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Symposium : Medical genetics

Editorial :

Medical genetics in India

The high infant and perinatal mortality rates observed in India generate the impression that genetic disorders are of little consequence in developing countries. However, the presence of numerous factors such as large family size, high rate of consanguineous marriages, and the selective pressure exerted by Falciparum malaria, lead to the occurrence of a large number of genetic disorders in developing countries.[1] Moreover, the vigorous implementation of primary health care is likely to bear fruit in the coming decade. For example, in an area of Haryana, the introduction of simple measures to combat neonatal tetanus, and distribution of oral rehydration fluid to control diarrheal disorders reduced the infant mortality rate, and increased the contribution of congenital malformations as a cause of infant mortality from 5% to 13%.[2] In the city hospitals congenital malformations are already the third commonest cause of perinatal mortality, contributing to about 15% of it.[3]

At what stage in the development of a country should genetic services be provided ? Scriver pointed out that the population structure (number of persons at different ages) in developing countries has a 'pyramidal' shape, as constrasted with that in developed countries wherein it is 'cylindrical'[4] (Fig 1). The pyramidal shape is due to the large number of children and a stepwise reduction in their number as a result of mortality. He suggested that when the population structure in developing countries assumes a cylindrical shape genetic disorders become important.[4] The present author feels that the morbidity and mortality patterns will change sooner than the alteration in the population structure, and one cannot depend upon this criterion for establishing genetic services. Others have suggested that when the infant mortality rate has been reduced to below 50, then genetic disorders deserve attention.[5] However, a review of the situation in India shows that this is not likely to happen until 2000 AD. Obviously the expectations of the people cannot remain unfulfilled for that long. WHO Hereditary Programme Advisory Committee has proposed that one should examine the graph correlating infant mortality rate with birth rate to determine the developmental status of a country.[5] The better the health services, more the curve reaches down to the left lower corner, and greater is the need for organising genetic services. Fig. 2 shows that India occupies a middle position in such a graph.

It is clear therefore that no single criterion can be used to signal the introduction of genetic services in a country. In India,[1,6] hemoglobinopathies, congenital malformations, and genetic causes

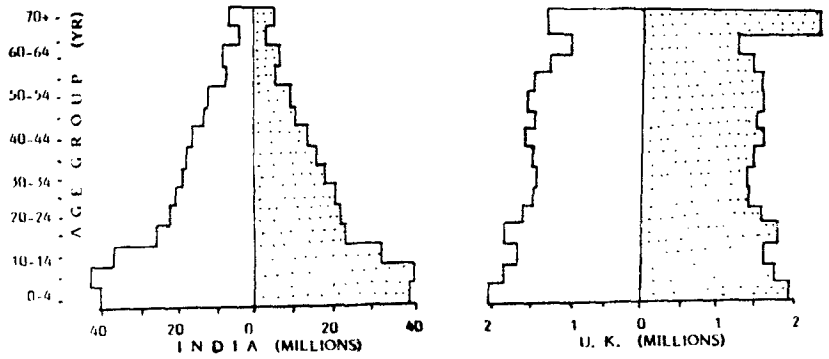

Fig. 1. Population structure of India (Census, 1971), contrasted with that of U.K. (Demographic Yearbook, 1971).

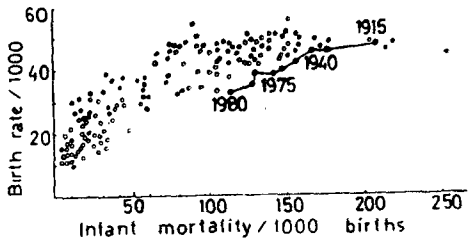

Fig. 2. Infant mortality per 1000 births related to birth rate per 1000 for the world and India.

of mental retardation affect a large number of children, the total reaching astronomical figures due to the huge population size (Table). It can be estimated that in India one infant with a genetic disorder is born every 40 seconds ! How long can we deny appropriate medical services to such children and their parents ?

Obviously, we need to establish genetic centres in all the medical schools in the country with 4-5 advanced regional centres. In the villages the health workers should be trained to identify those with possible genetic disease. The primary health doctor should be able to counsel for genetic disorders common in that area. The laboratories in the district hospitals need to be upgraded so that diagnosis of hemoglobinopathies, urinary aminoacid chromatography, and karyotypes from blood culture can be carried out. Alternately for the time being, genetic counsellors from the medical

Table. Genetic disorders in India

Disorder	Prevalence (%)	Total no. (millions)
Affected at birth		0·825
Congenital anomalies*	2·5	0·564
Single gene	0·6	0·135
Chromosomal	0·56	0·126
In general population		
Behavioural and C.N.S. disorders**	1·5	10·650
Late-onset multifactural disorders***	10·1	27·390

Data based on 1981 census. Details of computation are given in ref. 6. *A quarter of malformations are assumed to be genetic in origin; **Includes nonspecific mental retardation, schizophrenia, manic depressive psychosis, and epilepsy; *** Includes hypertension, diabetes mellitus, and coronary artery disease.

schools should visit the peripheral centres on designated days.[7] These are gigantic tasks which will need a concerted effort by health planners and specialists in the medical schools; but these are tasks which cannot be ignored if we are to provide health for all by the year 2000 AD.

In this issue we publish articles covering recent advances in the field of medical genetics. *Ramalingaswami* describes how modern genetics has applications in the diagnosis and control of infectious and parasitic disorders. A constant theme in clinical genetics is genetic heterogeneity, i.e. similar disorders or phenotypes being caused by different genes. *Kumar* illustrates this by discussing cerebellar ataxias. *Joshi* et al discuss the recent advances in Duchenne muscular dystrophy. Mental retardation is a common end result of many genetic disorders. Of the chromosomal syndromes which cause mental retardation Down syndrome is the commonest occurring in India with a frequency of 1 per 880 births or 1·13 per 1000 births, based on an analysis of 72,339 births in eight centres in India.[8] The next commonest chromosomal cause which has been recently recognized is fragile X syndrome. This explains the obvious increase of males with mental retardation admitted in schools for the mentally retarded. This syndrome is well covered by *Mixon and Dev*. Chromosomal basis of recurrent fetal loss is reviewed by *Tharapel and Wilroy*. *Pai* presents a practical approach to genetic counselling, while *Chakraborty* describes the utility of DNA technology in the detection and prevention of genetic defects.

One of the most significant advances in the field of prenatal diagnosis is the use of ultrasonography. The improvements in equipment design have led to a dramatic increase in the ability of a trained ultrasonographer to detect fetal abnormality. This is elegantly presented in a most illuminating paper by *Elejaldes*, who have done much to establish the new discipline of fetal diagnosis. May more experts in India acquire such skills !

Prenatal diagnosis by amniocentesis at 16-18 weeks of gestation had hardly been established in a few centres in India,[9] when chorionic villi sampling (CVS) invaded this field. This has led to first trimester diagnosis by remarkably simple methodology. Scientists in many centres in India are learning to master the technical skills of CVS, and we hope it will soon be available as a clinical service. This subject is well reviewed by *Bombard and colleagues* from USA and *Bartels and Hansmann* from West Germany.

In closing I must thank Professor Dharmdeo Singh from USA for his help in the organisation of this symposium, and the various contributors for their enthusistic response to our requests.

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Part II of the **Symposium on Medical Genetics** will appear in *November-December 1986* issue of the *Journal*. The following articles will appear in that issue :

1. DNA polymorphism and clinical genetics :
   Ranjit Chakraborty

2. Genetic aspects of congenital cerebellar ataxia :
   D. Kumar

3. Chorionic villus sampling : first trimester prenatal diagnosis :
   Allan T. Bombard, Joe Leigh Simpson, Sherman Elias and Alice O. Martin

4. Duchenne muscular dystrophy : recent concepts :
   R.N. Joshi, M.J. Shah, N.B. Trivedi and H.D. Joshi