How will personalised medicine change public health practice?

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Educational aims

- To increase awareness about the growing applications of personalised medicine
- To appreciate the tensions between public health practice and personalised medicine
- To gain an understanding of the manner in which personalised medicine can act as a bridge between public health practice and clinical medicine

Key words
Personalised medicine, public health practice, public health genomics, prevention, screening.

Abstract

Personalised medicine is challenging core elements of public health practice to bring about a paradigm shift. Traditional public health activities such as prevention, screening programmes, infectious diseases control, financing and planning of health systems will all be affected by developments in genomics. There is a need to move away from the traditional high-risk versus population approach debate and to engage with concepts of population stratification and public health genomics. Public health through its activities of surveillance, needs assessment, education and policy advocacy has a critical role to play in shaping the entry of personalised medicine into health systems.

Introduction

This article explores how personalised medicine is challenging core elements of public health practice to bring about a paradigm shift in the organisation of public health services and health care systems.

Personalised medicine is defined as "an emerging practice of medicine that uses an individual's genetic profile to guide decisions made in regard to the prevention, diagnosis and treatment of disease".[1] Personalised medicine is increasingly being used to develop custom-tailored individualised treatments in the field of oncology and rare diseases but also raises hopes of successful treatments for common illnesses such as cardiovascular disease, diabetes and mental disorders. A closely related approach known as 'stratified medicine' allows decisions to be made for sub groups of people depending on their genetic risk profile. Such concepts are increasingly being portrayed as the future for clinical diagnostic and therapeutic medicine.[2] However some authors have raised ethical issues[3] and others have called for personalised medicine to move away from focussing on targeted treatment for specific organ disease and to embrace personalised medicine as the holistic treatment of the whole person.[4,5] The fact that personalised medicine and stratified medicine have emerged from a focus on the genetic material of individuals or sub groups of people sharing the same characteristics may mistakenly lead one to assume that they bear no relevance to public health practice. Indeed, the World Health Organisation defines public health as "all organised measures (whether public or private) to prevent disease, promote health and prolong life among the population as a whole. Its activities aim to provide conditions in which people can be healthy and focus on entire populations, not on individual patients or diseases". Should public health practice be concerned with personalised medicine when the scope of activities for public health is typically at population level, while personalised medicine deals with individuals? Some public health pioneers recognised the importance of engaging with genetics and genomics early on.[6] Now, as genomics starts to develop a role in common chronic illness prevention and treatment, there is a need for contributions from public health to become more visible.[7] Public health practice will not

be able to ignore the impact of genomics and will need to take into account the concepts of population stratification.[8]

Discussion

Prevention strategies

The debate on the merits of the individual high-risk versus population-based approaches that characterised preventive epidemiology in the second half of the 20th century[9] has returned in full force, fuelled by the advances being made in genetics and genomics. It has been proposed that the traditional one-size-fits-all approach to disease prevention, diagnosis, and treatment would be progressively replaced by a more individualized and tailor-made approach.[10]

Tobacco prevention and control strategies provide an interesting example. In no other policy area has public health been so effective in advocating for an approach that is based on strong regulation with measures that aim to protect the whole population such as taxation and smoke free public places. Genetic and neuroscience research continues to enhance our understanding of addiction and tobacco dependence yet it is not known how best to integrate genetic information about a complex phenomenon like smoking into traditional public health population-based approaches. An empirical study of stakeholder perspectives found that whilst public health approaches remain the preferred vehicle for tackling tobacco, individualised treatment programmes through pharmacogenomics were viewed as useful complementary mechanisms to support individuals provided that sufficient evidence about their effectiveness becomes available.[11] Unsurprisingly, this study found that clinicians were far more open to the possibility of using genetic information to underpin tobacco prevention and control strategies than public health practitioners.

The way in which knowledge of genetic predisposition can affect attitudes to prevention is an important factor for public health strategies. A study amongst persons with Type 2 diabetes found that persons felt less responsible for their Type 2 diabetes if they received information about their genetic predisposition and this information also affected attitudes towards prevention.[12] This is relatively unexplored territory and has important implications for all persons involved in preventive work.

Screening programmes

The genetic basis for public health screening programmes is associated with neonatal screening. Developments in the genomics field are expected to shape the future of other screening programmes in areas such as cancer and familial hypercholesterolaemia.[13]

Cancer screening programmes are an important, if somewhat controversial, area of public health practice. While population-based programmes are commonly available in many countries for breast, cervical and colorectal cancer, the developments taking place in the area of genomics could allow screening for cancers not previously feasible. One such example is ovarian cancer, where genetic testing for germline mutations associated with higher risk is becoming increasingly affordable and offers an opportunity to identify higher risk women irrespective of known family history. Screening for familial breast cancer in younger women than those traditionally targeted in population based screening programmes is another key development. There is some evidence to indicate that women would be ready to participate in such screening programmes.[14] Stratified screening based on genetic testing is a new approach to prevention. Various organisational issues would need to be considered before it could be introduced. Potential issues that arise and would need to be addressed include how the offer of screening would be made, making sure consent is adequately informed, how individuals' risk would be assessed, the age at which risk estimation should occur, and the potential use of genetic data for other purposes.[15] Inter-country differences in the genetic profiles could also provide an explanation for the varying success rates between population based screening programmes and would need to be taken into account when determining the feasibility of establishing stratified screening programmes.

Infectious diseases

The 2009 influenza pandemic exhibited a wide spectrum of disease ranging from very mild to fatal. Traditional factors such as age, comorbidities and being immunocompromised failed to explain the variations observed. Several lines of evidence suggest that different populations have disparate degrees of susceptibility and that host variation in key genes associated with the appropriate immune response could play an important role in determining the outcome of infection. Genomics applied to infectious disease epidemiology, prevention and control has important implications to identify which populations or subgroups may be at highest risk of severe infection and target limited amount of countermeasures or vaccine to those at highest risk.[16]

Funding innovative treatment

The use of genotyping to predict outcomes and determine the most appropriate candidates for treatment has been well established in the field of oncology. More recently expensive treatment for hepatitis C can be carefully planned through the use of genotype testing to predict outcomes.[17] Such applications are important in the management of health systems that have to deal with competing demands on highly limited resources for the financing of innovative therapies. There is a need for public health tools such as health technology assessment to be adapted for evaluations on effectiveness of personalised medicine early on in the development stage.[18] Finer targeting of treatment for persons who truly demonstrate a capacity to benefit may radically alter fundamental public health concepts such as 'numbers needed to treat'. The call for new methods of financing personalised medicines requires strong public health stewardship.[19]

Conclusion

Balancing the aspirations for personalised medicine with public health approaches in fiscally constrained health systems will not be an easy task but could be facilitated through a strong medicine – public health partnership.[20] The extent to which genomics will change public health practice depends on the willingness of the public health community to embrace genomics. The areas of practice within the public health impact pyramid developed by Frieden[21] provide an alternative manner of envisioning the positioning of public health genomics[13] that goes beyond the dichotomous high-risk versus population approach which is unhelpful to take

forward the relationship between public health and personalised medicine. The scope of public health practice is sufficiently wide such that within it, it is possible to identify sectors such as screening, where public health genomics is already challenging and modifying the population-based approach. In other sectors such as tobacco control, personalised medicine at present does not realistically present feasible options to overturn established models of prevention and control. Public health practitioners have an important role to play at multiple levels in influencing the development and uptake of personalised medicine. The need to ensure that public health genomics and personalised medicine finds its way into public health teaching, research and practice cannot be underestimated if public health is to evolve to meet the needs and challenges facing society in the 21st century. Equally public health through its activities of surveillance, needs assessment, education and policy advocacy has a critical role to play in shaping the entry of personalised medicine into health systems such that the basic values of equity, utility and efficiency, underpinning welfare-based health systems, prevail. Indeed it is possible that the introduction of personalised medicine into health systems may become the bridge across the often separate worlds of medicine and public health.

Key points

- Personalised medicine is challenging core elements of public health practice.
- Prevention, screening and infectious diseases activities may need to take population stratification into account.
- Public health is not antagonistic to personalised medicine but has an important role to play in shaping the entry of personalised medicine into health systems.
- Personalised medicine may become the bridge between the specialities of medicine and public health.

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