Section of Physical Medicine

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Hypermobility

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Familial Joint Laxity

There are a number of disorders which exhibit the physical sign of excessive laxity of joints. It may occur alone, or as part of a clearly defined syndrome, and many different genes can be concerned in producing this one end effect. In addition to demonstrable laxity of joints, a number of associated conditions may be present, one of the commonest being hernia, whether inguinal, femoral, umbilical or at any other site.

My own interest in the subject was promoted by Dr Cedric Carter of the MRC Clinical Genetics Research Unit. He and Mr J Wilkinson carried out a survey of normal school children between the ages of 6 and 11 years in London, looking for excessive joint laxity, and on comparison with a group of children with congenital dislocation of the hip there seemed to be positive evidence that excessive generalized joint laxity was a feature in patients with this deformity (Carter & Wilkinson 1964).

A large genetic survey of congenital dislocation of the hip has been completed in Edinburgh, including not only children with established hip dislocation but also neonates with 'clunking' hips, and an investigation into joint laxity in all patients and their first degree relatives. In addition a survey of joint laxity control was completed in Edinburgh infants and school-children with ages ranging from one week to 18 years. It is common knowledge that laxity of joints diminishes with age. The survey established the rate of this, as well as showing that familial joint laxity is one etiological factor in congenital dislocation of the hip. Details are reported elsewhere (Wynne-Davies 1970*a*, *b*).

In summary, excessive joint laxity was not demonstrable in any child (with or without a dislocated hip) during the first week of life. All children were found to be most lax jointed around the age of 2 to 3 years. In the control group of that age, nearly 50% were affected, but the percentage rapidly declined with age. By 6 years only 5% of children were so affected and by 12 years the figure was under 1%. These results were obtained by taking the composite figure for three pairs of lax joints. Reviewed separately, results were as follows:

Ankles and feet: At birth some 50% of children could dorsiflex their ankles beyond 45 degrees. By the age of 3½ years this was reduced to 5%.

Knees: These were the most stable of the peripheral joints examined. At the age of 18 months, some 30% of children could hyperextend their knees, but by the age of 3½ years this again was reduced to 5%.

Elbows: Approximately one-third of children between the ages of 1 and 2 years could hyperextend their elbows. This diminished slowly until by the age of 7 years it was reduced to about 5%.

Wrists and metacarpal joints: These two regions followed each other closely and were the most frequently affected. Around the age of 2 years, some 65% of all children showed laxity. At the age of 3½ years it was about 40%, and by the age of 7 years about 5%.

Two other points emerged from the control survey. First, under the age of 2 years girls were more often lax jointed than boys. Secondly, and again only under the age of 2 years, children belonging to the higher income group families were more lax jointed than those of the lower social classes (significant at the 5% level).

In the congenital dislocation of the hip survey it was found that the neonatal group had the highest proportion of individuals with joint laxity, very much in excess of controls and also in excess of patients with 'late-diagnosis' dislocation (who were themselves still in excess of controls). Not only was this apparent in the patients, but also in their first degree relatives. It was also noted that a significantly high proportion of neonates with dislocation belonged to high income group families.

Hernia in Congenital Dislocation of the Hip

Enquiry was also made regarding hernia both in the index patients and in their families. The figure for inguinal hernia in males to the age of 15 years is usually quoted as about 9 per 1,000 or nearly 1%. In the congenital dislocation of the hip survey the figure for males was about 7% and it must be remembered that many children were not yet 15 years of age. It was also interesting to note that inguinal herniæ occurred in about 5% of their fathers and brothers.

Thus, one etiological factor in congenital dislocation of the hip is likely to be familial hypermobility of joints and this is probably a dominant trait. It is particularly a feature of neonatal dislocation but is also present in many cases of dislocation presenting at a later age.

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Is Hypermobility a Discrete Entity?

Hypermobility of joints is characteristic of the Ehlers-Danlos syndrome, and is the hallmark of the hypermobility syndrome described by Kirk *et al.* (1967). The implication of these statements is that hypermobility is a discrete entity. In other words, that people either exhibit hypermobility of joints or they do not. The other side of this coin is that limitation of motion is also a discrete phenomenon. I wish to question the validity of these concepts.

Conceptual appreciation in medicine undergoes a sequential development as far as biological characteristics are concerned. Cochrane has represented this process graphically (Wood 1971, Fig 1). When a characteristic is first linked with a disease, it is often concluded that the diseased and non-diseased states are qualitatively different from each other with regard to the characteristic. The everyday necessity of making a binary decision, treatment required or not required, predisposes to such a simplistic view.

With the passage of time the stark dichotomy gets eroded by gradual appreciation that there are distributions of the characteristic, both in the diseased and in the nondiseased. Finally, it is conceded that the characteristic is distributed continuously, so great is the overlap between the two states. However, there persists a reluctance to discard the notion that underlying the skewed distribution are the two discriminated occurrences.

This whole pattern of development reflects stages in the progressive acquisition of knowledge, as experience increases. To bring this pattern to life you have only to remember the celebrated controversy over the nature of hypertension, with which the names of Platt (1959) and Pickering (1963) are associated – is hypertension a discrete entity or only the extreme of a distribution? A similar conflict arose about the significance of hyperuricæmia (Neel 1968). The epidemiologist, by examining a representative sample, is protected from the intermediate biases.

My interest in the distribution of mobility in a joint was sparked off by observations on the Ehlers-Danlos syndrome. Two population samples of females in Buffalo, USA, were examined with my colleagues Floyd Green and David Sackett (Green *et al.* 1965), and the data in this report are derived from this work. However, I have comparable data from a British sample of both sexes in the Rhondda Fach, and in general the findings were similar in this group.

We restricted our attention to joints that move in only one plane, and the bulk of our data relates to the elbow and the interphalangeal joints of the upper limb. We made preliminary studies with a goniometer, but our alignment of the arms of this instrument in relation to the axis of the limb showed unacceptable variability. Inter-observer variation was much less with judgments made by standardized procedures and recorded on a seven-point ordinal scale (Fig 1).

The elbow provides a good example (Table 1). Although the neutral position was the limit of passive extension in more than half the individuals, the pattern of this distribution is nevertheless within the family described as normal or Gaussian. Two important conclusions stem from these observations. First, the mobility of a joint is a continuously distributed variable. In other words, neither hypermobility nor limitation of motion are discrete phenomena. Furthermore, in

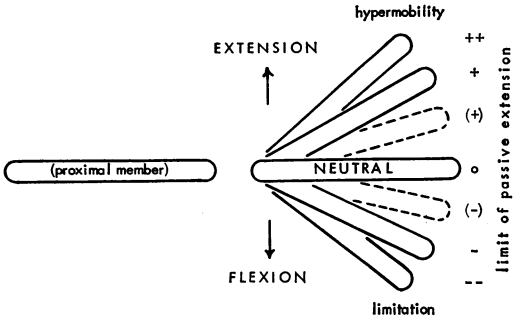

Fig 1 *Mobility of a joint which moves in only one plane, recorded on a seven-point ordinal scale. The neutral position was categorized as zero and departures from this as doubtful (symbol in parentheses), definite (single symbol) and marked (double symbol), hypermobility being indicated by plus and limitation by minus*