Editorial

Community Genetics

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Community Genetics in Developing Countries

A.H. Bittles[a] H.S. Savithri[b] N. Appaji Rao[b]

[a]Centre for Human Genetics, Edith Cowan University, Perth, Australia; [b]Department of Biochemistry, Indian Institute of Science, Bangalore, India

To date, studies on human genetic disorders have mainly been conducted on populations living in developed countries, and the results reflect their contemporary disease profiles. An equivalent assessment of the impact of genetic disorders on the health of people in developing countries has not been possible, largely because of the ongoing toll imposed by major endemic and epidemic diseases. However, with improvements in general living standards and successful preventive campaigns to control infectious diseases in virtually all developing countries, the effects of genetic disorders have become increasingly obvious, especially where sophisticated diagnostic facilities have been established.

The genetic structure of populations plays a very significant role in the distribution patterns of specific inherited disorders, and developed and developing countries differ quite markedly in this respect. Following industrialisation in the middle of the 19th century, Western countries were characterised by widespread population movement from the countryside into the rapidly expanding towns and cities, and subsequent migration of large numbers of inhabitants to other parts of the globe. These large-scale population changes also led to the breakdown of historical local, regional and national boundaries, which in turn exerted a partial homogenising effect on the gene pool.

By comparison, in most developing countries religious and social divisions are still in place, epitomised by the clan, tribal and ethnic groupings found throughout Africa, Central and South America, the Middle East, central, south and south-east Asia. In India, Pakistan and Bangladesh, which collectively account for 21% of the world's population, marriage continues to be arranged within caste and *biraderi* boundaries that may date back some 3,000 years. In India alone, there are an estimated 50,000–60,000 separate endogamous communities [1]. Some 25% of the population of 1,033 million belong to one of the more than 1,600 scheduled tribes and castes that exist outside the Hindu caste system [2], and a further 130 million are Muslim. In effect, each of these groupings, whether Hindu caste or non-caste, Muslim, Christian, Buddhist, Sikh, Jain or Parsi, ranging in numbers from hundreds to many millions, represents a separate breeding pool. A similar situation also exists in much smaller countries, such as Laos, with 68 ethnic groups in a population of 5.5 million [3]. This means that disease mutations that have arisen, for instance, over the last 50–100 generations may be restricted and unique to individual ethnic groups, sub-castes, tribes or clans [4].

Superimposed on this complicated genetic patchwork, consanguineous marriage is strongly favoured by many

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Alan H. Bittles
Centre for Human Genetics, Edith Cowan University
100 Joondalup Drive
Perth, WA 6027 (Australia)
Tel. +61 8 9400 5623, Fax +61 8 9400 5851, E-Mail a.bittles@ecu.edu.au

major populations. In Pakistan and countries of the Middle East, over 50% of marital unions are intra-familial, most commonly contracted between first cousins [4, 5]. The re-emergence of consanguinity as a significant factor in clinical genetics can largely be ascribed to the migration of many millions of individuals and families during the latter half of the 20th century from countries with a strong tradition of close-kin marriage. However, the recent adoption of homozygosity mapping for rare recessive genes has also added to research interest in the topic.

The Symposium on Community Genetics in Developing Countries, held in the Indian Institute of Science, Bangalore, from January 16–18, 2002, was devised and organised against this background. The papers in this special issue of *Community Genetics* represent a broad selection of the topics presented, and they are intended as appropriate starting points for further detailed studies. The importance of understanding and accounting for inter- and intra-population divisions is specifically considered in a range of settings: the patterns and intensity of human migrations into and within south-west Asia (McElreavey and Quintana-Murci); the role of the HLA and other immune systems in determining differential predisposition to major infectious diseases (Pitchappan); constraints on the design and utility of vaccines (Mehra et al.); haplotype differences at the *FMR1* locus involved in fragile X syndrome (Thelma and Sharma); and the aetiology of common psychiatric diseases (Mukherjee et al.). The potential adverse influence of consanguineous marriage on health is illustrated by papers on infant and childhood mortality among Palestinian communities (Pedersen), as well as on the prevalence and types of ocular disease (Kumaramanickavel et al.) and cerebral lipidoses (Christopher and Nalini) in South India. An overview of the profile of genetic disorders within the Indian population is presented (Verma and Bijarnia), with information on major haematological disorders, including counselling, screening and prenatal diagnostic programmes (Mohanty et al.). The final two papers deal with particular aspects of genetic disorders in the UK Pakistani community, where consanguineous marriage is strongly preferred (Corry), and problems faced in providing genetic counselling in multi-ethnic communities (Karbani), which affect both developed and developing countries.

Two major topics of interest emerge. The first applies to both developed and developing countries and involves recognition of the co-existence of sub-populations with differing disease susceptibilities and profiles. In Western countries, for example, there has been a tendency to broadly refer to individuals as being of south Asian or Maghrebian origin, which effectively ignores the very marked genetic subdivisions that exist within these regional categories. Likewise, although data are routinely collected on regional origins or consanguinity status in many developing countries, only limited attention has been paid to genetic differences between ethnic groups or specific communities. The second area that merits future action is mutual transfer of health-based information between developed and developing countries. Developing countries could immediately benefit from access to the voluminous data collected and stored in Western countries on resident migrant communities from Asia, Africa and South America. While substantial ethical and legal difficulties may need to be overcome, the potential benefits to developed countries of detailed information on inherited disorders that occur in their populations and sub-populations would be highly cost-effective, and greatly assist in focusing and accelerating the planning of future programmes of health care provision.

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