GENETICS AND SCHOOLS FOR THE BLIND

IDA MANN, *C.B.E.*, D.O.M.S., F.R.C.S., F.R.A.C.S., *Ophthalmologist, Western Australia*

It is common knowledge among ophthalmologists that the percentages of the various causes of blindness among children have altered materially during the last 50 years. It is probably time to take stock of this process and to assess its trend and its basic causes. 'Prevention of Blindness' was chosen by the World Health Organization as its theme for 1962, and among many possible preventive measures which could be discussed, that of investigation of the changing distribution of causes among blind persons in half a century might be expected to yield some pointers about the orientation of the work required to reduce their number.

It is well known and indeed self-evident that as the elimination of many of the lethal infectious diseases continues, the age of the population rises. Therefore, more people will reach the decades in which blindness is most common. Unfortunately, the three major causes of blindness in later life, glaucoma, cataract and macular degeneration, are still not fully understood and much more work on their aetiology is required. They will be referred to later. Blindness in later life at ages when, in most cases, activity is lessening and productivity is drawing to a standstill, is, although a tragedy, not so economically disastrous as blindness at earlier ages.

Blindness in childhood presents the most serious social problem of all. Not only does a blind child necessitate much outlay on special education, transport, etc. during school years, but the fact must be faced that full economic independence will be achieved in only a few instances. The expenses, therefore, of invalid pensions, subsidies to sheltered workshops, travel and other concessions will continue throughout life. It may, therefore, be profitable to review the situation with regard to serious eye defects among children in various parts of the world from the beginning of the century to the present time.

Civilized Communities

If we consult the 1915 *Report* of the National Committee for the Prevention of Blindness in the USA, we find that in 1907 26·5% of new admissions to the blind schools of America were on account of ophthalmia neonatorum, mostly caused by the gonococcus. A vigorous campaign for the education of the public and midwives in prophylaxis and treatment only succeeded in reducing this number to 15% by 1915. It will be realized that no specific treatment existed at that time, though prophylaxis was being used widely. Trachoma was also a cause of blindness at that time and 103 cases in children were known in New York city alone in 1916. Of these, 56 were in schools for the blind.

It is difficult to get a complete census of the causes of blindness from the figures given in the abovementioned report, but enough details are given to show that ophthalmia neonatorum headed the list, which in its incomplete form is as follows:

CAUSES OF BLINDNESS IN CHILDREN ENROLLED IN THE 31 STATE SCHOOLS FOR THE BLIND AND 4 CITY CLASSES: 1915

	%
Ophthalmia neonatorum	21·8
Congenital anomalies	21·3
Optic atrophy (cause not stated)	9
Trauma	7·9
Wood-alcohol poisoning	5
Interstitial keratitis	3·8
High myopia	1·9
Trachoma	1·4

It will be noted that the two major causes were infections and congenital defects. If ophthalmia neonatorum, trachoma and interstitial keratitis are included under infections, the total is 27%. With regard to the congenital anomalies, it is not certain whether some of the optic atrophy cases should be included in this group. If they all were included, then the figure would rise to 30·3%, which even in 1915 - 16 is slightly higher than that for infections. It is, therefore, apparent that the two great causes of blindness in children were infections and congenital anomalies.

These figures can be somewhat paralleled in England in 1920 - 25 where we find in the Sunshine Homes the following distribution of cases:

Ophthalmia neonatorum	31·8	45·1%
Other infections	13·3	
Congenital anomalies	34·8%	
Indefinite aetiology	5%	
Trauma	0·79%	

This shows that the rate of infection as a cause of blindness in civilized countries was not diminishing in the 10 years under consideration. Infection and congenital anomalies still presented the major problem.

By 1950, however, in England (*Causes of Blindness in England 1948 - 50:* Report by Arnold Sorsby) the picture had completely changed. For example, only 7 children (aged from 0 to 14) were registered as blind from ophthalmia neonatorum in 1948 - 50, 0·03% of the total registered blind persons for those years (18,150). On the other hand, 336 were registered blind from congenital abnormalities in the same age group. Therefore, 98% of blind children in England in 1950 were blind from developmental defects. In other words there are now 49 times as many children blind from congenital abnormality as from infection. None in this age group are blind from congenital syphilis.

Sorsby analysed the causes of blindness for *all* age groups in England as follows:

CAUSES OF BLINDNESS RESPONSIBLE FOR 1% OR MORE OF CASES: 1948 - 50

	%
Cataract	30·4
Senile macular degeneration	15·3
Glaucoma	14·0
Myopia	8·9
Congenital and genetic defects	7·6
Diabetes	4·3
Iritis and iridocyclitis	3·5
Optic atrophy of unknown origin	2·7
Vascular diseases	2·6
Corneal lesions of indefinite aetiology	2·1
Affections of the globe of indefinite aetiology	1·5
Syphilis	1·2
Choroidal lesions of indefinite aetiology	1·0
Primary retinal detachment	1·0
All other causes	3·9

It is thus obvious that infection has been practically eliminated and that we are left with two major problems, congenital abnormality in children and the big group of cataract-glaucoma-senile macular degeneration in old age.

If we turn to other civilized areas of the world we find much the same situation. A survey of 50 blind and partially blind white children in Western Australia shows that only 15 of them became blind from infection or trauma, as is seen from the list which follows:

Toxoplasmosis	4
Spastic with optic atrophy	3
Rubella in the mother	2
Still's disease	2
Meningitis	2
Cerebral tumour	1
Birth injury	1

The remaining 35 children fall within the following categories:

Nystagmus and partial albinism	13
Microphthalmia	5
Buphthalmia	3
Aniridia	2
Cataract	6
Optic atrophy	1
Coloboma	1
Dislocated lenses	1
Anophthalmia	1
Day blindness	1
High myopia	1

Among these 35 cases (70% of the total) 8 are known to be familial and many of the others may be so. They all fall into the category of developmental defects and all the conditions have been reported at some time or other as hereditary.

A similar situation was found in a school for the blind in South Africa. These were Indian children and here the rarity of infection as a cause was equally striking, thus:

INFECTIVE CASES

Corneal scars	1
Meningitis	1
Congenital syphilis	1
Tick fever	1
Phthisis bulbi	1

NON-INFECTIVE CASES (DEVELOPMENTAL)

Congenital amblyopia and optic atrophy (probably retinal aplasia)	7
Nystagmus	6
Retinitis pigmentosa	4
Cataract and fragilitas ossium	2
Buphthalmia	2
Coloboma	1
Cataract	1
Microphthalmia	1
Retinoblastoma	1

Of these there was a family history in 8 cases and a possibility in the rest. Developmental anomaly here accounts for 83% of blindness in children and it is obvious that there has been a most remarkable change in emphasis in the last 20 years. This is obviously due to the discovery of the sulpha drugs and the antibiotics.

If we would lessen the number of blind children therefore, we must consider more closely the genetic aspect of the situation. In the Indian community in South Africa there is naturally a tendency to inbreeding and this is also found to a certain extent in parts of Australia where cousin marriages were common in the first and second generations of pioneers. This inbreeding would naturally have no effect on the ratio of infective to developmental eye defects, but it would tend to increase the total number of children blind from congenital anomaly. It may, therefore, be of interest to consider some so-called uncivilized races in which (although the problems of infection have not yet been tackled) the exogamous marriage laws could have influenced the amount of genetic defect.

Less Civilized Communities

Various surveys of eye disease have been made since 1950 on aboriginal Australians. In 1953, 2,185 part- or full-blood aboriginals were examined in the Kimberley division of Western Australia. Of these, 111 were blind in one eye and 149 blind in both eyes. This is a very high rate of blindness by 'civilized' standards, yet of these only 1 was blind in one eye and 1 in both eyes from congenital defect (buphthalmia). All the rest were the result of infection or trauma. Four other cases of congenital defect short of blindness were found.

The surveys were continued for areas farther south. In the Eastern Goldfields and the Warburton Ranges 1,105 aboriginal people were examined and 28 persons were found to be blind in both eyes, all from infection, injury or senile cataract. One case of monocular buphthalmia was found and one coloboma of the iris. Therefore, among 3,290 aboriginal Australians (of whom 325 were blind in one or both eyes) only 1 was blind from congenital defect (0·3%). Only 8 cases of congenital defect in all were found (0·2% of all persons examined).

Surveys in the Territories of Papua and New Guinea are also of interest. In 1955, 13,268 native persons were examined on the mainland in New Britain, New Ireland, in the Trobriand Islands, the Marshall Bennetts and the Admiralty group. Here the marriage laws differ from tribe to tribe, but on many of the smaller islands inbreeding is unavoidable. This is sometimes apparent by the presence of several albinos in one village. Altogether 58 cases of congenital anomaly were found (0·4% of persons examined). Of these 58, 9 were blind in one eye and 11 in both eyes. The defects found could mostly have been genetic, but no family histories were available (except in the case of the albinos). The defects were as follows:

Microphthalmia	12
Congenital cataract	11
Albinism and nystagmus	7
Grape-seed bodies on iris	5
Persistent pupillary membrane	4
Buphthalmia	3
Retinitis pigmentosa	3
Conical cornea	2
Microcornea	2
Corectopia	2
Atypical coloboma	1
Epicanthus	1
Epiblepharon	1
Congenital ptosis	1
Fibrolipoma	1

The total number of persons blind in both eyes was 96 and in one eye 312. Therefore, congenital anomaly accounted for 11·4% of blindness in both eyes and 2·8% in one eye. Even this is far below the figure for the USA even in 1915, and strikingly below those for England, white Australia and the Durban Indian school described above. It is also in contrast to the findings for Australian aborigines, whose marriage laws tend usually more towards exogamy. This evidence appears to show that improved conditions of life, by largely eliminating infection and trauma as causes of blindness in children, are linked with a rise in the percentage of blindness from developmental defect. How much is truly hereditary is not known with certainty, but today there are very few developmental defects which have not at some time been reported to be familial or inherited.

Heredity in 'Senile Degenerations'

The field of genetics in ophthalmology is continually widening and has recently been shown to include conditions previously explained as senile degenerations. There is continually increasing evidence, for example, that glaucoma simplex is hereditary and indeed partly racial, being extremely common in Icelanders, Chinese, Nigerians and most European races. It is rare in Polynesian peoples, in Australian aborigines and Papuans. Senile cataract and macular degeneration also show familial and hereditary tendencies in many cases, so that a genetic background study is of importance, not only in childhood but also in old age.

CONCLUSION

The question whether the genetic causes of blindness can ever be influenced is a vexed one. When we consider that at present 98% of children in blind schools in England alone are there because of genetic defect we should take stock of the situation. Although recent biochemical discoveries adumbrate the possibility of influencing genetic constitution there is little immediate hope of this. In the meantime the way in which improvement in the situation can be hoped for is through education, propaganda and advice.

It is strange that, in an age when everyone is aware of the refinements of animal breeding, the knowledge that the same principles apply to the human race should be as limited as it appears to be. Education could overcome this ignorance. The teaching of human genetics in a simple way should form part of compulsory biology in all school curricula. The setting up of free guidance councils, possibly attached to the genetics department of universities, where advice on the risk of transmitted defect could be available to intending parents is much needed. Propaganda advocating the use of such a service and

setting forth the reasons for its need (not only in ophthalmic problems) is necessary. Teachers of the blind should understand their pupils better and should know the genetic risk to each individual, so that they could help by explanation to foster a sense of responsibility in the students. At present, since blind schools are mostly co-educational, there is every chance of the continued perpetuation of defects. Very many instances are known of two and even three generations of blind children of the same family attending the same school, their parents (and grandparents) having met there originally.

Finally, this is not only an ophthalmic problem. It extends to every type of physical and mental handicap among children. A very simple research programme, in which a genetic analysis of all children in schools for the handicapped in any country was done, would bring the matter into proper perspective and make it certain that our next advance in prevention of disease must be along genetic lines.