INFANTILE IDIOPATHIC SCOLIOSIS

CAUSATIVE FACTORS, PARTICULARLY IN THE FIRST SIX MONTHS OF LIFE

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An attempt has been made to determine the aetiological factors in infantile idiopathic scoliosis from a clinical, genetic and epidemiological survey of 134 infants, ninety-seven of whom developed a curve in the first six months of life. Plagiocephaly was present in all cases; mental retardation occurred in 13 per cent of males with progressive scoliosis; congenital dislocation of the hip occurred in 3-5 per cent of cases and congenital heart disease in 2-5 per cent; and inguinal hernia was found in 7-4 per cent of males. Approximately 3 per cent of parents and 3 per cent of sibs had the same deformity, thirty times the general population frequency for the Edinburgh area. Other positive findings included an excess of breech presentations and of premature, low birthweight males, and a preponderance of curves developing in the winter months. Infants with progressive scoliosis tended to have older mothers and to come from poorer families. Only three children, all with resolving scoliosis, habitually lay prone in early infancy, in marked contrast to North American infants where this posture is usual. The almost complete absence of infantile idiopathic scoliosis in North America is noted and it is thought that the two facts may be related. The aetiology is likely to be multifactorial, with a genetic tendency to the deformity which is either "triggered off" or prevented by external factors.

Clinical details of idiopathic scoliosis as an entity have been available for only about twenty-five years (Ponseti and Friedman 1950). The first large groups of patients with infantile idiopathic scoliosis were described by James (1951, 1954), "infantile" being defined as under the age of four years. It was noted that the curve was seldom present, or noticed, at birth, but usually developed during the first few months of life. The sex ratio was unequal, approximately three males to two females. Nearly all curves were thoracic and about four-fifths were convex to the left (James, Lloyd-Roberts and Pilcher 1959). Two types of structural scoliosis were noted, resolving and progressive, the range being from a mild, transient curve to the beginning of one of the most severe progressive deformities known, causing death from cor pulmonale in early middle age. It was known that at least half the infants attending specialist scoliosis clinics had resolving curves, but that the true proportion was likely to be much higher because many curves would not be considered serious enough to cause the child to be brought to hospital, or escaped notice altogether. Indeed, Lloyd-Roberts and Pilcher (1965) estimated that 90 per cent resolved.

In a survey of idiopathic scoliosis from Edinburgh (Wynne-Davies 1968) it was found that all infants, whether with resolving or with progressive curves, invariably had plagiocephaly as well, the "recessed" side of the head being always on the same side as the convexity of the curve (Fig. 1). This cranial deformity had usually disappeared by the age of five years. In preliminary studies on the plasticity of the infant axial skeleton Hay (1971) noted that plagiocephaly was not apparent in normal infants at birth but developed during the first few weeks of life. He suggested that its onset was related to the side of habitual lying of an infant, especially if the child was bundled up with clothes and unable to move, and he thought that this habitual posture could be a possible cause of scoliosis in this age group.

There had been speculation for many years before, particularly by Browne (1956, 1965) and by Lloyd-Roberts and Pilcher (1965), that posture in utero could cause infantile scoliosis. More recently, Dunn (1969) and

FIG. 1
Left-sided plagiocephaly.

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1 Based on a thesis submitted to the University of Edinburgh in 1973 for the degree of Ph.D.

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Watson (1971) considered the problem of infantile "postural" deformities, and concluded that pressures in utero were likely to be one cause of "positional" deformities such as scoliosis, torticollis, club foot and congenital dislocation of the hip. However, it is difficult to account for the fact that scoliosis, like plagiocephaly, is not seen at birth, or only very rarely. It is during the first few weeks or months of life that they both become apparent.

In the 1968 Edinburgh survey Wynne-Davies noted that infants with idiopathic scoliosis had an increased proportion of relatives with the same disorder, but there was no definite pattern of inheritance. It was stated that "it is probable that there is a strong environmental factor acting in the early onset type", though the findings did suggest genetic factors acting in addition.

It had been suspected for many years that the frequency of infantile scoliosis was very low in North America compared with Europe, but no precise figures were available until recently. Riseborough and Wynne-Davies (1973) have reported only 0.5 per cent from a scoliosis clinic in Boston, Massachusetts, the comparable figure from Edinburgh being 50 per cent. Because the genetic factors are likely to be very similar, this pointed to differing environmental factors.

A more detailed survey of idiopathic scoliosis has recently been completed in Edinburgh (Wynne-Davies 1973), one feature of which was the separate analysis of observations on infants developing scoliosis during the first six months of life. It was found that they had significantly different adverse factors in their birth and peri-natal histories.

It is not known when or by whom the significance of infant posture, particularly the prone position, was first suggested. Certainly the different habits of American and British babies and the possible relationship to infantile scoliosis have been freely discussed by scoliosis surgeons over the past decade.

MATERIAL AND METHOD

One hundred and thirty-four infants with scoliosis who attended the Edinburgh Scoliosis Clinic between October 1958 and December 1971 have been investigated. Ninety-seven developed a curve during the first six months of life and eighty-four of these were the subject of a genetic survey. Thus the proportions of relatives with scoliosis are known.

Clinical details were noted, including the sex ratio, the side of the curve, whether it resolved or progressed, and whether any other developmental anomalies were present. Information was collected relating to the pregnancy and birth history, including mother's age, length of gestation, presentation and birth weight. The season of birth and the time at which the curve developed was noted, and a history taken from the mother as to the infant's preference for lying on the left or right sides, on his back, or prone. The father's occupation was recorded as giving some indication of social class. Data from pregnancy and birth histories, and for "social" factors were compared with control figures obtained from the Annual Reports of the Registrar-General for Scotland (1939–1967) and from the Edinburgh Register of the Newborn (1964–68).

A survey relating to plagiocephaly was carried out in 107 normal infants aged between two weeks and six months, and data were also used from 116 infants in a previous survey (Wynne-Davies 1968). The object was to determine, in normal children, the side of the plagiocephaly, the age of onset and the relationship to the season of year and social class of the family.

RESULTS

Clinical findings.—The expected ratio of three males to two females was found, with a preponderance of left-sided curves (76 per cent); 98 per cent were thoracic. Of curves developing during the first six months, 64 per cent resolved, but it is again stressed that these are figures from a specialist scoliosis clinic. The true numbers with resolving scoliosis will be higher. Only six curves were present at birth; ninety-one developed between one and six months and the remaining thirty-seven between seven months and three years.

Plagiocephaly was present in all ninety-seven infants, the "recessed" side agreeing always with the side of the convexity of the curve. The next most common associated defect was mental retardation, but in infants it occurred only in males with progressive scoliosis, 13 per cent being affected. No female infant and no child with resolving scoliosis was mentally retarded. Congenital dislocation of the hip occurred in 3-5 per cent of all cases and congenital heart disease, usually a septal defect, in 2-5 per cent. These associated anomalies occurred also amongst juveniles and adolescents with scoliosis. The figures were all in excess of those expected from a random survey. There was, however, no excess of these defects among the relatives of patients. The single exception to this was inguinal hernia, which was present in 7-4 per cent of males with infantile scoliosis, in 6-5 per cent of their fathers and in 2-4 per cent of brothers—figures all in excess of the expected figure of around 1 per cent (Knox 1959; Pringle, Butler and Davie 1966).

Genetic findings.—The proportions of relatives of infants with idiopathic scoliosis did not differ significantly from any other group of idiopathic scoliosis patients, being approximately 3 per cent of parents and 3 per cent of sibs, or thirty times those expected for the normal population (Wynne-Davies 1968). The proportions of second and third degree relatives with scoliosis were 0.4 and 0.2 per cent respectively, which is near the figure for the general population.

Epidemiological findings.—There were no significant findings relating to the mother's previous obstetric or menstrual history or to illness during pregnancy. With regard to length of gestation and birth weight of the ninety-seven infants developing scoliosis in the first six months, the females did not differ significantly from normal, but 29 per cent of the males were born under thirty-eight weeks (controls 15 per cent), and their birth weights were significantly less than the figures both for the general population and for their own sibs.
There were only six infants in whom scoliosis was noticed at or very soon after birth, three males and three females. All three males and one of the females were premature (four, five and ten weeks, and one with dates not known but a birth weight under three pounds). Three of the curves resolved and three progressed. It was also noted that five of these patients had associated anomalies indicating some disorder of connective tissue; two had large herniae, one had generalised joint laxity and finger contractures, and two had congenital dislocation of the hip. The sixth infant was ten weeks premature and mentally retarded.

Presentation—There was a significant excess of breech presentations (17.6 per cent) amongst infants developing scoliosis during the first six months of life, the figure dropping sharply to the usual 4 per cent when a curve developed at seven months or more.

Seasonal variation—Of the ninety-seven infants, 64 per cent were born during the second half of the year, July to December, and exactly the same proportion, 64 per cent, developed scoliosis during the two winter quarters (October to March). The ratio in each case was 1:8 to 1, whereas the expected ratio for normal babies born in the first half of the year compared with the second half is equal.

Side of lying in early infancy—Retrospective history-taking is notoriously unreliable and the investigation proved to be equivocal. That is, infants with left-sided or right-sided curves were reported as equally likely to lie on the concavity or on the convexity of the curve, or on their backs. However, the survey did establish that only three of the ninety-seven infants habitually lay prone; all three had resolving scoliosis.

An inspection of maternity hospitals in Boston, Massachusetts (Riseborough 1973) showed that, without exception, all infants lay prone. There is, however, no detailed information relating to their habits after leaving hospital, though it is common knowledge that the prone position is preferred in North America.

Maternal age—There was no significant finding among infants with resolving scoliosis, but the mothers of infants with progressive curves were significantly older than normal, 50 per cent being over thirty years of age compared with the expected 28 per cent.

Social class—In the 121 cases of infantile scoliosis in which the father’s occupation was known, there was a difference which almost reached significance at the 5 per cent level—a greater proportion of infants with resolving scoliosis came from families with higher incomes, and more with progressive scoliosis came from families with lower incomes.

Plagiocephaly in normal infants—Examination of 223 normal infants aged between two weeks and six months showed that 28 per cent had a mild to moderate degree of plagiocephaly, the sexes being equally affected. There was a preponderance of left-sided deformity (83 per cent) and 50 per cent of the children came from poorer families, compared to an “expected” figure of 32 per cent. It was also noted that, of infants examined in the winter months from October to March, 30 per cent were affected, but of those examined in the summer months only 8 per cent showed the deformity.

DISCUSSION

Three anomalies occurring with idiopathic scoliosis—mental retardation, congenital heart disease and congenital dislocation of the hip—were not found amongst the relatives of patients, which suggests there is no common genetic aetiologial factor. If one were present, an excess of these abnormalities should be appearing within scoliosis families. It is possible, therefore, that mental retardation and congenital heart disease themselves predispose an individual to scoliosis.

Congenital dislocation of the hip did not occur amongst the relatives of patients, but inguinal herniae of the congenital type did. Congenital dislocation of the hip is itself of complex aetiology (Wyne-Davies 1970), one feature being a connective tissue disorder evidenced by generalised joint laxity and congenital type herniae, both in patients and relatives. It is possible that genetic factors are involved in a connective tissue disorder, leading to some cases of idiopathic scoliosis, congenital dislocation of the hip and/or herniae. Certainly, scoliosis is not uncommonly a feature of some of the known genetic disorders of connective tissue, such as Marfan’s syndrome, Ehlers-Danlos syndrome and Morquio’s disease.

It is reasonable to suppose that adverse factors during pregnancy and birth can influence events during the early months of life. An excess of breech presentations has been noted in other “positional” deformities, such as congenital dislocation of the hip and torticollis. The reason for the excess of premature males with scoliosis is not clear. If these infant boys are removed from the survey then the sex ratio is equal.

Seasonal variation, shown by the greater numbers born between July and December, together with the excess of curves developing between October and March, suggest that infants are particularly susceptible to the development of scoliosis from the age of three to nine months, and perhaps limitation of free movement, due to the child’s being heavily wrapped up in cold weather, increases this tendency.

Several special features were noted in children with progressive scoliosis. Thirteen per cent of males, but no females, were mentally retarded; the mothers were older than average and the children tended to come from poorer homes than did those whose curves resolved.

It is difficult to know the significance of plagiocephaly in scoliosis. Certainly many otherwise completely normal children develop and then recover from plagiocephaly. The survey of normal infants showed an equal number of males and females affected, but as in scoliosis, there was some excess among lower social classes, in the winter months, and on the left side. Whatever factors lead to plagiocephaly presumably can also lead, less
commonly, to spinal curvature, since in 100 per cent of cases of infantile idiopathic scoliosis plagiocephaly is also present.

It is apparent that infants with scoliosis may have several adverse factors to contend with—prematurity, low birth weight, mental retardation, older mothers, poorer homes, congenital heart disease, and perhaps muscle weakness associated with undue generalised joint laxity as evidenced by congenital dislocation of the hip and herniae. One can speculate that these factors in their external or internal environment lead to relative immobility, and that that leads to retarded, possibly unequal, maturation. Evidence suggests that the "static" or "floppy" baby is particularly at risk of developing scoliosis.

The aetiology of infantile idiopathic scoliosis must be multifactorial, with a genetic tendency to the deformity which can then be "triggered off" in different individuals by different factors, some medical, some themselves genetic and some social. The exact cause in each individual is likely to be different and the balance must vary from patient to patient; thus someone with a strong genetic tendency would need very little "triggering" action. At the other end of the scale, scoliosis can probably be produced in a child with no genetic tendency to the deformity, entirely by adverse environmental factors.

However, these adverse factors must also be present in North America, and the virtual absence of infantile scoliosis there is still unexplained. The difference is very unlikely to be genetic, and therefore one must look for some aspect of the environment of North American infants which is preventing or correcting any tendency to curvature. Clearly, infantile scoliosis is preventable, and if so, why is it not prevented in this country?

Evidence relating to the habitual side of lying in early infancy was equivocal, though in the Edinburgh area it is not prone as it is thought to be in North America. The only three infants in this survey who habitually lay prone had resolving scoliosis. There has been speculation on each side of the Atlantic as to whether infant posture is significant in the development of scoliosis: whether the prone position in some way promotes physical development of the spinal muscles and corrects any tendency to scoliosis at this early age. The significance of infant posture is not proven, and it is only suggested here that the prone position could be relevant. More information is needed from orthopaedic surgeons and paediatricians in different parts of Britain in relation to the frequency of infantile scoliosis and the habits of infant posture in the first few months of life. The results of a current Liverpool survey are awaited with interest (Owen 1974). If indeed the prone position is significant, it should be a simple matter to prevent what is potentially a most crippling deformity.

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REFERENCES


Edinburgh Register of the Newborn, 1964-1968. Held at the Department of Community Medicine, University of Edinburgh.


Riseborough, E. J. (1973) Personal communication.


