they gathered more information and became more knowledgeable through the focus group informative videos and discussions, participants started developing the skill of recognising the potential risks, and they consequently argued for greater control over the process of participating in genomic research and biobanking, as they, on varying levels, started to question the trustworthiness of their governance structures and procedures. Moreover, this research establishes that, for all focus group participants, the risks and benefits associated with participating in genomic research and biobanking are not mutually exclusive. Motivation often superseded the concerns that were raised after delving into the topic more deeply. This said, various participants felt that they should still maintain some level of control, and thus insisted that willingness to participate might depend on the research projects' approach towards specific issues, such as the consenting process, data sharing and incidental findings. This shows that reflexivity does not necessarily hinder collaboration, however, it empowers research participants to raise concerns and set standards.

6.1 Research limitations and recommendations

A limitation of this research is that data were collected towards the end of 2019, thus before the Covid-19 pandemic hit the globe. The recent events have likely impacted on perceptions and attitudes towards the medical system in general, and biomedical research in particular, which impact could not be analysed in this research.

The key challenge of this research was that the focus group participants lacked knowledge on the topic in question, and although the necessary tools were created to overcome this challenge and to enhance participation, it remains a fact that opinions might not always have been fully informed. Nonetheless, a lack of genomic knowledge generally is also the case amongst potential participants of genomic research and biobanking, and thus what emerged through this research is crucial, as it informs the scientific community about the key issues which need to be discussed on a deeper level during the consenting process.

Based on what emerged in this research, it is suggested that researchers and scientists treat the issue of a lack of scientific communication with urgency, in the hopes of improving awareness levels. The dependence of such a venture on human participation makes scientific communication indispensable for success.

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Appendix A - Quantitative study verbal consent

Your participation in this research programme is highly appreciated. This is a preliminary study which aims to examine the level of awareness about biobanks in the general public in Malta. If you agree to continue, I will ask you 4 very brief questions which should take about 5 minutes to complete.

Participation is completely voluntary and you may change your mind about participating at any point during our conversation. In this case all data you would have provided will be discarded.

By agreeing to answer my questions, you are giving your verbal consent and you are confirming that you are aged 18 years or over.

All the information that you do provide will remain anonymous; your identity details are not required for this research thus total anonymity and confidentiality are ensured.

The data will be analysed and findings will be presented as part of research project which is currently being carried out at the University of Malta.

Kunsens verbali (kwestjonarju):

Il-parteċipazzjoni tiegħek f'din ir-riċerka hija apprezzata. Din ir-riċerka qed issir biex teżamina l-għarfien tal-pubbliku ġenerali Malti dwar il-*'biobanks'.* Jekk int taċċetta li tipparteċipa, jien se insaqsik 4 mistoqsijiet qosra li m'għandhomx jieħdulek aktar minn 5 minuti.

Il-parteċipazzjoni tiegħek hija kompletament volontarja u int għandek id-dritt tieqaf tirrispondi x'ħin trid. F'dak il-kas, l-informazzjoni li tkun tajt tiġi eliminata.

Jekk taċċetta li tirrispondi dawn il-mistoqsijiet inti tkun qed tagħti kunsens orali u tkun qed tikkonferma li għandek eta' ta' 18-il sena jew aktar.

L-informazzjoni li se tipprovodi se tibqa' anonima; l-identita' tiegħek mhiex se tkun magħrufa għallura int se tibqa' totalment anonimu/a.

L-informazzjoni se tkun analiżata u r-riżultati se tkunu preżentati bħala parti minn riċerka li qed issir bħalissa fi ħdan l-Università ta' Malta.

Appendix B - Survey questionnaire

1. DO YOU KNOW WHAT A BIOBANK IS?

YES NO (If answer is ‘no’ skip to question 4)

2. WHAT IS IT?

_____

3. HOW DID YOU LEARN ABOUT BIOBANKS? (Skip to question 5)

_____

4. WHAT DO YOU THINK A BIOBANK IS?

_____

5. WHAT IS YOUR LEVEL OF EDUCATION?

Secondary Post-secondary Tertiary

6. GENDER:

Male Female Other

Kwestjonarju:

1. TAF X'INHU BIOBANK?

IVA LE (Jekk it-tweġiba hi le, mur għan-numru 4)

2. X'INHU?

_____

3. KIF SIRT TAF DWAR BIOBANKS? (Aqbeż għall-mistoqsija numru 5)

_____

4. X'TAĦSEB LI HU BIOBANK?

_____

5. X'INHU L-LIVELL TA' EDUKAZZJONI TIEGĦEK?

Sas-sekondarja Sixth Form Terzjarja

6. ĠENERU:

Maskil Femminil Iehor

Appendix C - Quantitative study information sheet

Thank you for participating in our research project which is being carried out within the Centre for Molecular Medicine and Biobanking, University of Malta.

Your participation in this research is completely anonymous, and you will not be identifiable in any way.

If you would like to know more about this research, please feel free to contact Dr Gillian Martin on 23402301 or Maria Desira on 23403218 (email: maria.desira@um.edu.mt).

Nota ta' informazzjoni (kwestjonarju):

Grazzi talli għadek kemm ipparteċipajt fir-riċerka li qed nagħmlu fi ħdan is-*'Centre for Molecular Medicine and Biobanking',* fl-Università ta' Malta.

Il-parteċipazzjoni tiegħek f'din ir-riċerka hija kompletament anonima u int ma tistax tkun identifikat/a bl-ebda mod.

F'każ li tkun trid issir taf aktar dwar din ir-riċerka, tista' ċċempel lil Dr Gillian Martin fuq 23402301 jew lil Maria Desira fuq 23403218 (e-mail: maria.desira@um.edu.mt).

Appendix D - Focus group recruitment letter

Dear Sir/Madame,

You are being invited to participate in a research study exploring local understandings, beliefs and attitudes related to the process of biobanking and genomic research. Biobanks are collections of blood and/ or other tissues amassed for the purpose of medical research.

This research project is being conducted by MA student & Research Support Officer Ms Maria Desira, under the supervision of Dr Gillian Martin, resident academic in the Department of Sociology Faculty of Arts, in collaboration with the Centre for Molecular Medicine and Biobanking, University of Malta. The objective of this research project is to further our understanding of how people in Malta feel about participating/ collaborating in genomic research, and donating their samples to biobanks.

There are no known risks associated with participating in this research study. The information you provide will help us understand how best to devise biobank governance policies in order to make them more trustworthy and facilitate more active participation on a national level. It will also help us in the development of an IT portal that is currently being designed to act as an interface between the researchers at the Centre for Molecular Medicine and Biobanking, the participants and the general public. The information you share will potentially enhance the possibilities for important bio-genetic research which could translate into new and revolutionary ways of treating genetic diseases. The collected data will also be used for a Master's degree dissertation which is being carried out by Maria Desira.

If you decide to participate, you will be invited to attend a focus group discussion where you, and other participants will view an introductory video about genomic research and biobanking and participate in a group discussion where we will explore your views and concerns related to the topic. The sessions are expected to last about an hour and a half and you will be given refreshments and a thank you gift for attending.

These group discussions will be voice recorded with your permission, however you will be assigned a pseudonym, and this will be used in connection with your data from then on. It will not be possible to link any of your input to your personal identity in the final publication of the research findings. Your data will be coded so that only the researchers are able to link your comments or data to your name. All data presented in reports, presentations or other final summaries will be in a summarised and pseudonymised so that no one will be able to identify you from your comments or data.

If you agree to participate, please contact me on phone number 23403218 or by email at maria.desira@um.edu.mt. Should you require any further information please send an email either to Maria Desira or to Dr Gillian Martin at gillian.martin@um.edu.mt.

Respectfully yours,

Maria Desira & Dr Gillian Martin PhD,

Centre of Molecular Medicine and Biobanking, L-Università ta' Malta

Ittra ta' reklutaġġ (focus groups):

Għażiż/a,

Inti mistieden/ mistiedna biex tipparteċipa fi studju li qed isir biex jesplora l-fehmiet, it-twemmin u l-attitudnijiet dwar il-proċess tal-*biobanking* u riċerka ġenomika. *Biobanks* huma kollezzjonijiet ta' demm u/ jew *tissues* oħrajn miġbura bil-għan li jintużaw fir-riċerka medika.

Din ir-riċerka qed issir minn studenta tal-Masters u riċerkatriċi, Maria Desira, taħt is-superviżjoni ta' Dr Gillian Martin. Din ir-riċerka iegħa ssir fi ħdan id-dipartiment tas-soċjoloġija u f'kollaborazzjoni ma' '*Centre for Molecular Medicine and Biobanking*', fi ħdan l-Università ta' Malta. L-għan ta' din ir-riċeka huwa li biex nifhmu iżjed kif in-nies jaħsbuha dwar is-sehem jew il-kollaborazzjoni tagħhom f'riċerka ġenomika, u dwar li jagħtu l-kampjuni tagħhom lil *biobanks*.

M'hemm l-ebda riskju assoċjat ma' li tipparteċipa f'din ir-riċerka. L-informazzjoni li tipprovdi se tgħinna nifhmu kif ikun l-aħjar mod biex jinħolqu *policies* dwar l-immaniġjar ta' *biobanks* li jkunu aktar fdati mill-poplu u li jiffaċiltaw u jżidu l-partiċipazzjoni fuq skala nazzjonali. Din se tgħinna wkoll fl-iżvilupp ta' sit tal-I.T li bħalissa qed tiġi ddisinjata sabiex tkun użata mir-riċerkaturi fiċ-Ċentru, il-parteċipanti u l-pubbliku ġenerali. L-informazzjoni li tagħti potenzjalment tista' tkabbar il-possibilitajiet għal riċerka importanti tal-bioġenetika li tista' twassal għal modi ġodda u revoluzzjonarji biex jiġi trattat mard ġenetiku. L-informazzjoni li tingabar se tintuża ukoll minn Maria Desira għar-riċerka tagħha fl-istudji tal-Masters fl-Università ta' Malta.

Jekk inti tiddeċiedi li tipparteċipa, se tiġi mitlub/a tattendi diskussjoni f'forma ta' *focus group* fl-Università ta' Malta, fejn inti, u parteċipanti oħra, se tintwerew filmat dwar ir-riċerka ġenomika u *biobanking*, u wara tipparteċipaw f'diskussjoni bejn il-grupp fejn inti tkun tista' taqsam dak li taħseb u dak li jinkwetak dwar ir-riċerka ġenomika u l-*biobanking*. Is-sessjoni mistennija ddum madwar siegħa u nofs. Se jkun hemm *refreshments* u se tingħata rigal żgħir bħala ringrazzjament talli tkun attendejt.

Dawn id-diskussjonijiet se jkunu rrekordjati b'*voice recorder* bil-permess tiegħek, però inti se tkun qed tingħata isem fitizzju. Dan se jintuża b'rabta mal-informazzjoni tiegħek minn dak il-ħin 'il quddiem. B'dan il-mod, l-informazzjoni li tingabar se tkun ikkodiċizzata. Mhux se jkun possibbli li wieħed jassoċja dak li inti tgħid mal-identità personali tiegħek fil-pubblikazzjoni tar-riżultati tar-riċerka. L-informazzjoni tiegħek se tkun ikkodiċizzata sabiex ir-riċerkaturi biss ikunu jistgħu jorbtu l-kummenti tiegħek jew l-informazzjoni mal-isem. L-informazzjoni kollha li tagħti inti, u li tiġi ippreżentata f'rapporti, fi preżentazzjonijiet u/ jew f'kitbiet oħrajn, se tkun qed tiġi ppublikata b'mod li ħadd ma jkun jista' jidentifikak mill-kummenti u l-informazzjoni tiegħek.

Jekk tixtieq tipparteċipa inti mitlub titkellem miegħi fuq in-numru 23403218 jew permezz ta' email fuq maria.desira@um.edu.mt. Għal aktar informazzjoni tista' tibgħat email lil Maria Desira jew lil Dr Martin fuq gillian.martin@um.edu.mt

Dejjem tiegħek,

Maria Desira & Dr Gillian Martin PhD, Centre of Molecular Medicine and Biobanking,

L-Università ta' Malta

Appendix E - Focus group consent form

This research study is aimed at exploring local understandings, beliefs and attitudes related to the process of biobanking and genomic research. This research project is being conducted by MA student & Research Support Officer Maria Desira, under the supervision of Dr Gillian Martin, resident academic in the Department of Sociology Faculty of Arts, in collaboration with the Centre for Molecular Medicine and Biobanking, University of Malta. The objective of this research project is to further our understanding of how people in Malta feel about participating/ collaborating in genetic research, and donating their samples to biobanks.

There are no known risks associated with participating in this research study. The information you provide will help us understand how best to devise biobank governance policies in order to make them more trustworthy and facilitate more active participation on a national level. It will also help us design and produce educational video clips which will form part of an IT portal that is currently being designed to act as an interface between the researchers at the Centre for Molecular Medicine and Biobanking, the participants and the general public. The information you share will not necessarily benefit you directly, however it will potentially enhance the possibilities for important bio-genetic research which could translate into new and revolutionary ways of treating genetic diseases. The collected data will also be used for a Master's degree dissertation which is being carried out by Maria Desira.

This group discussion will be recorded with your permission, however you will be assigned a pseudonym at the start, and this will be used in connection with your data from then on. It will not be possible to link any of your input to your personal identity in the final publication of the research findings. Your data will be coded so that only the researchers are able to link your comments or data to your name. All data presented in reports, presentations or other final summaries will be in a summarised and pseudonymised so that no one will be able to identify you from your comments or data.

CONFIDENTIALITY

Every effort will be made to ensure confidentiality of any identifying information collected in this study:

- We will ask you to sign below to indicate that you will keep all comments made during the focus group confidential and not discuss what happened during the focus group, after the meeting.
- Your identity, however, will be known to other focus group participants and it would be unrealistic to guarantee that others in these groups will respect the confidentiality of the group.
- Focus groups will be recorded and transcribed at which point, all identifying information, will be removed.
- Recordings will be downloaded onto a password-protected computer and the original recordings will be deleted.
- All transcriptions will be stored on a password-protected computer.
- Only the researcher Ms Maria Desira & Dr Gillian Martin, will have access to focus group data.

HANDLING AND SECURITY OF DATA

- Every effort will be made to ensure that your privacy and confidentiality is protected throughout the study. Data will be kept for a maximum of 5 years and then destroyed by the researchers.

Your participation in this study is voluntary.

Please remember that you may choose to withdraw from the study at any time. You may exercise the

option of removing your data from the study, however please note that once all identifying information has been removed, all focus group responses will become anonymous and it will not be possible for participants to ask for their data to be removed from the study.

If you agree to participate, please fill in the consent section below. You will be given a copy of this letter for your reference.

Maria Desira

University of Malta

==========

Consent section

I have read and understood the above information and agree to participate in the research project as described above. I understand that my input, when published, will be anonymous and that I may decide to stop participating at any stage. I also understand that the content of the discussion of the focus group is confidential and that I agree to maintain confidentiality of information shared in this group.

Name and surname _____ Date _____ Signature _____

Maria Desira, Master of Arts (Sociology) Student & Research Support Officer

_____ (Signature)

Formola ta' kunsens (focus groups):

Dan l-istudju qed isir biex jesplora l-fehmiet, it-twemmin u l-attitudnijiet dwar il-proċess tal-*biobanking* u riċerka ġenomika. Din ir-riċerka qed issir mill-istudenta tal-Master of Arts (Soċjoloġija) u Research Support Officer Maria Desira, taħt is-supervizjoni ta' Dr Gillian Martin, li hija akkademika residenti fid-Dipartiment tas-Soċjoloġija fil-Fakultà tal-Arti. Din qed isir f'kollaborazzjoni ma' '*Centre for Molecular Medicine and Biobanking*', fi ħdan l-Università ta' Malta. L-għan ta' din ir-riċeka huwa li biex nifhmu iżjed kif in-nies jaħsbuha dwar is-sehem jew il-kollaborazzjoni tagħhom f'riċerka ġenetika, u dwar li jagħtu l-kampjuni tagħhom lil *biobanks*.

M'hemm l-ebda riskju assoċjat ma' li tipparteċipa f'din ir-riċerka. L-informazzjoni li tipprovdi se tgħinna nifhmu kif ikun l-aħjar mod biex jinħolqu *policies* dwar l-immaniġjar ta' *biobanks* li jkunu aktar fdati mill-poplu u li jiffaċiltaw u jżidu l-partiċipazzjoni fuq skala nazzjonali. Din se tgħinna wkoll biex niddisinjaw u nipproduċu filmati edukattivi li jkunu parti mis-sit tal-I.T li bħalissa qed tiġi ddisinjata użata mir-riċerkaturi fiċ-Ċentru, il-parteċipanti u l-pubbliku ġenerali. L-informazzjoni li tagħti mhux bilfors se tibbenefika minnha inti direttament, però potenzjalment tista' tkabbar il-possibilitajiet għal riċerka importanti tal-bioġenetika li tista' twassal għal modi ġodda u revoluzzjonarji biex jiġi trattat mard ġenetiku. L-informazzjoni li tingħabar se tintuża ukoll minn Maria Desira għar-riċerka tagħha fl-istudji tal-Masters fl-Università ta' Malta.

Din id-diskussjoni se tkun rrekordjata bil-permess tiegħek, però inti se tkun qed tingħata isem fitizzju. Dan se jintuża b'rabta mal-informazzjoni tiegħek minn dak il-ħin 'il quddiem. B'dan il-mod, l-informazzjoni li tingħabar se tkun ikkodiċizzata. Mhux se jkun possibbli li wieħed jassoċja dak li inti tgħid mal-identità personali tiegħek fil-pubblikazzjoni tar-riżultati tar-riċerka. L-informazzjoni tiegħek se tkun ikkodiċizzata sabiex ir-riċerkaturi biss ikunu jistgħu jorbtu l-kummenti tiegħek jew l-informazzjoni mal-isem. L-informazzjoni kollha li tagħti inti, u li tiġi ippreżentata f'rapporti, fi preżentazzjonijiet u/ jew f'kitbiet oħrajn, se tkun ippreżentata b'mod li ħadd ma jkun jista' jidentifikak mill-kummenti u l-informazzjoni tiegħek.

Il-Kunfidenzjalità

Se jsir kull sforz biex nassiguraw l-kunfidenzjalità ta' kull informazzjoni miġbura f'dan l-istudju:

- Ir-*recordings* se jinżammu f'kompjuter li għandu *password* protetta u r-rekordings oriġinali se jiġu mħassra.
- It-traskrizzjonijiet kollha se jiġu maħżuna f'kompjuter li għandu *password*.
- Ir-riċerkaturi Maria Desira u Dr Gillian Martin biss se jkollhom aċċess għall-informazzjoni li tidentifika l-parteċipant.

Is-sigurtà tal-informazzjoni

- Se jsir kull sforz biex nassiguraw li l-privatezza u l-kunfidenzjalità tkun protetta matul l-istudju. L-informazzjoni se tinżamm għall-massimu ta' ħames snin u tinqered mir-riċerkaturi kif jgħaddi dan il-perjodu.

Il-parteċipazzjoni tiegħek issir b'mod voluntarju. Jekk tiddeċiedi li tieħu sehem, jekk jogħġbok ftakar li tista' tieqaf tieħu sehem u tista' tagħżel li titħassar l-informazzjoni kollha li tkun tajt għal dan l-istudju. Pero' fi stadju aktar avvanzat, meta l-informazzjoni li tkun tajt tkun ġiet anonimizzata, ma jkunx possibli li l-parteċipanti jagħżlu li titħassar l-informazzjoni li jkunu provdew.

Jekk inti taqbel li tipparteċipa, jekk jogħġbok imla s-sezzjoni ta' hawn taħt u li biha qed tagħti l-kunsens. Inti se tingħata kopja ta' din l-ittra għar-referenza tiegħek.

Maria Desira

L-Università ta’ Malta

==========

Sezzjoni ta’ Kunsens

Jiena qrajt u fhimt l-informazzjoni t’hawn fuq u naqbel li nipparteċipa f’din ir-riċerka kif deskritta hawn fuq. Jiena nifhem li l-informazzjoni li se nagħti, meta tiġi ppubblikata, se tinżamm anonima u li jiena nista’ nieqaf nipparteċipa f’kwalunkwe stadju. Nifhem ukoll li l-kontenut li joħroġ mid-diskussjoni tal-*focus group* huwa kunfidenzjali u jien naqbel li għandi nżomm il-kunfidenzjalita’ tal-informazzjoni kollha li toħroġ fil-grupp.

Isem u Kunjom _____ Data _____ Firma _____

Maria Desira, Studenta tal-Master of Arts fis-Soċjoloġija u Research Support Officer

_____ (Firma)

Appendix F - Focus group discussion guide

Before we kick off the discussion, I need to raise a few important points:

- I kindly ask you to keep all that the other participants say confidential. Please do not repeat anything that the others say outside of this room.
- There is no right or wrong answer, so please feel free to say whatever you feel is right. We are only after your opinions, and thus whatever you say will be considered valid.
- (In the case of patients/ parents of patients) You are not obliged to give details about your condition or that of your child – if you prefer to refer to the condition please feel free, but it's important that you know that you are not obliged to do so.

I have prepared some video clips which will help you understand better what biobanking is, and what research is currently being carried out at the University of Malta:

1. Genetika Info Clip
2. Donors Clip
3. Lawyer Dr Alistair De Gaetano Clip

Guidance points for focus group discussion:

(In the case of patients/ parents of patients) Do you mind people knowing about your condition? Do you think that there are people who try to keep it a secret?

What interest do you have in genomic research? [patient? family members of a patient? scientist? doctor? neither of these?]

Are you interested in research? Do you keep yourself updated?

Do you think that genomic research is important? [why? why not? for who? who has the most to gain? what is there to lose?]

What motivates you to participate?

(In the case of patients/ parents of patients) Do you think that enough is being done with regards to your condition? If there was the need for you to contribute, would you be willing to do so? Even if you knew that an outcome would take many years?

Do you think that there any problems related to the donation of blood, saliva or tissues to the biobank? [privacy? personal information? genetic information? breach of privacy? control over who uses your sample and how? what worries you? discrimination? insurance? employment? use of samples which goes against your morals?]

How do feel about giving consent to researchers to use your sample and data for research? [any conditions? one-time consent for a variety of studies? would you want to know the details of each study? why? why not? if you have already participated, do you feel that you were given enough information?]

What motivates you to participate? [science? the common good? cure for my condition? to learn more about my DNA? for a cure to be found for future generations?]

Is there anything which would demotivate you from participating? [privacy? data protection? fear of what might result?]

Would you accept that your data, or that of your child, is shared with other researchers? What about international research bodies? Do you trust local researchers more or less than foreign ones?

What do you think about the profit that is made by the pharmaceutical industry from research which is conducted using the samples of patients?

How do you feel about the researchers? Do you think that your data can be breached? And if that happened would you mind? [crucial for research? fear of a lack of privacy? would you want information about secondary findings which might emerge? is there a risk? what is it?]

Would you expect to receive something back? [results? what kind? updates regarding the progress of research?]

How do you think that researchers should handle incidental findings? Do such findings scare you?

What do you think about the future? [aims? future medical treatments? future generations?]

Is there anything else that you would like to add?

Punti ta' gwida għad-diskussjoni fi gruppi:

Importanti li qabel nibdew naqsam magħkom dawn il-punti:

- Dak li se jingħad, għandu jibqa' kollu f'din il-kamra, nitlobkom biex dak li tisimgħu hawn ġew iżżommuh għalikom.
- Hawnekk m'hawnx tweġiba korretta jew skoretta, jiġifieri m'hemmx xi ħaġa li suppost tridu tgħidu, ħlief l-oppinjoni tagħkom, jiġifieri tgħidu x'tgħidu huwa validu… Aħna irridu biss l-oppinjonijiet tagħkom.
- (Fil-każ ta' pazjenti u ġenituri ta' pazjenti) Meta qegħdin titkellmu m'intomx obbligati li tagħtu dettalji tal-kundizzjoni ta' uliedkom jew tagħkom – għalkemm jekk intom tixtiequ li titkellmu dwar dan għandkom tħossukom komdi li tagħmlu dan – pero' m'intomx obbligati li tiżvelaw dettalji.

Ħa nibdew mill-ewwel billi naraw il-clips li bihom se tifmhu aktar x'qed isir fl-Università bħalissa:

1. Filmat Ġenetika Info
2. Filmat Donaturi
3. Filmat Avukat Dr De Gaetano

Punti għal *focus groups:*

(Fil-każ ta' pazjenti u ġenituri ta' pazjenti) Tiddejjaq li ħaddieħor ikun jaf dwar il-kundizzjoni tiegħek? La qegħdin ifisser li komdi titkellmu fuqha? Taħseb li hemm persuni li jippruvaw jaħbuha?

X'interess għandek fir-riċerka ġenetika? [pazjent? familjari ta' pazjent? xjentist? student? tabib? l-ebda?]

Tinteressa ruħek fir-riċerka? Izzomm ruħek aġġornat?

Taħseb li r-riċerka ġenomika hija importanti? [għaliex? għaliex le? għal min? min għandu l-aktar x'jigwadanja? hemm x'titlef?]

X'jimmotivak u xi jħeġġek biex tipparteċipa?

(Fil-każ ta' pazjenti u ġenituri ta' pazjenti) Taħseb li qed isir bizzejjed dwar il-kundizzjoni tiegħek? Jekk ikun hemm bzonn il-kontibut tagħkom għal xi studju lesti li tgħinu? Anki jekk taf li biex joħroġ xi ħaġa minn dik ir-riċerka jridu jgħaddu ħafna u ħafna snin?

Taħseb li jeżistu problemi inkwetanti relatati mad-donazzjoni ta' demm jew *tissues* oħrajn lil *biobank*? [privatezza? informazzjoni personali? informazzjoni ġenetika? breach of privacy? kontroll fuq min u kif jintużaw il-kampjuni tiegħek? x'jinkwetak? diskriminazzjoni? insurance? xogħol? użu kontra l-morali tiegħek?]

Xi tħoss dwar li tagħti permess u l-kunsens lir-riċerkaturi sabiex jużaw id-demm tiegħek? [x'kundizzjonijiet? tagħti kunsens kull darba għal kull riċerka differenti? trid tkun taf id-dettalji dwar kull studju li jsir? għaliex? għaliex le? jekk diġà qed tipparteċipa f'riċerka, ingħatajt bizejjed informazzjoni meta ġejt biex tagħti kunsens? tħossok li fhimt biżżejjed?]

X’inhi l-akbar motivazzjoni biex tipparteċipa? [xjenza? għal ġid tal-komunità? biex tinstab kura għal marda tiegħek? biex issir taf aktar dwar id-DNA tiegħek? biex tinstab kura għal ta’ warajna?]

X’jista’ jkun li tkun ir-raġuni biex tagħżel li ma tipparteċipax? [privatezza? protezzjoni tad-*data*? biża minn dak li jista’ jirriżulta?]

Taċċetta li l-informazzjoni tieġħek/ ta’ uliedek tingħata lil riċerkaturi oħrajn? U jekk tintbgħat barra? Tafda r-riċerka li ssir Malta aktar minn dik li ssir barra jew inqas?

X’taħseb dwar il-profit li jsir minn kumpaniji tal-farmaċewtika għal mediċini li jkunu rriżultaw permezz tar-riċerka ġenomika li għaliha jkunu kkontribwixxew il-pazjenti stess?

Kif tħossok dwar ir-riċerkaturi? Taħseb li jista’ jkun li jsiru jafu li dak il-kampjun partikolari hu tiegħek? U jekk jiġri hekk tiddejjaq? [kruċjali għal proċess tar-riċerka? biża minn nuqqas ta’ privatezza? informazzjoni dwar riżultati sekondarji li jistgħu joħorġu mir-riċerka? riskju? għaliex? għaliex le?]

Hemm xi ħaġa li tkun qed tistenna li tirċievi lura talli tkun ipparteċipajt? [riżultati? x’tip? *updates* dwar il-progress tal-proġett ta’ riċerka? oħrajn?]

X’taħseb li għandu jsir minn informazzjoni li jsiru jafu biha r-riċerkaturi li ma jkollix x’taqsam mal-kundizzjoni li tafu biha diġà? Din tbeżżakom? [skoperti inċidentali]

X’taħseb dwar il-futur? [għanijiet tar-riċerka tal-futur? futur tal-kampjuni għar-riċerka? ġenerazzjonijiet futuri? trattamenti mediċi futuri?]

Hemm xi ħaġa oħra x’iżżid? Ħallejna xi ħaġa ta’ importanza barra?

Appendix G - Focus group information sheet

Thank you for participating in a research study exploring local understandings, beliefs and attitudes related to the process of biobanking and genomic research. This research project is being conducted by Maria Desira under the supervision of Principal Investigator Dr Gillian Martin, resident academic in the Department of Sociology Faculty of Arts, in collaboration with the Centre for Molecular Medicine and Biobanking, University of Malta. The objective of this research project is to further our understanding of how people in Malta feel about participating/ collaborating in genetic research, and donating their samples to biobanks.

There are no known risks associated with participating in this research study. The information you provided will help us understand how best to devise biobank governance policies in order to make them more trustworthy and facilitate more active participation on a national level. It will also help us design and produce educational video clips which will form part of an IT portal that is currently being designed to act as an interface between the researchers at the Centre for Molecular Medicine and Biobanking, the participants and the general public. The information you shared will not necessarily benefit you directly, however it will potentially enhance the possibilities for important bio-genetic research which could translate into new and revolutionary ways of treating genetic diseases.

This focus group discussion has been recorded with your permission, however you will be assigned a pseudonym for yourself, and this will be used in connection with your data from then on. It will not be possible to link any of your input to your personal identity in the final publication of the research findings. Your data will be coded so that only the researchers are able to link your comments or data to your name.

CONFIDENTIALITY

Every effort will be made to ensure confidentiality of any identifying information collected in this study:

- Your identity will not be revealed, however, will be known to the other focus group participants and it would be unrealistic to guarantee that others in these groups will respect the confidentiality of the group.
- The recordings of the focus group will be transcribed at which point, all identifying information, will be removed.
- Recordings will be downloaded onto a password-protected computer and the original recordings will be deleted.
- All transcriptions will be stored on a password-protected computer.
- Only the researchers Ms Maria Desira & Dr Gillian Martin, will have access to focus group data.

HANDLING AND SECURITY OF DATA

- Every effort will be made to ensure that your privacy and confidentiality is protected throughout the study. Data will be kept for a maximum of 5 years and then destroyed.

Please remember that you may choose to withdraw from the study at any time. You may exercise the option of removing your data from the study, however please note that once all identifying information has been removed, all focus group responses will become anonymous and it will not be possible for participants to ask for their data to be removed from the study.

Should you have any questions or concerns about being in this study, you may contact me on 23403218 or maria.desira@um.edu.mt

Respectfully yours,

Maria Desira

Informazzjoni (focus groups):

Grazzi talli pparteċipajt f'dan l-istudju li se jesplora l-fehmiet, it-twemmin u l-attitudnijiet dwar il-proċess tal-*biobanking* u riċerka ġenomika. Din ir-riċerka qed issir mill-istudenta tal-Master of Arts (Soċjoloġija) u Research Support Officer Ms Maria Desira, taħt is-supervizjoni ta' Dr Gillian Martin, li hija akkademika residenti fid-Dipartiment tas-Soċjoloġija fil-Fakultà tal-Arti. Din qed isir f'kollaborazzjoni ma' '*Centre for Molecular Medicine and Biobanking*', fi ħdan l-Università ta' Malta. L-għan ta' din ir-riċeka huwa li biex nifhmu iżjed kif in-nies jaħsbuha dwar is-sehem jew il-kollaborazzjoni tagħhom f'riċerka ġenetika, u dwar li jagħtu l-kampjuni tagħhom lil *biobanks*.

M'hemm l-ebda riskju assoċjat ma' li tipparteċipa f'din ir-riċerka. L-informazzjoni li tipprovdi se tgħinna nifhmu kif ikun l-aħjar mod biex jinħolqu *policies* dwar l-immaniġjar ta' *biobanks* li jkunu aktar fdati mill-poplu u li jiffaċiltaw u jżidu l-partiċipazzjoni fuq skala nazzjonali. Din se tgħinna wkoll biex niddisinjaw u nipproduċu filmati edukattivi li jkunu parti mis-sit tal-I.T li bħalissa qed tiġi ddisinjata użata mir-riċerkaturi fiċ-Ċentru, il-parteċipanti u l-pubbliku ġenerali. L-informazzjoni li tagħti mhux bilfors se tibbenefika minnha inti direttament, però potenzjalment tista' tkabbar il-possibilitajiet għal riċerka importanti tal-bioġenetika li tista' twassal għal modi ġodda u revoluzzjonarji biex jiġi trattat mard ġenetiku.

Id-diskussjoni se tkun rrekordjata bil-permess tiegħek, però inti se tkun qed tingħata isem fitizzju. Dan se jintuża b'rabta mal-informazzjoni tiegħek minn dak il-ħin 'il quddiem. B'dan il-mod, l-informazzjoni li tingabar se tkun ikkodiċizzata. Mhux se jkun possibbli li wieħed jassoċja dak li inti tgħid mal-identità personali tiegħek fil-pubblikazzjoni tar-riżultati tar-riċerka. L-informazzjoni tiegħek se tkun ikkodiċizzata sabiex ir-riċerkaturi biss ikunu jistgħu jorbtu l-kummenti tiegħek jew l-informazzjoni mal-isem. L-informazzjoni kollha li tagħti inti, u li tiġi ippreżentata f'rapporti, fi preżentazzjonijiet u/ jew f'kitbiet oħrajn, se tkun mqassra u mqassam f'sezzjonijiet b'mod li ħadd ma jkun jista' jidentifikak mill-kummenti u l-informazzjoni tiegħek.

Il-Kunfidenzjalità

Se jsir kull sforz biex nassiguraw l-kunfidenzjalità ta' kull informazzjoni miġbura f'dan l-istudju:

- Ir-*recordings* se jinżammu f'kompjuter li għandu *password* protetta u r-rekordings oriġinali se jiġu mħassra.
- It-traskrizzjonijiet kollha se jiġu maħżuna f'kompjuter li għandu *password*.
- Ir-riċerkaturi Maria Desira u Dr Gillian Martin biss se jkollhom aċċess għall-informazzjoni li tidentifika l-parteċipant.

Is-sigurtà tal-informazzjoni

- Se jsir kull sforz biex nassiguraw li l-privatezza u l-kunfidenzjalità tkun protetta matul l-istudju. L-informazzjoni se tinżamm għall-massimu ta' ħames snin u tinqered mir-riċerkaturi kif jgħaddi dan il-perjodu.

Infakkrek li għandek id-dritt li tieqaf tieħu sehem f'dan l-istudju u tista' tagħżel li titħassar l-informazzjoni kollha li tkun tajt għal dan l-istudju. Pero' fi stadju aktar avvanzat, meta l-informazzjoni li tkun tajt tkun ġiet anonimizzata, ma jkun possibli li l-parteċipanti jagħżlu li titħassar l-informazzjoni li tkunu provdew.

Jekk għandek xi mistoqsijiet jew xi tħassib dwar dan l-istudju, tista' iċċempilli fuq 23403218 jew tibgħatli imejl fuq maria.desira@um.edu.mt

Dejjem tiegħek,

Maria Desira

Appendix H - Expert interview recruitment letter

You are being invited to participate in a research study exploring local understandings, beliefs and attitudes related to the process of biobanking and genomic research. This research project is being conducted by MA student & Research Support Officer Ms Maria Desira, under the supervision of Dr Gillian Martin, resident academic in the Department of Sociology Faculty of Arts, in collaboration with the Centre for Molecular Medicine and Biobanking, University of Malta. The objective of this research project is to further our understanding of how people in Malta feel about participating/ collaborating in genetic research, and donating their samples to biobanks, and also about how you experts feel about the process of biomedical research and biobanking.

There are no known risks associated with participating in this research study. The information you provide will help us understand how best to devise biobank governance policies in order to make them more trustworthy and facilitate more active participation on a national level. It will also help us design and produce educational video clips which will form part of an IT portal that is currently being designed to act as an interface between the researchers at the Centre for Molecular Medicine and Biobanking, the participants and the general public.

If you decide to participate, you will take part in an in-depth, semi-structured interview exploring issues related to your work in the area of genomic research and biobanking. Issues of key interest are those related to participant rights, trust, risk, consent, and data sharing.

ANONYMITY

The interview will be audio-recorded with your permission however you will assigned a pseudonym, and this will be used in connection with your data from then on. In this way, data collected during your interview will be coded (pseudo-anonymised). It will not be possible to link any of your input to your personal identity in the final publication of the research findings. Your data will be coded so that only the researchers are able to link your comments or data to your name. All data presented in reports, presentations or other final summaries will be pseudonymised so that no one will be able to identify you from your comments or data.

CONFIDENTIALITY

Every effort will be made to ensure confidentiality of any identifying information collected in this study:

- Recordings will be downloaded onto a password-protected computer and the original recordings will be deleted.
- All transcriptions will be stored on a password-protected computer.
- Only the researchers Ms Maria Desira & Dr Gillian Martin, will have access to focus group data.

HANDLING AND SECURITY OF DATA

- Every effort will be made to ensure that your privacy and confidentiality is protected throughout the study. Data will be kept for a maximum of 5 years and then destroyed by the researcher.

Your participation in this study is voluntary. If you decide not to take part, this decision will have no impact on you at all. If you do decide to take part, please remember that you may choose to withdraw from the study at any time and may exercise the option of removing your data from the study.

If you agree to participate you may contact me at maria.desira@um.edu.mt or by calling on 23403218. Should you require any further information you may also email Dr Martin at gillian.martin@um.edu.mt

Repectfully yours,

Maria Desira

Ittra ta' reklutaġġ (intervista mal-esperti):

Inti mistieden/ mistiedna biex tipparteċipa fi studju li qed isir biex jesplora l-fehmiet, it-twemmin u l-attitudnijiet dwar il-proċess tal-*biobanking* u riċerka ġenomika. Din ir-riċerka qed issir mill-istudenta tal-Master of Arts (Soċjoloġija) u Research Support Officer Ms Maria Desira, taħt is-supervizjoni ta' Dr Gillian Martin, li hija akkademika residenti fid-Dipartiment tas-Soċjoloġija fil-Fakultà tal-Arti. Din qed isir f'kollaborazzjoni ma' '*Centre for Molecular Medicine and Biobanking*', fi ħdan l-Università ta' Malta. L-għan ta' din ir-riċeka huwa li biex nifhmu iżjed kif in-nies jaħsbuha dwar is-sehem jew il-kollaborazzjoni tagħhom f'riċerka ġenetika, u dwar li jagħtu l-kampjuni tagħhom lil *biobanks,* u kif ukoll biex nifhmu kif intom l-esperti taħsbuha dwar il-proċess tar-riċerka medika.

M'hemm l-ebda riskju assoċjat ma' li tipparteċipa f'din ir-riċerka. L-informazzjoni li tipprovdi se tgħinna nifhmu kif ikun l-aħjar mod biex jinħolqu *policies* dwar l-immaniġjar ta' *biobanks* li jkunu aktar fdati mill-poplu u li jiffaċiltaw u jżidu l-partiċipazzjoni fuq skala nazzjonali. Din se tgħinna wkoll biex niddisinjaw u nipproduċu filmati edukattivi li jkunu parti mis-sit tal-I.T li bħalissa qed tiġi ddisinjata użata mir-riċerkaturi fiċ-Ċentru, il-parteċipanti u l-pubbliku ġenerali.

Jekk inti tiddeċiedi li tipparteċipa, inti se tieħu sehem b'mod profond f'intervista semistruttura li tesplora *issues* relatati mal-ħidma tiegħek fis-settur ta' riċerka genomika u *biobanking*. L-aktar dwar kwistjonijiet relatati mad-drittijiet, il-fiduċja, ir-riskju, il-kunsens, it-tqassim tal-informazzjoni tal-parteċipant ma' sorsi terzi.

L-Anonimità

L-intervista se tiġi rrekordjata bil-permess tiegħek, però inti se tiġi assenjat isem fitizzju. Dan se jintuża b'rabta mal-informazzjoni tiegħek minn dak il-ħin 'il quddiem. B'dan il-mod, l-informazzjoni li tinġabar matul l-intervista se tkun ikkodiċizzata. Mhux se jkun possibbli li wieħed jassoċja dak li inti tgħid mal-identità personali tiegħek, la waqt il-proċess u lanqas fil-pubblikazzjoni tar-riżultati tar-riċerka. L-informazzjoni tiegħek se tkun ikkodiċizzata sabiex ir-riċerkaturi biss ikunu jistgħu jorbtu l-kummenti tiegħek jew l-informazzjoni mal-isem. L-informazzjoni kollha li tagħti inti, u li tiġi ippreżentata f'rapporti, fi preżentazzjonijiet u/ jew f'kitbiet oħrajn, se tkun ippreżentata b'mod li ħadd ma jkun jista' jidentifikak mill-kummenti u l-informazzjoni tiegħek.

Il-Kunfidenzjalità

Se jsir kull sforz biex nassiguraw l-kunfidenzjalità ta' kull informazzjoni miġbura f'dan l-istudju:

- Ir-*recordings* se jinżammu f'kompjuter li għandu *password* protetta u r-rekordings oriġinali se jiġu mħassra.
- It-traskrizzjonijiet kollha se jiġu maħżuna f'kompjuter li għandu *password*.
- Ir-riċerkaturu Maria Desira u Dr Gillian Martin biss se jkollhom aċċess għall-informazzjoni li tidentifika l-parteċipant.

Is-sigurtà tal-informazzjoni

- Se jsir kull sforz biex nassiguraw li l-privatezza u l-kunfidenzjalità tkun protetta matul l-istudju. L-informazzjoni se tinżamm għall-massimu ta' ħames snin u tinqered minn Maria Desira u Dr Gillian Martin kif jgħaddi dan il-perjodu.

Il-parteċipazzjoni tiegħek issir b'mod voluntarju. Jekk inti tiddeċieda li ma tiħux sehem, id-deċiżjoni

tiegħek mhux se tħalli impatti fuqek, bl-ebda mod. Jekk tiddeċiedi li tieħu sehem, jekk jogħġbok ftakar li tista' tieqaf tieħu sehem f'kwalunke ħin u tista' tagħżel li titħassar l-informazzjoni kollha li tkun tajt għal dan l-istudju.

Jekk tixtieq li tippartecipa nitolbok tagħmel kuntatt miegħi fuq 23403218 jew permezz ta' imejl fuq maria.desira@um.edu.mt. Jekk għandek xi mistoqsijiet jew xi tħassib dwar dan l-istudju, tista' iċċempel jew tibgħat imejl lili, jew inkella tagħmel kuntatt ma' Dr Martin fuq gillian.martin@um.edu.mt.

Dejjem tiegħek,

Maria Desira

Appendix I - Expert interview consent form

You are being invited to participate in a research study exploring local understandings, beliefs and attitudes related to the process of biobanking and genomic research. This research project is being conducted by MA student & Research Support Officer Ms Maria Desira, under the supervision of Dr. Gillian Martin, resident academic in the Department of Sociology Faculty of Arts, in collaboration with the Centre for Molecular Medicine and Biobanking, University of Malta. The objective of this research project is to further our understanding of how people in Malta feel about participating/ collaborating in genetic research, and donating their samples to biobanks, and also about how you experts feel about the process of biomedical research and biobanking.

There are no known risks associated with participating in this research study. The information you provide will help us understand how best to devise biobank governance policies in order to make them more trustworthy and facilitate more active participation on a national level. It will also help us design and produce educational video clips which will form part of an IT portal that is currently being designed to act as an interface between the researchers at the Centre for Molecular Medicine and Biobanking, the participants and the general public.

Participation will involve taking part in an in-depth, semi-structured interview exploring issues related to your work in the area of genomic research and biobanking. Issues of key interest are those related to participant rights, trust, risk, consent, and data sharing.

ANONYMITY

The interview will be recorded with your permission however you will be assigned a pseudonym, and this will be used in connection with your data from then on. In this way, data collected during your interview will be coded (pseudo-anonymised). It will not be possible to link any of your input to your personal identity in the final publication of the research findings. Your data will be coded so that only the researchers are able to link your comments or data to your name. All data presented in reports, presentations or other final summaries will be pseudonymised so that no one will be able to identify you from your comments or data.

CONFIDENTIALITY

Every effort will be made to ensure confidentiality of any identifying information collected in this study:

- Recordings will be downloaded onto a password-protected computer and the original recordings will be deleted.
- All transcriptions will be stored on a password-protected computer.
- Only Maria Desira and Dr Gillian Martin, will have access to identifier links.

HANDLING AND SECURITY OF DATA

- Every effort will be made to ensure that your privacy and confidentiality is protected throughout the study.
- Data will be kept for a maximum of 5 years and then destroyed by the researchers.

Your participation in this study is voluntary. Please remember that you may choose to withdraw from the study at any time and may exercise the option of removing your data from the study.

If you agree to participate, please fill in the consent section below. You will be given a copy of this letter for your reference.

Maria Desira

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CONSENT section

I have read and understood the above information and agree to participate in the research project as described above. I understand that my input, when published, will be anonymous and that I may decide to stop participating at any stage.

Name and surname _____ Date _____ Signature _____

Maria Desira, Master of Arts (Sociology) Student & Research Support Officer

_____ (Signature)

Formola ta' kunsens (intervista mal-esperti):

Inti mistieden/ mistiedna biex tipparteċipa fi studju li qed isir biex jesplora l-fehmiet, it-twemmin u l-attitudnijiet dwar il-proċess tal-*biobanking* u riċerka ġenomika. Din ir-riċerka qed issir mill-istudenta tal-Master of Arts (Soċjoloġija) u Research Support Officer Ms Maria Desira, taħt is-supervizjoni ta' Dr Gillian Martin, li hija akkademika residenti fid-Dipartiment tas-Soċjoloġija fil-Fakultà tal-Arti. L-għan ta' din ir-riċeka huwa li biex nifhmu iżjed kif in-nies jaħsbuha dwar is-sehem jew il-kollaborazzjoni tagħhom f'riċerka ġenetika, u dwar li jagħtu l-kampjuni tagħhom lil *biobanks*, u anki dwar kif inthom l-esperti taħsbuha dwar dan.

M'hemm l-ebda riskju assoċjat ma' li tipparteċipa f'din ir-riċerka. L-informazzjoni li tipprovdi se tgħinna nifhmu kif ikun l-aħjar mod biex jinħolqu *policies* dwar l-immaniġjar ta' *biobanks* li jkunu aktar fdati mill-poplu u li jiffaċiltaw u jżidu l-partiċipazzjoni fuq skala nazzjonali. Din se tgħinna wkoll biex niddisinjaw u nipproduċu filmati edukattivi li jkunu parti mis-sit tal-I.T li bħalissa qed tiġi ddisinjata użata mir-riċerkaturi fiċ-Ċentru, il-parteċipanti u l-pubbliku ġenerali.

Jekk inti tiddeċiedi li tipparteċipa, inti se tieħu sehem b'mod profond f'intervista semistruttura li tesplora *issues* relatati mal-ħidma tiegħek fis-settur ta' riċerka genomika u *biobanking*. L-aktar dwar kwistjonijiet relatati mad-drittijiet, il-fiduċja, ir-riskju, il-kunsens, it-tqassim tal-informazzjoni tal-parteċipant ma' sorsi terzi.

L-Anonimità

L-intervista se tiġi rrekordjata bil-permess tiegħek, però inti se tiġi assenjat isem fitizzju. Dan se jintuża b'rabta mal-informazzjoni tiegħek minn dak il-ħin 'il quddiem. B'dan il-mod, l-informazzjoni li tinġabar matul l-intervista se tkun ikkodiċizzata. Mhux se jkun possibbli li wieħed jassoċja dak li inti tgħid mal-identità personali tiegħek, la waqt il-proċess u lanqas fil-pubblikazzjoni tar-riżultati tar-riċerka. L-informazzjoni tiegħek se tkun ikkodiċizzata sabiex ir-riċerkaturi biss ikunu jistgħu jorbtu l-kummenti tiegħek jew l-informazzjoni mal-isem. L-informazzjoni kollha li tagħti inti, u li tiġi ippreżentata f'rapporti, fi preżentazzjonijiet u/ jew f'kitbiet oħrajn, se tkun ippreżentata b'mod li ħadd ma jkun jista' jidentifikak mill-kummenti u l-informazzjoni tiegħek.

Il-Kunfidenzjalità

Se jsir kull sforz biex nassiguraw l-kunfidenzjalità ta' kull informazzjoni miġbura f'dan l-istudju:

- Ir-*recordings* se jinżammu f'kompjuter li għandu *password* protetta u r-rekordings oriġinali se jiġu mħassra.
- It-traskrizzjonijiet kollha se jiġu maħżuna f'kompjuter li għandu *password*.
- Ir-riċerkaturi Maria Desira u Dr Gillian Martin biss se jkollhom aċċess għall-informazzjoni li tidentifika l-parteċipant.

Is-sigurtà tal-informazzjoni

- Se jsir kull sforz biex nassiguraw li l-privatezza u l-kunfidenzjalità tkun protetta matul l-istudju.
- L-informazzjoni se tinżamm għall-massimu ta' ħames snin u tinqered mir-riċerkaturi kif jgħaddi dan il-perjodu.

Il-parteċipazzjoni tiegħek issir b'mod voluntarju. Jekk inti tiddeċieda li ma tiħux sehem, id-deċiżjoni tiegħek mhux se tħalli impatti fuqek, bl-ebda mod. Jekk tiddeċiedi li tieħu sehem, jekk jogħġbok ftakar li tista' tieqaf tieħu sehem f'dan l-istudju f'kwalunke ħin u tista' tagħżel li titħassar l-informazzjoni kollha li tkun tajt għal dan l-istudju.

Jekk inti taqbel li tipparteċipa, jekk jogħġbok imla s-sezzjoni ta' hawn taħt u li biha qed tagħti l-kunsens. Inti se tingħata kopja ta' din l-ittra għar-referenza tiegħek.

Maria Desira

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Sezzjoni ta' Kunsens

Jiena qrajt u fhimt l-informazzjoni t'hawn fuq u naqbel li nipparteċipa f'din ir-riċerka kif deskritta hawn fuq. Jiena nifhem li l-informazzjoni li tajt, meta tiġi ppubblikata, se tinżamm anonima u jiena nista' nieqaf nipparteċipa f'kwalunkwe stadju.

Isem u kunjom _____ Data _____ Firma _____

Maria Desira, Studenta Master of Arts (Sociology) u Research Support Officer

_____ (Firma)

Appendix J - Expert interview guide

What is your particular interest in the issue of genetic research. [describe professional role]

How long have you been involved in this area of research? [how have things changed?]

What would you say is your key motivation to work in this area? [science? Common good? Health of future generations? Personal satisfaction?]

Who do you think benefits most from your research? [patients? Families? General public? Academia?]

Do you think there are any worrying issues related to participants donating blood or tissues to the Malta biobank? [re participants privacy? Personal data? Genetic information? Data sharing?]

Do you feel there is a particular service that is lacking locally, in relation to supporting participants in genomic research?[if yes - what is it? How do you get by without it at the moment?]

Do you currently use the Biobank at the CMMB for your research? [why? Why not? If no, might you in the future?]

What do you feel about the consenting procedure currently in use? [what is it? Can it be improved? Dynamic consent?]

What do you think about the issue of dealing with incidental findings? [what do you currently do in practice? Is this entirely satisfactory? Why? Why not?]

What about the future? [future research aims? Future for samples? Future funding?]

Is there anything we have missed here?

Punti ta' gwida għall-intervisti mal-esperti:

X'inhu l-interess partikolari tiegħek fejn tidħol ir-riċerka ġenetika. [iddeskrivi l-irwol professjonli]

Kemm ilek involut f'din it-tip ta' riċerka? [kif inbiddlu l-affarijiet?]

X'tgħid li hi l-akbar motivazzjoni biex taħdem f'dan is-settur? [ix-xjenza? biex tgħin lis-soċjeta'? is-saħħa tal-ġenerazzjonijiet futuri? sodisfazzjon personali?]

Min l-aktar li jibbenifika mir-riċerka tiegħek? [pazjenti? familji? pubbliku ġenerali? akademija?]

Taħseb li jistgħu jeżistu problemi meta persuni jagħtu d-demm tagħhom, jew *tissues* oħrajn, lil biobank ta' Malta? [dwar il-privatezza? informazzjoni personali? informazzjoni ġenetika? qsim ta' informazzjoni ma' riċerkaturi terzi?]

Taħseb li hemm servizz partikolari li f'Malta ma jeżistix, fejn jidħol is-sapport għal min jipparteċipa f'riċkera ġenomika? [jekk iva-x'inhu? Kif qed tgħaddu mingħajr dan is-servizz bħalissa?]

Bħalissa qed tuża il-biobank ta' CMMBB għar-riċerka tiegħek? [għaliex? għaliex le? jekk le, hemm ċans li tużah fil-futur?]

X'taħseb u xi tħoss dwar is-sistema' ta' kunsens li qed tintuża bħalissa? [x'inhi? tista' titjieb? kunsens dinamiku?]

X'taħseb dwar l-*issue* ta' x'għandek tagħmel b'informazzjoni sekondarja li tirriżulta waqt il-proċess tar-riċerka? [bħalissa x'tagħmel meta jiġri hekk? taħseb li dan hu kompletament tajjeb? għaliex? għaliex le?]

X'taħseb dwar il-futur? [għanijiet ta' riċerka għal futur? il-futur tal-kampjuni? *Funding* tal-futur?]

Hemm xi ħaġa oħra x'iżid? Ħallejna xi ħaġa ta' importanza barra?