

eligible for prenatal diagnosis at the time the Scientific Group met (November 1971) are listed in the Annex; those which had actually been diagnosed *in utero* are printed in italics. Neither sickle cell disease nor cystic fibrosis is printed in italics; moreover, the former is keyed to a footnote which indicates that, since the condition is known to be genetically heterogeneous, particular care must be taken with those methods which at the time were thought to be of some use in prenatal diagnosis of this particular disease. The literature sources of the material in the table are clearly stated on the same page of the WHO report. Professor Edwards or any careful reader can obtain the details by reading the source material—as we did.

Professor Edwards criticises our use of compound interest to estimate the doubling time for multifactorial disorders, as cited in the final pages of the report (Annex 3). Although he criticises this approach, he does not suggest an alternative. The final graph, showing the time in generations required to double the frequency of harmful genes for single gene diseases, should reassure Professor Edwards rather than alarm him; doubling times of 1,000 or 100 or even 10 generations would hardly seem alarming, especially for frequencies so minute.

While we are waiting for Professor Edwards's precious grail of "orderly development of either practice or theory" to be found, we hope that he will permit those who have access to the report to use the great part of its 46 pages which escaped unscathed from his comments, for the purposes intended, namely, to begin to apply current genetic knowledge to the immediate benefit of the patient with genetic disease.

#### Error

We, "an assembly of experts with a dedicated secretariat . . . appropriately stratified by hemisphere, power block and language", remain grateful to Professor Edwards for pointing out an error in the report. We stated that relaxed selection, acting over 100 generations, would increase the frequency of a trait from 0.001 to 0.0086; the correct final frequency should be 0.0102. We fail, however, to see how this or any other alleged error in the report affected the conclusions that were drawn by the committee. If, as Professor Edwards asserts, "some" of the "doubtful assumptions, erroneous facts and uncertain inferences" he finds in the report "overflow into recommendations", would it be too much to ask how correction of a single one of the "errors" he cites would, or should, have altered the recommendations the committee made?

CHARLES R. SCRIVER CHAIRMAN

ITALO BARRAI	}	SECRETARIAT
F. CLARKE FRASER		
WALTER E. NANCE		
HORST BICKEL	}	RAPPORTEURS
C. O. CARTER		
N. P. BOCHKOV		I. HALBRECHT
J. A. BOOK		M. F. LAMY
N. FUJIKI		E. ROSSI

PROFESSOR EDWARDS writes: Quotations are by nature out of context, and any obtuseness in understanding that small fraction of this technical report which was not technical, and which, rightly or wrongly, I considered could lead to an exaggerated claim of the need for counselling, must be left to the reader. So far as Annex 1 goes it seems reasonable to regard categories specified by percentages which sum to 100% as exclusive. In Annex 2 the heading "All the diseases in the following list can now be diagnosed prenatally" also seems clear. I was certainly in error in overlooking the conditional following the unfortunate apposition of "counsel" and "urge".

I do not think I am alone in feeling some anxiety at the casualness with which genetic counselling is being advanced as a salve for the problems of either our natural variability or for the therapeutic intractability of some of its consequences and, where emotion and enthusiasm run high, there is no substitute for precision in word and number and caution in application. The greater the expertness of a committee, or the weight of authority of the organisation, or its area of influence, or the distinction of its previous reports, the greater the need to expose to public accountancy any doubtful claims which may be influential. I am not doubting the benefits available to the populations of, for example, Montreal or Moscow, or the advantages of their wider availability, provided that they are subordinated to an efficient administration for diagnosis and therapy and do not compete with this basic requirement in the allocation of funds. I am doubting the scale of benefits to populations of counselling *per se* and disturbed at the casual widening in terminology by which counselling is extended to diagnosis and unsolicited advice, therapy to selective abortion, genetic disease to multifactorial disease (I hardly claim any originality for the view that all diseases are multifactorial, but some are less multifactorial than others), and prevention to elimination. In neither Montreal nor Moscow, nor in any other city, would I wish to see unsolicited genetic counselling being given on the basis of computerised files of so-called multifactorial disease (which includes schizophrenia) in relatives. I do not share the optimism of the group which "is convinced that genetic registries can be designed and used in ways that will contribute to the diagnosis and prevention of genetic disease without

endangering individual rights", unless genetic is used in the restricted sense which excludes multifactorial, or the registries are decentralised and maintained within hospital records departments. Quixotry implies an aggressive and well-intended reaction to a delusion and, while I hope my anxieties are delusions, I remain unconvinced.

It would be a great tragedy if drawing attention to any apparent *non sequiturs* should in any way be regarded as critical of the technical reports in general, or of the administration under which they are edited. Effective documents are necessarily controversial and the genetic future of man is too important a subject to be constrained by the niceties of private dissent.

#### BIRTH DEFECTS

### Hope for Spina Bifida

SINCE the publication of the WHO report number 497 there have been several advances in the prediction in the uterus of a defective embryo. Perhaps the most significant of these has been the discovery that foetuses which are anencephalic or which suffer from spina bifida release a high amount of alpha foetoprotein into the amniotic fluid. This high protein level, if detected as a result of amniocentesis, is a sign of a deformed foetus.

In Britain about 2,000 anencephalic children, and a similar number of spina bifida cases are born annually. In principle these births can now be avoided if a positive amniocentesis is followed by abortion.

It is, however, impracticable to study every pregnancy in Britain (731,000 births in 1972). Therefore the discovery by Dr D. J. H. Brock and colleagues at the University of Edinburgh (*Lancet*, ii, 923; 1973) that for anencephaly an excess of alpha foetoprotein is found in maternal blood, is welcome.

Spina bifida, however, throws up greater problems than anencephaly for such children who undergo surgery soon after birth, live for a long time, although they are usually both physically and mentally handicapped afterwards. Anencephalic children die at birth.

An excess of alpha foetoprotein is also found in the blood of some mothers who are carrying spina bifida foetuses but Dr Brock said this week that the test on maternal blood was not as reliable as in the case of anencephaly. It seems that an excess of protein is found when the foetus is very deformed but when the deformation is slight there is not always a detectable amount of the protein in the blood.