World Health Organization and Genetic Disorders

This critique of the World Health Organization report number 497 on *Genetic Disorders : Prevention, Treatment and Rehabilitation* is by Professor J. H. Edwards, of the Department of Human Genetics at the University of Birmingham

A COMMITTEE of cobblers, if asked to advise on the health of the world, would doubtless draw attention to the scandal of millions of naked feet and to the danger of foot-borne diseases, and advise on the need for counselling of the barefooted and for the training of both counsellors and cobblers.

This form of negative stability is by no means peculiar to health; beekeepers, chess-players, numismatists and pot-holers share an equal missionary zeal. Even solipsists, according to Bertrand Russell, complain of their rarity.

Medical geneticists, whose report was reviewed briefly in *Nature* (**238**, 7; 1972), might be thought to be free from this hazard by their training in the biases of ascertainment, but this is not so.

The report makes a number of recommendations which are neither surprising nor inappropriate. For reasons which are not clear, however, it contains a strange mixture of doubtful assumptions, erroneous facts, and uncertain inferences, some of which overflow into recommendations. The first paragraph speaks of "the relative increase in morbidity and mortality attributable to slow changes in gene frequencies". No explanation is given. There is said to be "no reliable information on the current prevalence of genetic disease"—in fact, the information is adequate for practical purposes; the numbers of the deaf, the blind, and the ineducable are necessarily known in all countries with compulsory education, and the proportions of these due to mendelian and chromosomal disorders have been estimated on good data.

The word genic is extended from its regular use to include multifactorially determined disorders, a term used variously. In the table on multifactorial disorders, congenital malformations and conditions of unknown and "non-genetic" disease are used as exclusive categories. In Table 2 a series of malformations, mostly of unknown aetiology, are called multifactorial, which admittedly means the same thing.

Later we are redeemed with robust and orderly descriptions of biochemistry until we again leave the ground in a cloud of numerals. On page 31 a mutation rate for recessives of 10^{-4} is "assumed". On the next page $100 \times 5 \times 10^{-5} \times 2 + 0.0001$ somehow makes 0.0086. On page 33, "genetic counselling, urging couples total abstinence from reproduction" is recommended in recessive traits, but, on page 34, we are almost all carriers—"every zygote is estimated to carry 1 or 2 deleterious recessive genes".

On page 43 the remarkable claim that fibrocystic disease can be diagnosed *in utero* is made. The claim that sickle cell disease can be diagnosed, which is also made, was hardly stronger when the committee met. On page 45 compound interest is used to provide the doubling time for "multifactorial disorders". The theory is difficult and no mention of the procedure is made, but certainly stabilizing mechanisms are likely to reduce this. The report ends on a most alarming graph, whose credentials are hardly as firm as the chronology of Archbishop Ussher, showing that the end of the world is at hand.

How, we may ask, can such an assembly of experts, with a dedicated and able secretariat, lead to the publication of a document so rich in platitude and inconsistency?

The reason is not far to seek: an assembly of experts, appropriately stratified by hemisphere, power-block and language, is expected to create, within one week, an agreed text on a subject beside which Vietnam and the Common Market are mere tea-cup storms restricted to parts of continents for a mere generation or two.

The problems presented by the genetic future of man are far more important than anything under the control of warriors, bankers, or tradesmen, and, by their nature, no executable advice can be expected from a week's discussion. What seems important is that no one should expect the committees or secretariats to undertake such impossible tasks. Given its magnitude the achievement may well be commended, but the results are not conducive to the orderly development of either practice or theory.

Perhaps the World Health Organization technical report series should restrict itself to technical reports and provide summaries of what is known, linguistic recommendations, and recipes for laboratory procedures; this may seem a rather pedestrian approach, but, in a world short of bread, elaborate schemes for cake-making should be thoroughly tested on the rich for acceptability and digestibility before being offered to the poor.

Technical reports should surely be reports of technical matters. By tackling some specific problem, such as G6PD, clotting factors, or immunoglobulins, this series has had a profound influence on the standards of human genetics throughout the world and the informal benefits of meetings of world experts have been accompanied by formal reports which have defined problems, increased the precision of words, and provided a framework from which local policy decisions could be made.

The recent report on haemoglobins shows once again the benefits which a central organization can confer simply by assembling experts and reviewing facts.

Recommendations

THE WHO Scientific Group on Genetic Disorders met in Geneva from November 16 to 22, 1971. Its chief recommendations included the following:

- medical genetics services, including counselling clinics, should be provided at medical centres;
- physicians and the public should be better educated in the principles of genetics and their relevance to human welfare;
- better facilities for prenatal diagnosis should be developed;
- more research should be undertaken, particularly in certain areas, including prenatal diagnosis, population screening for heterozygous carriers of specific mutant genes, and the identification of environmental pollutants that might increase mutagenesis. Follow-up studies on children born after amniocentesis are also needed to assess the long term effects of the procedure, as are studies on the effects of inborn errors of metabolism in the mother or the fetus;
- medical genetic centres should set up registries of genetically determined disorders.