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New Paradigms in Medical Genetics

In the past few decades, medical genetics has been established as a major discipline in medicine in many parts of the world. The contributing factors behind this move included changes in the epidemiological pattern of diseases, emphasis on disease prevention and health promotion, and major advances in science and technology. Regarding epidemiological changes, it has been well documented that the contribution of infectious diseases to morbidity and mortality in childhood has greatly declined. Birth defects, in the form of inborn errors of morphogenesis or metabolism, however, have become major factors in disease causation in this age group. Furthermore, genetic determinants of diseases continue to affect our health well beyond childhood. This is exemplified by the fact that most of the commonest contributors to mortality in Hong Kong have one or more genetic determinants. These include cancers, cardiovascular and cerebrovascular diseases, diabetes mellitus, mental disorders and susceptibility to infections. Coupled with this epidemiological change, healthcare providers became increasingly aware of the important role of prevention of diseases and promotion of health in the modern healthcare system. Besides its contribution to decreasing suffering in individuals, this approach helps to improve the general well being of the whole population. Under this approach, medical genetics has much to offer as a major medical discipline. Equipped with advances in genetic epidemiology, cytogenetics, biochemical genetics, and more recently, molecular diagnostics, medical geneticists can now provide a wide spectrum of services to prevent genetic diseases and promote genetic health in the population. These services include genetic diagnosis and counselling, carrier detection, prenatal diagnosis, predictive genetic testing, and genetic screening at different phases of life. In this issue of Hong Kong Journal of Paediatrics (Issue), we witness how recent genetic advances have been put to good use in elucidating the genetic factors behind a number of birth defects. Most of these reports are the first of its kind describing these conditions among Chinese. They serve as examples of how far Hong Kong has progressed in medical genetics in the past quarter of century.

Of recent, though, we have witnessed a quickening of the pace of progress in medical genetics. Two major driving forces are at work here, one technological and the other social. The technological advance is the spectacular early completion of the Human Genome Project in 2003. It

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promised to offer an important basis to decipher how our genes are coded, how they would function individually and in concert with other genes in a bigger environmental context. The subsequent HapMap Project added further information towards our understanding of these issues. It is envisaged that further progress in genomics will shed more light on the causation of diseases, be it rare genetic conditions which usually land at clinical genetic centers or common diseases encountered by most healthcare workers. As for the social forces behind our progress in medical genetics, one should take note of the increasing concern of the public in any health related topic, and in particular the interaction of genetic determinants of health and environmental factors. Advocacy from the public and professional groups have helped government to shape our healthcare policies.

The result is that we are witnessing four obvious paradigm shifts in medical genetics. First of all, there is a growing demand on the emphasis of genetic research and services for common conditions affecting our population. Traditionally, studies of rare conditions have contributed greatly to our understanding of disease pathogenesis. For the past few decades, limitations in technology posed certain restrictions on researches in the genetic basis of common and complex diseases. However, with the advent of genome studies and bioinformatics, the path is now opened. In the foreseeable future, much more information will be available for advancing the treatment and prevention of these conditions. This shift of emphasis will in turn lead to our second major paradigm change, which can be paraphrased as 'from bed-side to the community'. While rare conditions are mostly attended to in the hospital setting, where a strictly clinically approach has to be adopted, the changing emphasis on common, and perhaps more chronic, conditions means that genetic practices would have to adopt a more community sort of orientation. In the genetic context, one speaks of a shift of emphasis from clinical genetics to community genetics, or public health genetics, as it is more recently termed. The promotion of genetic health will find more footing in a community setting. Prevention of genetic diseases will be grounded in the mass. Yet, this shift in emphasis does not negate the necessity for personal patient care. If anything, recent genetic advances allow for greater potential in curative care. Developments in genetics, genomics and cell biology have been put to good use in saving the lives of thousands of previously untreatable patients. The use of enzyme replacement therapy and organ or tissue transplantation for genetic diseases are good examples. Research in gene therapy may point to further areas of therapeutic development. In addition, the potential of pharmacogenetics with its promises of personalised medicine probably at the primary healthcare care level will be a very interesting area of medical development. This can be considered the third major shift of paradigms of medical genetics, and it can be best coined as 'from prevention to cure'. Medical geneticists are increasingly finding themselves associated with the development of therapeutic agents or modalities for the treatment of patients.

Hence, on top of their traditional role in prevention of genetic diseases and promotion of genetic health, they are partaking in therapeutic and curative management of patients. Medical genetics has indeed moved through a full circle of development. This leads us to the fourth paradigm shift. As we move along the rim of the developmental cycle of medical genetics outlined above, one can easily be impressed by the fact that health and diseases are not wholly determined by genetic factors, as the previous studies in rare 'single gene diseases' led us to believe. Neither are they wholly determined by environmental factors, as the term 'lifestyle diseases' construe. It involves an interplay of both genetic and environmental forces, with varying contributions in different situations, much like the pictorial presentation of Tai Qi. Graphically, the logic of genetic determinism can be represented as a straight line from one point of

determinant leading to an endpoint in health or disease. Hence, this fourth paradigm shift in medical genetics can be rightly coined 'from linearity to Tai Qi'. This philosophical concept of development in medical genetics demands a different approach to its practice in a healthcare setting. Research and healthcare need to pay full attention to the interplay between genetics and environment. The latter, of course, include lifestyle, social, cultural, ethical and institutional determinants of health. Besides understanding genomics, we need to equip ourselves with insights of humanity. It is timely that this Issue carries messages on ethics, information disclosure and genetic knowledge.

STS LAM
Guest Editor