Lax ligament syndrome in children associated with blue sclera and bat ears

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SUMMARY. The child that is slow to walk causes concern. When cerebral palsy, mental retardation and muscular dystrophy have been excluded, what remains? Thirty five children (19 boys and 16 girls) with hypermobile joints, blue sclera and bat ears (the 'lax ligament syndrome') were referred by general practitioners to a general paediatric outpatient clinic over two years. Three were referred in the first three months of life because of clicking hips; 14 children aged one to two years, had delayed milestones of motor development and exhibited bottom shuffling; 10 children aged four to five years presented with 'growing pains' or 'funny gait' and eight older children had multiple minor complaints.

The lax ligament syndrome is a comparatively common mild collagenopathy. It may well come to light on routine surveillance in general practice. It is dominantly inherited and improves with time; management is therefore expectant and symptomatic. A firm and reassuring diagnosis can be given which saves both anxiety and investigations.

Introduction

'CHILDREN begin self-movement when they can crawl and especially when they can walk. But it is not good to compel them lest their legs become bent' (Galen). It is, however, worrying when a child does not begin to walk at the appropriate time. The child with lax ligaments frequently presents in this way, and the purpose of this paper is to describe and to review this condition.

Children with lax ligaments have the triad of hypermobile joints, blue sclera and bat ears. The syndrome lies on the spectrum ranging from normality, through simple joint hypermobility to the Ehlers–Danlos syndromes.

Method

A total of 35 children with the triad of signs of lax ligament syndrome presented to a district general hospital outpatients department in Surrey over the two year period January 1987 to January 1989. The first nine children together with their parents were examined in greater detail than the remaining 26.

Joint laxity was assessed by Beighton's modification of the Carter and Wilkinson method.[1] Patients were considered to be hypermobile if they could meet at least three of the five criteria: (1) extension of the wrist and metacarpal phalanges so that the fingers are parallel to the dorsum of the forearm; (2) passive apposition of the thumb to the flexor aspect of the forearm; (3) hyperextension of elbows by more than 10°; (4) hyperextension of knees by more than 10°; (5) flexion of trunk with knees extended so that palms rest on floor.

The blueness of the sclera was estimated on an arbitrary scale of 0–5 by the author and independently by a consultant geneticist. The skin was classified as normal, lax, elastic or soft. Ears were measured and tested by tweaking. Collagen biopsies were performed on two patients and their similarly affected mothers for lysyl oxidase level and collagen growth patterns. All the boys in the group of 35 children had creatinine phosphokinase levels measured, and karyotypes examined for fragile X sites. Birth centiles and current centiles for height, weight and head circumference were plotted. A full developmental and family history was taken for all 35 children.

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Submitted: 23 June 1989; accepted: 9 November 1989.

Mode of presentation

In this series of 35 children recognized to have the lax ligament syndrome 19 were boys and 16 girls. The age range at presentation was two months to 10 years (median 3.3 years). There was no appreciable difference in age of presentation between girls and boys. The ages of presentation are shown in Table 1. There were differing symptoms according to age but similar signs.

The clinical picture was of a child between the 50th and 90th centiles for height and head circumference, with weight nearer the 50th. Epicanthic folds were present in most of the children until they reached the age of two years. Sclera were blue (grade 2–4) but tended to fade after the age of seven years. The ears were prominent and unfolded; they were also remarkably soft, and could be easily tweaked and bent over, like those of a rabbit. The cheeks tended to be full, and the children often appeared younger than their chronological age. The palate was normal, the anterior fontanelle closed at around 18 months, and the teeth were normal, with no enamel problems. The skin could be described as soft, but not lax or elastic. Six children had prominent veins over the chest wall. The majority were described as bruising easily, but there was no increased skin fragility, and scars healed normally. No mitral valve murmurs were elicited, neither were any hernias found. However, the characteristic posture was lordotic, with very flat feet, and a 'toddler tummy' persisted in most children until the age of five years.

Complications

Complications observed in the 35 children were mostly those of the hypermobility syndrome.[2] Recurrent dislocation of the

Table 1. Presentation of lax ligament syndrome according to age.

Age	Number of children	Presentation	Comments
1–3 months	2M 1F	Clicking hips, head lag	No CDH. Slithered through examiners hands
1–2 years	7M 7F	Delayed motor milestones	Girls walked alone at mean of 17 months (range 13–25 months) Boys walked alone at 24 months (12–42 months) 4 bottom shuffled
4–5 years	5M 5F	'Growing pains' 'Funny gait'	'Keeps falling over' 2 girls had ureteric reflux 6 were constipated
6–9 years	5M 3F	Multiple minor complaints	Retrospectively delayed motor milestones 'Growing pains'

CDH = congenital dislocation of the hip.

elbow and shoulder occurred in two girls, especially when they were lifted up by one hand. Minor trauma was universal, and every child, once mobile, had more than five bruises per leg from frequent tumbles. Effusions[3] were common in the older children, and three mothers had noted crepitus.[4] They all had flat feet with valgus heels. Forefoot adduction was present in three children, which compounded the tendency to trip.

It appeared that the boys had more problems than the girls up to the age of about seven years, when the girls appeared to have more problems, especially around the knee.[5] As well as delayed milestones of motor development, four of the boys had delayed, unclear speech until aged three years. This added to the impression of mental retardation, although all were alert, had good attention spans, and scored well on the personal/social side of Denver assessments. Visual/spatial orientation is normal in children with this syndrome, but difficulties may be experienced with fine motor control as the fingers and thumb bend back when gripping pens and forks.

Another problem, again more evident in the boys than the girls, was late toilet training. Most tended to have constipation, and did not appear to be able to control their bowels until aged three to four years. This may, like oesophageal and ureteric reflux, be an effect of low conduit tone.

Growing pains, that is 'intermittant pains localized deeply in the arms and legs, occasionally accompanied by sensations of restlessness, but never by tenderness, redness or local swelling'[6] were common, both in the patients and their siblings. The pains mainly occurred in the late afternoon and at bedtime.[7] Sometimes there was a clear relationship with over-use, but often there was no obvious precipitating cause.

Interestingly, the similarly affected mothers of the children with the lax ligament syndrome all had quick and easy labours.

Treatment

The lax ligament syndrome improves with time and many of the more worrying signs have improved by the time the child is seven years old. Management is therefore expectant and symptomatic. The important thing is to recognize the syndrome and exclude the more severe collagenopathies, cerebral palsy, muscular dystrophy and mental retardation. A firm diagnosis and reassurance as to the absence of serious disease saves both anxiety and investigations. The hypermobile joints should be demonstrated, the complaints explained and a simplified account of collagen formation offered along the lines that the child's type of collagen provides more stretch than strength at the moment, but more cross linkages will form with time, and the symptoms will improve. Parents are usually comforted by finding that they, and other close relatives have the same condition and it can explain a lot of minor symptoms. It is worthwhile to encourage them to take the child swimming and to buy properly supportive shoes.

Differential diagnosis

Varying degrees of bat ears, blue sclera and ligament laxity[8] are common in early childhood. The lax ligament syndrome can be diagnosed by the doctor by observing the child walking, or rather bottom shuffling, through the door and by noting the combination of unfolded, sticking-out ears, full 'hamster' cheeks and blue sclera (Figure 1).

It is very important to differentiate this innocuous clinical picture from other causes of hypotonia. First, one should check that the child is not weak and hypotonic. In lower motor neurone lesions the reflexes will be reduced or absent. In muscular disorders the power will be reduced and the child unable to keep a leg or head up at 45° from the horizontal for any length of time. The creatine phosphokinase level should always be measured in a boy of 18 months who is not walking, to eliminate Duchenne muscular dystrophy. The level was within the normal range for all the boys in this study.

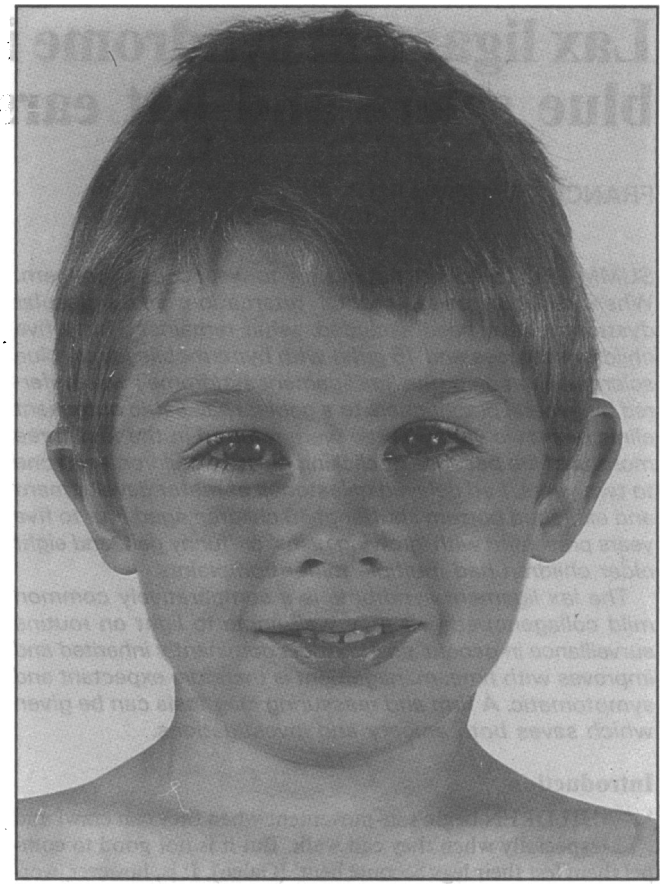

Figure 1. *Typical boy with lax ligament syndrome; note soft bat ears and full cheeks. He had blue sclera at the age of five years.*

Floppiness can also be due to mental retardation, so other dysmorphic features should be looked for. Early cerebral palsy may present with hypotonia. The reflexes however will be increased and the primitive responses will persist. The major collagenopathies of the Ehlers–Danlos syndromes are the other major conditions to consider. Type I (gravis), II (mitis), two hypermobile types-III (benign) and VII (severe) are the most likely candidates. The cardinal clinical features of the Ehlers–Danlos syndromes[9] are skin hyperextensibility, skin fragility (easy splitting of the dermis leading to thin shiny atrophic scars), easy bruising and hypermobile joints. In the children with lax ligaments studied here and their similarly affected parents the skin was soft, but not stretchy or elastic. Some children did bruise easily and had frequent tumbles, but there was no evidence of cutaneous fragility, and scars, when they occurred, were normal.

In the fragile X syndrome, the mentally retarded males have prominent ears, macroorchidism, attention deficit and language delay; 20–60% have hyperextensible finger joints and flat feet. Some have loose stretchable skin owing to increased fragmentation of elastin.[10] Fragile X sites were not found in any of the boys who were tested in this study, but should be sought in boys who appear to have lax ligament syndrome and delayed speech. Marfans syndrome should be considered if the child or parent is exceptionally tall or slender. The hypermobile joint syndrome,[2,4,11,12] which may lead to joint instability and early osteoarthritis, is well described in the literature. In none of the published papers are the clinical features of blue sclera or bat

ears mentioned, in fact several[12,11,13,14] make a point of noting that there are no facial abnormalities.

Inheritance of syndrome

A positive family history was found in 90% of the 35 cases in this study. Transmission from male to male was observed. Histories of late walking, bottom shuffling, and double jointedness were commonly obtained and affected individuals had simple sticking out ears. Blue sclera tend to fade with time, although several of the mothers still had pale blue sclera (grade 1–2). The lax ligament syndrome appears to be a mild, dominantly inherited collagenopathy, of as yet undefined type. In the two patients and their affected mothers receiving biopsy, the collagen appeared ultrastructurally normal, and the lysyl oxidase enzyme levels were normal (Pope M, personal communication). Collagen forms a tightly coiled triple helix with cross linkages between hydroxylated lysines.[15] Ten main collagen types have been identified so far, and more are likely as gene mutations are identified. A small type I collagen mutation is a possible cause of lax ligament syndrome, giving rise to a mild connective tissue defect.

Conclusion

Thirty five children were observed in a general outpatient clinic to have a triad of signs: bat ears, blue sclera and joint hypermobility. Many presented with delayed milestones of motor development, as has previously been observed in the articular hypermobility syndrome.[13] However, the facial signs have not been recorded in the hypermobility syndrome, and this fact together with a soft, but not elastic skin, permit a rapid and reassuring diagnosis of the lax ligament syndrome.

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Acknowledgements

I wish to thank Dr M Patten for additional examination of the first nine patients, Dr M Pope for examination of the collagen biopsies and David Pearce for help with computing and statistics.

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