# THE SOUTHEAST ASIAN JOURNAL OF TROPICAL MEDICINE AND PUBLIC HEALTH

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# EDITORIAL

## HUMAN GENETIC DISEASE IN ASIA

On July 27-29 1994 in Bangkok was held the first Asia-Pacific Conference on Medical Genetics. In a field that has exploded over the past four decades this may seem to be a very late start, yet it is, in reality, an important landmark at this time in history in the region with the world's fastest economic growth.

Infectious disease has been the major focus of attention in Asia during those forty years, for good reason: viral, bacterial, parasitic, fungal diseases represented the most urgent threat in terms of potentially preventable mortality and morbidity where decisive action could make a difference. Indeed, communicable disease programs in the region have had substantial impact, particularly in early childhood, so that in many countries life expectancy has risen markedly, as infant mortality has dropped. While infectious diseases still represent a major burden, other problems are rising in perceived importance, their relevance recognized as economic performance increases.

While accidents, vascular disease and cancer figure more prominently with rising affluence, the genetic component of a wide range of diseases is now the focus of rapt attention as the result of a wave of technological advances that have changed the face of medicine: the genetic components of a growing number of human diseases are being discovered in quick succession. The precision with which mutations of medical importance have been and are being defined, and the rate at which the consequent data banks are growing provides not just the scientific rationale for satisfying curiosity but the opportunity for concerted action of a positive kind in this broad geo-political arena where economic growth curves dominate.

What this meeting did was to bring forward information of the widespread activity already extant in human genetics in many countries in the Asia-Pacific region. This will be evident in detail in the proceedings to be published as a supplement by this journal. A great deal has been achieved over these past decades in many countries and there was evidence at the meeting both of growing capacity to handle the diagnosis of a range of genetic diseases and concern about the cost-benefit of achieving this capability where there would appear to be limited opportunity for productive intervention.

Indeed, there are in the region widely differing approaches to the underlying ethical issues involved, reflecting the enormous diversity of cultures and traditions from country to country. It was perhaps possible to discern a small element of uncertainly about the justification of embracing the technical elegance that molecular genetics has brought to pass, when knowing the mutant gene sequence often cannot be followed up by appropriate constructive action. Against this, being able to offer precise diagnosis at this level brings its own potential satisfaction to parents of young children whose illness would otherwise remain in the vexing basket of uncertainty: arguably for some this alone merits investment in the technology. However, there may be justification in the caution, especially where application of the technology leads to severe strains on human and fiscal resources.

The meeting highlighted some areas of human genetics where high gene frequencies bring the clinical consequences to the forefront of public health challenges. Globin chain gene mutations, manifected clinically as thalassemias and hemoglobinopathies, fall into this category, with their distribution across many countries in the region, collectively representing a sizeable disease burden and a substantial economic cost both to individual families and to society as a whole. More than 60 different mutations giving rise to thalassemia syndromes have been categorized in the region, some occurring in high frequency clusters in population groups with singularly long cultural histories. How to approach this broad problem in economically viable fashion is moving beyond the drawing board into community assessment.

A spin-off from molecular investigation of genetic diseases on a regional basis is the insight given into demographic divergence of human populations over historical time throughout Asia. A number of papers addressed this question, providing a starting point for building more complete genetic distance maps in the future. While this may to the clinician seem to be somewhat tangential to the central issue of disease delineation, the potential contribution to broader questions concerning the genesis of cultural diversity is considerable and should be encouraged in the context of regional collaboration. Asia also is te repository of important archeological evidence concerning the antiquity of *Homo sapiens* and its evolutionary relationship with *Homo erectus*, an even wider historical debate also addressed in an erudite paper at the conference.

The meeting provided a venue to explore ways in which regionally concerted effort might be able to enhance individual country capacity to grapple with both a broad approach to genetic disease and to particular high frequency clusters such as the thalassemias. Regional networking was indeed initiated in the latter case, while agreement to move ahead with further human genetics collaboration as a whole in the region received strong support in the form of an offer by Indonesia to host the next meeting in 1995, coincident with that country's fiftieth anniversary celebration of independence. The current status of information technology provides for much more effective networking than would have been possible just a couple years ago, another reason for the opportune timing of this meeting. The need to identify complimentary diagnostic capabilities underscores the parallel need to pursue the goal of bringing genetic disease management within affordable economic constraints, goals to which regional collaboration and resource rationalization may be able to contribute substantially in the years ahead.

The major focus of most single gene diseases has always been on their effects in childhood, leading as they do to severe morbidity or to early mortality. Attention was also drawn, however, to the growing fund of knowledge about multigene diseases, common problems which have their major impact on older age groups. A casual glance at the leading causes of adult morbidity and mortality in the region reveals that this sphere of genetics contributes risk factors to many of these disease classes. With the aging population structure of countries such as Japan, China, Korea at this time, with others in close pursuit, investment in this large area of medical genetics will likely pay dividends in the decades ahead. Just at what rate attention thereto will be justified on grounds of public and private sector funding priorities remains to be seen. While many nations in the region harbor growing inequity in access of their populations to adequate basic health care, the justifiable rate of moving to this high ground may be rather slow: this is a matter perhaps for future discussion among the medical genetics gatherings in the region. What funding authorities may be willing or even enthusiastic to support will no doubt differ greatly from country to country. Realistically such support may not be forthcoming in the near future in many cases.

An important question is the direction that research in human genetics might take in future in the region. The region is large and is comprised of countries with greatly differing culture, religions and economic status. There are some inriguing puzzles waiting to be interpreted, such as sudden unexplained death syndrome, with its peak occurrence in northeast Thailand: just how definitive is the genetic component if any is as yet uncertain, but the suspicion in high. This is a case where unravelling may contribute to a more rational solution of what has become a significant problem among adults in their prime working period of life. The full extent of genetic components of cardiovascular disease in the region remain to be catalogued: these are likely to be similar in many respects to those in other regions of the world but their practical importance necessitates elucidation. The cancer profiles differ country by country; while these no doubt reflect the greatly differing food habits and environmental variations, again genetic components may well reflect gene pools containing mutations from antiquity that are pertinent to the particular disease patterns. There are many challenges, including those that fall into the special multidisciplinary field of molecular genetic epidemiology.

Perhaps the most important outcome of the meeting was the recognition it has given to the fine work of many groups which previously had too little contact with each other. If the networking planned on this occasion can be put in place in a proactive way, the greater interaction can lead to the genesis of both more productive basic research and applied public health operations. Not all countries in the region were represented; it is hoped that more will join in the fun of collaboration in the near future. A start has been made to address this exciting field in a more concerted manner on a regional basis and great productivity is expected over the next few years.

> Suthat Fucharoen Chev Kidson