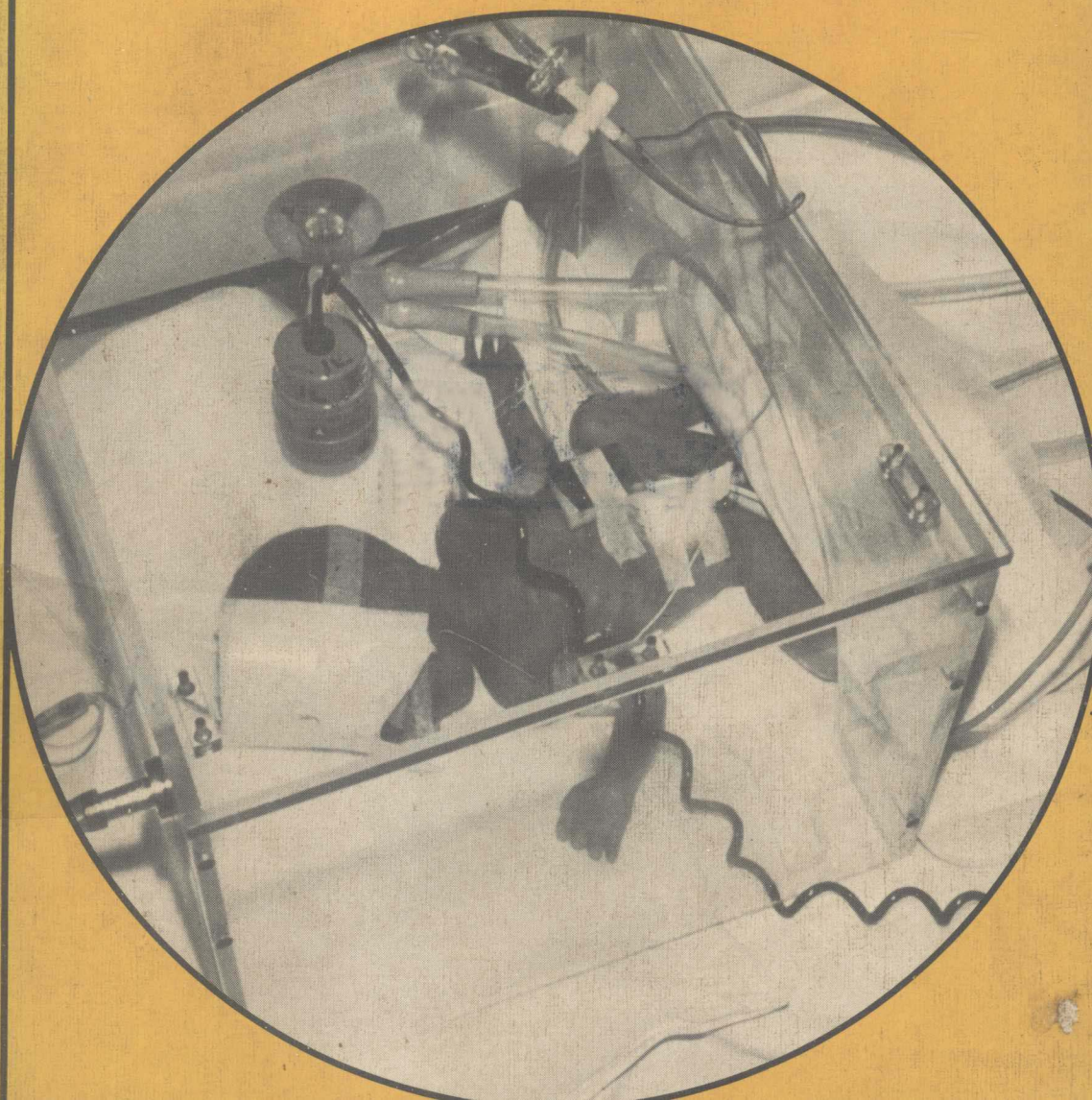
Ciba Foundation Symposium 59 (new series)

Major Mental Handicap: methods and costs of prevention



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Chairman's introduction

C. O. CARTER

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The main point of our meeting is to discuss the extent to which severe mental handicap may be prevented by better prenatal and perinatal care and to consider the cost of providing such care.

First the problem must be set in the perspective of the aetiology of severe mental retardation and the opportunities for, and perhaps the relative cost of, prevention of other types of severe mental retardation, such as those due to chromosomal and inborn metabolic errors. The first contribution, by Dr Alberman, will set out this perspective. The birth frequency of severe mental retardation, like that of the major congenital malformations, has been little affected by the public health measures which have been so successful in reducing the rate of infantile and more recently perinatal mortality. The prevalence at school age is usually taken as between 3 and 4 per 1000, and Professor Hagberg will be giving us information on this. Such reduction as there has been is perhaps largely confined to Down syndrome and is the consequence of the reduction in births at late maternal ages. We now have a clear picture of the aetiology of half to two-thirds of all cases of severe mental retardation. Of the genetic types trisomy 21 is outstanding, being itself responsible for about a third of all severe cases of mental retardation at primary school age. This group, as Dr Alberman will tell us, is now amenable to secondary prevention by prenatal diagnosis and abortion, though we have no way yet of preventing the primary error in gametogenesis. Cost–benefit analysis for such prenatal screening and abortion has been made in two or three countries and these measures appear to be cost-effective at least down to maternal age 38. Monogenic disorders are less important, perhaps accounting for 10 to 15%, and again Professor Hagberg has some up-to-date figures from two areas of Sweden which show the expected differences in relation to inbreeding. Known prenatal environmental causes are a relatively small

group: intrauterine rubella is responsible for perhaps less than 1% of cases and is now preventable; cytomegalovirus infection is possibly more frequent than rubella, though we need more information on this. Maternal alcoholism does not appear to be making any significant contribution to severe retardation in the UK at present, and smoking in pregnancy perhaps contributes only indirectly, through low birth weight for dates. Spina bifida again makes a small and now largely preventable contribution. Clear-cut postnatal causes, such as cerebral and meningeal infections, again make only a small contribution and are preventable. Kernicterus due to rhesus incompatibility is already preventable and mostly prevented.

This leaves, say, 40% of cases with no certain cause and Professor Hagberg will be reminding us that it is in this group that associated neurological disorders such as cerebral palsy, epilepsy and disorders of the senses are relatively common. Some cases in this group (Dr Drillien will be giving us an estimate of the proportion) are due to perinatal events, mainly asphyxia and cerebral haemorrhage. Probably this is not a large group in relation to all severe mental retardation, but these conditions are in part preventable and well worth preventing. A more difficult group perhaps, both for evaluation of frequency and for attempting prevention, are the cases with prenatal acquired causes other than infection. Dr Hagberg's Swedish colleagues have described this group as having the 'fetal deprivation syndrome'; it includes babies whose mothers suffered from associated bleeding in pregnancy, placental infarction, toxaemia and diabetes. Dr Hobel will discuss some of the methods available to the obstetricians dealing with high-risk pregnancies—methods that may reduce the impact of such prenatal causes. Professor Reynolds, Dr Fitzhardinge and Dr Stewart will discuss the methods available to the neonatal paediatrician for preventing perinatal damage. Not only may the survival rate of such high-risk infants be improved, but the rate of survival without mental handicap. Dr Amiel-Tison will discuss methods of measuring some of the less severe neurological defects.

Finally, if severe mental retardation can be prevented by better obstetric and neonatal care, what is the cost? Ms Holtermann will discuss this and then Dr Chapalain will tell us something of the experience in France, which has recently reduced its infant mortality from well above to well below that of England and Wales.

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Main causes of major mental handicap: prevalence and epidemiology

EVA ALBERMAN

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Abstract The prevalence of educational subnormality of a severe form (between 3 and 3.6 per thousand children of school age) and the prevalence of cerebral palsy (between 2 and 2.4 per thousand) have been fairly stable up to recent years. This stability has also applied to the relative proportions of the different major causes contributing to the handicaps.

Where the ascertainment of such conditions is good, their prevalence monitored and the life expectancy of affected individuals estimated, any changes in prevalence can be used to measure the effectiveness of new forms of prevention, or alternatively to indicate the existence of new environmental hazards. Only a multi-pronged campaign against many of the recognized causes will have a substantial impact on prevalence.

The term 'major mental handicap' is a very broad one, encompassing an enormous variety of pathological conditions, and it can be defined in many different ways. In practice, there is general agreement on one group to be included: those individuals needing lifelong and constant care because of their inability to look after themselves. It seems that an arbitrary cut-off point of an IQ of below 50 defines such a group and, following Kushlick (1965), it has become usual to add to this group individuals with Down syndrome, if their IQ has not been tested. With these I will group, for the purposes of this symposium, the heterogeneous collection of conditions known as cerebral palsy, which is often, but not always, associated with mental defect.

There has been a vast amount of work on the prevalence and epidemiology of the conditions which together make up the group I have defined in this way, and one factor stands out clearly. This has been an apparent stability in over-all age-specific prevalence, which is in marked contrast to the large variations in reported prevalence of milder forms of mental defect. The latter can be shown to vary strikingly with cultural factors, and with differing

educational provisions for this special group. The severe forms of handicap do not show such variations, and by common consent can be regarded as largely biological or pathological in origin, rather than cultural or just the tail of a normal distribution.

The actual prevalence of major mental handicap as I have defined it depends on several basic factors. Most directly, it depends on the incidence at birth of its constituent conditions. This in turn depends on the frequency in the population of parents at high risk of producing offspring with specific abnormalities. After this, the ultimate prevalence at different ages will depend on the survival of affected individuals. The reported prevalence will also depend on the thoroughness of ascertainment of the handicapped individuals (ascertainment is the technical term used by educational authorities who have a statutory duty to seek out children who will require special educational provision). Considering the very complex nature of these interacting factors, and the extremely large number (probably in the thousands) of different conditions making up the total group, I never fail to be surprised at the stability of the prevalence of handicap, both over the past 20 years in the UK and in many cases also between countries.

This stability can be demonstrated by reference to specific well-recognized disorders such as Down syndrome; or to such a heterogeneous group as the cerebral palsies; or to the reports of the so-called 'administrative prevalence', at different ages, of individuals in need of special care by reason of their handicap.

DOWN SYNDROME

Table 1 shows estimates, from different sources and at different times, of age-specific population incidence rates of Down syndrome at birth. Overall the Swedish rates are consistently higher, but Lindsjö (1974) shows that, with the exception of the small group of mothers in the very oldest age-band, these differences can probably be ascribed to better ascertainment in Sweden. Allowing for this, there is on the whole a close similarity between the rates, particularly in the age-groups with the largest numbers, with the well-known steep rise with increasing age. Such examples can be multiplied many times. This is surprising, because even Down syndrome is heterogeneous in origin, with about 3% of the cases in most reported series resulting from a chromosomal translocation and the remainder from primary trisomy 21. The variation of risk with age is seen only with the latter.

TABLE 1

Incidence per thousand births of Down syndrome by maternal age group (adapted from Lindsjö 1974)

Maternal age	*Sweden 1968-70 (331 000 births)*	*UK 1951-63 (1 700 000 births)*	*Australia 1942-57 (780 000 births)*
-19	0.59	0.45	0.42
20-24	0.74	0.49	0.62
25-29	0.88	0.65	0.82
30-34	1.46	1.08	1.13
35-39	3.74	3.37	3.45
40-44	14.96	10.74	9.80
45-	62.10	24.94	21.56

CEREBRAL PALSY

In view of the difficulties of diagnosis and classification of cerebral palsy it is not surprising that the variation in reported rates is a little larger than for Down syndrome. Table 2 summarizes the data from the literature presented by Henderson (1961) and adds more recent data from the National Child Development Study (Davie *et al.* 1972). The most carefully ascertained studies

TABLE 2

Reports of the prevalence of cerebral palsy in Great Britain in children of school age (adapted from Henderson 1961)

	Rates per thousand	*No. of studies*
Range of rates reported in children of school age in 19 studies in UK, 1948-1957	<1	5
	1.0-1.4	4
	1.5-1.9	4
	2.0-2.5	6
National Child Development Study, 1965 (Davie *et al.* 1972)	2.4	

included were those from Scotland, Henderson's (1961) from Dundee and Ingram's (1964) from Edinburgh, and their rates were 2.04 and 2.3 per thousand, respectively. Together with the National Child Development Study rate of 2.4, and with reports from overseas suggesting about the same level in the 1950s and 1960s, we may accept that the real prevalence at that time was between 2 and 2.4 per thousand children of school age. Moreover, there has been fairly good agreement from many different sources on the proportions of different types of cerebral palsy found (Mitchell 1961). Table 3 gives the proportion of the different types found by Henderson (1961) and his col-

TABLE 3

Distribution of different types of cerebral palsy (adapted from Henderson 1961)

	Dundee (Henderson 1961) %	*Edinburgh (Ingram 1964)* %	*Bristol (Woods 1957)* %
Hemiplegia	37.5	36.1	36.6
Double hemiplegia	1.3	3.9	
Diplegia	38.3	38.0	
Ataxic diplegia	2.9	5.8	Different
Ataxia	1.7	7.2	classification
Dyskinesia	8.4	8.2	
Other	10.0	1.0	

leagues in the population study carried out in Eastern Scotland in 1955, and compares this with other similar studies.

For most published surveys the relative proportion of the main aetiological features is also fairly constant. Typical of the findings are those described by Henderson (1961). In this series of 240 children, comprising all known cases from Eastern Scotland in children of school age in 1955, 12% of all cases were of 'postnatal origin', a term which included kernicterus; and the incidence of birth weights of 2500 g or less was 27.5% and of twins 10%. In the subgroup of cerebral diplegia, well known to be associated with premature births, the incidence of low birth weight was 58.9%.

We can therefore estimate that even a halving of the incidence of low birth weight, or alternatively the effect of reducing long-term defects in such births by 50% or more, could only reduce the prevalence of cerebral palsy by 13%, all other factors remaining equal. It will be seen later that the effect of this reduction in cerebral palsy on the prevalence of severe mental defect would be considerably smaller.

In most series for which data are available, about 25% of children with cerebral palsy have an IQ of below 50, and a further 20% or so have an IQ between 50 and 70 (Cockburn 1961).

EDUCATIONAL SUBNORMALITY (SEVERE)

As I said earlier, the very mixed group classified as educationally subnormal (severe) itself showed a surprisingly stable prevalence within age-bands. Indeed, there is evidence that much of the reported variation in prevalence is due to difficulties in ascertainment, for in all studies the reported prevalence rises with age to a maximum at ages 15–19. It is presumed that in the younger and the older populations ascertainment is incomplete. Table 4 gives the

TABLE 4

Comparison of the prevalence rates of IQ under 50 in age groups where all subjects are likely to be known (adapted from Birch *et al.* 1970)

	Age of children	*Total rate per 1000 with IQ < 50*
England & Wales (Lewis 1929)		
1925-1927 urban	7-14	3.76
Middlesex (Goodman & Tizard 1962)		
1960	7-14	3.45
1960	10-14	3.61
Salford (Susser & Kushlick 1961)		
1961	15-19	3.64
Wessex (Kushlick 1964)		
1964 county boroughs	15-19	3.54
1964 counties	15-19	3.84
Baltimore, MD, USA (Lemkau *et al.* 1943)		
1936	10-14	3.3
Aberdeen, Scotland		
1962	8-10	3.7
Edinburgh, Scotland (Drillien *et al.* 1966)		
1962-1964	7-14	4.9
Quebec, Canada (McDonald 1973)		
1966-1969	10	3.8
England, Wales & Scotland (Frew & Peckham 1972)		
1969	11	3.6

prevalence of this group in several studies both inside and outside the United Kingdom and shows how little variation there is within the age-ranges indicated, given the difficulties of full ascertainment.

Table 5 gives the proportions of different conditions found in a series of severely retarded children in Hertfordshire between 1965 and 1967 (Laxova *et al.* 1977) and in whom particular care was taken to diagnose the cause where possible. The general findings are fairly typical of many other series, with Down syndrome accounting for about one-third of all cases in most published studies. Local variations may occur, as in areas where neural tube defects are unduly common, or in remote areas, such as some parts of Sweden, where certain genetically determined conditions are frequent, but in general the pattern has been very stable.

TABLE 5

Distribution of different types of condition in severely retarded children in Hertfordshire, 1965-1967 (adapted from Laxova *et al.* 1977)

	Distribution (n = 146) (%)	
Genetically determined		
Down syndrome	32.2	
Trisomy 21		30.1
Translocation		2.1
Other	16.5	
Epilepsy - idiopathic	1.4	
Neural tube defect	4.8	
Cerebral palsy	6.8	
Postnatal brain damage (including one case of rubella syndrome)	4.1	
Other	34.2	
All	100	

IMPLICATIONS OF THE STABILITY FOUND

It is because of the generally stable pattern of prevalence that it should be possible to plan and to evaluate the most effective methods of prevention. With such a heterogeneous collection of conditions, one must study each condition individually as far as possible and, for each, determine the major aetiological factors and the extent to which they are preventable. Given a baseline derived from well-ascertained studies, substantial changes in trend should be detectable, if surveillance of incidence and prevalence of conditions such as I have discussed is maintained.

THE FORMS WHICH PREVENTION CAN TAKE

Prevention of these conditions may take many different forms, largely related to the nature of the cause of the defect. Thus, for purely genetically determined conditions, the only form of prevention may be the avoidance of conception by women known to be at high risk, or termination of pregnancy where an affected fetus can be diagnosed. For maternal infection, prevention can take the form of preconceptional immunization together with termination

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