

Our human genome — how can it serve us well?

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Good public health, like all good medicine, rests on empirical data of high quality. If we have records of the incidence of disease, and of how this responds to interventions, we will be able to learn the best approaches for each community. Nowhere is this more true than for genetic diseases, where medical knowledge is increasing rapidly and personal issues, cultural differences and ethical dilemmas abound.

Until recently, it was possible to dismiss genetics as an issue affecting a few children in rich countries. Genetics looked unimportant compared to infectious diseases, high infant mortality and lack of proper sanitation. While those problems have not gone away, there are many countries (including India and China, the two largest in population) in which most people face the same medical issues as in “first world” countries — cancer, heart disease, psychiatric disorders, diabetes, and Alzheimer’s disease.

With this shift, the burden of genetic disease has changed dramatically. The adult diseases listed above all have a large genetic component, often due to changes in more than one gene, which interacts with the environment in susceptible individuals. Even the consequences of cigarette smoking, one of the most serious public health issues in developing countries, are not uniform but vary with genetic susceptibility. Greater knowledge of genetic factors that lead to high risk, which must be acquired for each ethnic group separately, will help public health planning and the targeting of interventions where they are most needed.

Among children, once a country has lowered mortality and morbidity due to poverty, genetic diseases due to single gene mutations such as haemoglobinopathies, cystic fibrosis and muscular dystrophy consume a large proportion of paediatric resources. Of these diseases, the haemoglobinopathies (sickle cell anaemia and thalassaemia) are by far the most important internationally.

There are about 200 million healthy carriers of thalassaemia or sickle cell anaemia. These carriers have a small added resistance to malaria in infancy, which is why the mutations causing haemoglobinopathies have spread throughout countries in tropical and sub-tropical regions where malaria is common. If two carriers have a child, it has a one in four chance of inheriting the mutation from both parents, causing a serious anaemia. Over 300 000 children each year are born with a severe haemoglobinopathy. With worldwide migration, these diseases are as much a feature of Europe, the United States and Australia as of the countries where they originated.

In this issue, Professor Bernadette Modell, who pioneered the reorientation of clinical genetics to public health and coined the term “community genetics”, puts forward evidence on the usefulness of genetic registers, in this case for thalassaemia in the United Kingdom (pp. 1006–1013). It is fascinating that even in the UK, where there is a highly organized national health system, the quality of the data depends upon the curator of the genetic register (“Personal contact is the key to success”). It is a pity that Professor Modell did not comment on this point, since it will be important when judging transferability to a country where transport may be poor and the phones may not work well. However, it is encouraging that in the UK there were no problems about confidentiality, and families were confident that the register would be used in a positive and not in a discriminatory way.

What of the ethical dimension? WHO is at last beginning to take a lead in offering policy on ethics of “the new genetics”, as can be seen from consultation statements on “Genomics and world health” and on “Ethical, legal and social implications of genomics” issued during 2001 by WHO’s Advisory Committee on Health Research. How do these statements fit with Professor Modell’s argument for better data registers?

Some of the most sensitive issues in genetics relate to the ethics of pregnancy choice. Some believe it is unethical for a country to push families towards screening, with the implication that an abortion may be a choice. Others believe it is equally unethical to deprive a couple of that

information and force them to have a child who will be seriously handicapped with a condition for which there may be no effective therapy. Choices by individual families can only be made if they are legal and the resources are there to provide the information needed. In the UK, where prenatal diagnosis is available, some ethnic groups used this option, while others did not. This would be completely appropriate provided that the choices were made by the couples themselves in the context of their beliefs and in good time, but unfortunately the UK study reveals that many couples who did not use prenatal testing were not offered it, which is surely a denial of their human rights to make their own decisions.

Now that the sequence of the human genome is available, it should be possible to ensure that every family has reproductive choice in relation to serious handicap. This does not in any way put down those with a handicap, any more than reducing the incidence of spina bifida by adding folate to bread reduces our determination to do our best for the smaller number of persons with a neural tube defect who are still born today.

WHO, as is appropriate, has recognized both the crucial importance of the understanding coming from the human genome project for human health, and the issues that arise from these data for world health in general and developing countries in particular. Much of genome science is low technology and could easily be used by every country were it not for the patenting of DNA sequences, an issue on which indigenous people are particularly sensitive as “their genomes” may give vital clues to susceptibility genes for common diseases such as diabetes.

Genes are about health. It is foolish to talk of cancer genes, or disease genes, or even thalassaemia genes. Most genes are healthy genes. They code for proteins and functions that allow us to survive and, usually, flourish. Their rich diversity ensures both our endless and wondrous variety as people, and our evolutionary survival. The human genome project helps us to know the power of the genome for humankind, for all our people, for now and the future. Appropriate data will allow us to harness that power better. ■

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