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Keynote Report:

Genomics and world health: hopes and realities*

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Abstract: Due to lack of appreciation of the complexities of the interactions between nature and nurture, claims for the rapid improvements in medical care following the human genome project have been exaggerated. Although some progress has been made in certain fields, the full scope of genomic medicine may not be realised for many years.

Key words: Genomics, Global health, Genetic disease

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The completion of the human genome project was accompanied by quite remarkable claims about the benefits for health that would follow this outstanding scientific endeavour. In this short article, I shall try to put some of these claims into perspective. Currently, the poorer countries of the world are still suffering from the effects of malnutrition, ineffective healthcare systems, and the scourges of tuberculosis, malaria, and HIV/AIDS. The richer countries are attempting to deal with the costs of caring for the chronic diseases of middle and old age and of the problems arising from an increasingly aged population. While there is no doubt that a better understanding of the mechanisms of the human genome will play a role in helping to combat all these diseases, it is quite clear that claims that this information will produce a major change in the pattern of healthcare in the near future have been greatly overstated, largely because of the multi-layered complexity of human disease. The full benefits of the human genome project for human health will only come to fruition by the close interaction of clinical epidemiology and population genetics combined with a revitalisation of clinical research to dissect the complex interactions between the genome and environment that are responsible for the extremely complex phenotypes of all human diseases. These principles were illustrated

in a recent report from the World Health Organization (WHO, 2002). Currently, the application of genetic technology for healthcare is most advanced in the cases of the diagnosis and control of monogenic diseases, notably the inherited haemoglobin disorders, and communicable disease. For the latter, applications include the identification of new pathogens, the diagnosis of conditions in which organisms are difficult to grow or identify rapidly by standard techniques, the identification of drug resistant organisms, and, in the future, from the discovery of new drug targets or the development of vaccines. Genetic medicine is also making major progress for the diagnosis and management of certain forms of cancer. Applications for other common diseases in which there is a major environmental component, heart disease, stroke, and diabetes for example, much more work is required to identify some of the genes involved in the multigenic systems before the clinical relevance will become apparent.

Some of the approaches to the application of genomic technology for medical care, particularly in the developing countries, were also outlined in the World Health Organization report and by Weatherall (2003).

References

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