

Professor Joseph Borg

B.Sc.(Hons)(Melit.),M.Sc.(Melit.),Ph.D.(Melit.)

Science Odyssey

It is indeed a great honor for me to be here. As I stand right here, I cannot ignore, nor forget that I was filling the exact same place you are now not so many years ago.

I am a scientist. What defines a scientist? In all areas of studies and health care professions, what defines us is our high-quality work ethos and drive to discover something new.

We should be proud to live in a country that, despite its size, has managed to produce innovation that surpasses that from much larger countries: I am, here, not talking about scientific innovation, but am referring to events in our history such as the building of the oldest free-standing Megalithic Temples¹ which still stand proudly to this day. I shall not delve into the genetics of the temple builders of Malta; however even if not much heredity has been bequeathed to the present generation, surely, the physical masterpieces which are still extant must inspire today's island citizens as well as visitors to Malta who witness these amazing feats.

Now we live in a different time. A time when personalized medicine and tailor-made therapy are available, albeit at some considerable cost (for now). A time when we can predict diseases before a child is even born.

Almost 20 years have elapsed since the golden age of the human genome project². We have also just completed our very own Malta Human Genome Project thanks to funding by the Malta Council for Science and Technology. We are now in an era, where opportunity is broader but more demanding than it used to be. I can only recommend to you to take time to daydream, to work hard, and equally important, to try things out no matter how impractical you think they are, no matter how discouraged you might feel. You may be right in also thinking that the current and subsequent years of your early career appear to be the hardest. Please be mentally prepared for some degree of chaos and failure. Many a time you will feel frustrated, or about to give up all together, however, these moments of near-despair can very well prepare you to become adapted to challenges that may lie ahead.

I was absorbed by the beauty of molecular cell biology; the intricate networks and biochemical pathways that occur inside a living cell, enabling it to divide, grow, and die in a programmed manner. My subsequent years of study, at both a Master's and Ph.D. level, were done at the University of Malta, at the Laboratory of Molecular Genetics, now part of the Centre for Molecular Medicine and Bio Banking, that brings together the Faculty of Health Sciences, Medicine and Surgery and Science. My research question was to uncover the cause as to why certain members of a Maltese family exhibited very high levels of foetal haemoglobin without showing any signs or symptoms of blood complications synonymous with Thalassaemia, a word which is based on the Greek roots for blood and the sea. I was drawn to experimental haematology and genetics in the first months of my post-graduate studies. These became my area of specialization and I no longer felt a novice; so much so that I naïvely believed that I could answer my research question in a short time. So I took up the challenge a step further and worked on the research question as my Ph.D. project.

What happened next, in the span of a few years, was nothing short of amazing. It was, for me, a science odyssey, that I don't hesitate one bit recommending to any of you sitting here tonight. The break in all this, came rather unexpectedly, whilst I was visiting a very small and remote town in Forli-Cesena, Italy. I was attending a small course on Thalassaemia. One night, I was given the opportunity to take my research question to the Erasmus Medical Center, in Rotterdam, Holland. There I could make use of equipment that was and still isn't readily available at our University. However, this was an excellent opportunity to further my skills and knowledge abroad and bringing back all this to Malta. The research exchange was so productive, that I travelled more frequently, each time adding more and more bits of the puzzle to understand the intricate control of haemoglobin and how we can devise ways for the treatment of thalassaemia. The "Eureka Moment" came when I discovered a new DNA mutation in a gene, known as *KLF1*, that acts as a master regulator of blood erythropoiesis.³

Fast forward a few months, and the group I worked with in Malta, alongside our collaborators from Holland, published two front cover papers in Nature Genetics.^{4,5} I am, but one example of a student turned scientist and later an academic member of this prestigious University. I predict that many other success stories are waiting to be written by several of you present here tonight, students, especially at the postgraduate level, embark on the challenges which academic supervisors set for them, and once that cocktail works, the magic of achievement can happen and excellent research papers in journals may flow.

Malta is now a member state of many high-level scientific Organizations, including the European Molecular Biology Laboratory (EMBL), making our local scientific enterprise part of a much bigger effort that can lead

more effectively to major scientific breakthroughs. But innovation is not just about competitiveness and economic growth. As we consider the big challenges facing societies today – climate change, inclusiveness, energy, health and ageing, just to mention a few. Innovation is at the heart of our ability to find solutions that are economically, socially, and environmentally sustainable. The focus on gender equality, and encouraging participation of women in research, as well as initiatives such as increasing funding and not only towards research in science, but also that of instilling in younger generations the passion for discovery and the quest for more knowledge. Research and Education go hand in hand. That is why you are here tonight, it is your ambition and thirst to know more. This will entice you to further your studies, like I've done and many others you look up too in the academic sector. Finally, I must mention how crucial and important it is, to work in teams, where each individual contributes the expertise necessary. I am now in a position to not only collaborate with Faculty departments, but also in between different Faculties such as in my recent experience with the group at the Institute of Space Sciences and Astronomy. We are combining elements of Molecular Biology and state of the art next generation DNA technology with astrophysics and laws of the Universe. Trying to uncover how harsh and extreme environments on Earth can sustain life and at times, even thrive better and how this in turn can help us understand better our health and life in the context of medical science.

I am proud to be working in a scientific field contributing to advances in genomics and applied biomedical science. The implications of this go beyond finding a cure for diseases such as cancer, but to also help to break barriers of stigma in scenarios such as mental health. This helps fight ignorance with sound knowledge, replace fear with reason and offer an explanation where there was no hope.

To conclude, I am sure that all branches of science and research are rapidly progressing with evolving times. The scientist we know, is no longer the one wearing a white lab coat and handling test tubes, but are present in all spheres of life across all continents. I end with a final quote by Carl Sagan, an American astronomer and astroscientist who said:

“We can judge our progress by the courage of our questions and the depth of our answers, our willingness to embrace what is true rather than what feels good.”

¹The Prehistoric Archaeology of the Temples of Malta. Bradshawfoundation.com. Retrieved on 2018-10-15

² International Human Genome Sequencing Consortium. Initial sequencing and analysis of the human genome. Nature 409, 860-921 (2001)

³ Miller, I.J., & Bieker, J.J. A novel erythroid cell-specific murine transcription factor that binds to the CaCCC element and is related to the Kruppel family of nuclear protein. Molecular Cell Biology, 13(5), 2776-2786 (1993)

⁴ Borg, J., Papadopoulos, P., Georgitsi, M., Gutiérrez, L., Grech, G., Fanis, P., et al. Haploinsufficiency for the erythroid transcription factor KLF1 causes hereditary persistence of fetal hemoglobin. Nature Genetics, 42(9), 801 (2010)

⁵ Giardine, B., Borg, J., Higgs, D. R., Peterson, K. R., Philipsen, S., Maglott, D., et al. Systematic documentation and analysis of human genetic variation in hemoglobinopathies using the microattribution approach. Nature Genetics, 43(4), 295 (2011)