Benign joint hypermobility in childhood

Children inherently have a greater range of joint motion than adults, the prevalence of hypermobility, as defined by several criteria, varying in different populations from 5 to 30% [1–5]. This variation probably represents ethnic differences, but also the different ages at which joint examinations were undertaken and the different populations chosen for study. The frequency of musculoskeletal disorders (MSDs) arising from such hypermobility in childhood is quite variable, both across populations and within individuals. Some studies have suggested a definite causal link between hypermobility of joints and MSDs, but others have not found such a link [6–13]. In paediatric services, and in particular paediatric rheumatology services, a referral bias is undoubted, and studies including control groups of healthy asymptomatic individuals with and without hypermobility are essential to fully establish such links. Nonetheless, the body of evidence suggests that hypermobility is associated with significant MSDs in childhood that lead to consultation with medical practitioners and health-care providers. We believe that manifestations may occur at any age, with a spectrum of potential disorders ranging from congenital dislocation of the hip at birth, through growing pains in early school age children to back pain and occasionally spondylolysis in teenagers.

When studying such problems, it is important to be clear that the presence of hypermobility in children does not equate to having the benign joint hypermobility syndrome (BJHS). The latter is defined as the presence of a degree of joint hypermobility measured by a prearranged and validated scoring system, associated with musculoskeletal symptoms and signs, and other connective tissue problems likely to be attributable to it [14]. Hypermobility of joints may be considered a physiological or pathological phenomenon in this context. Why certain children will have MSD associated with hypermobility and others not is unclear, but the
more extreme degree of joint laxity and the associated clinical features (such as skin elasticity, easy bruising or slow tissue healing) in children with MSDs may indicate that inherent differences are important. Children with more easily definable disorders, such as Ehlers–Danlos syndrome and Marfan syndrome, have long been recognized as having potentially pathological consequences of hypermobility. Other conditions or syndromes, such as osteogenesis imperfecta, Down syndrome and Stickler syndrome, are also associated with hypermobility of joints, which may be symptomatic [15]. It is now believed that mild, unrecognized or *forme fruste* variants of these and other conditions may occur and be associated with hypermobility and attendant MSDs [6]. There may be considerable overlap between such children and those who are perhaps towards the end of the normal Gaussian distribution curve of joint mobility. It is postulated that the characteristics who are described as having the BJHS are not identical and may have quite different body morphology and associated clinical characteristics. It is likely, perhaps, that hypermobility of joints, or indeed the BJHS, represents a complex genetic trait with multiple genes contributing to the phenotype and degree of hypermobility. Implicit in this idea is that contributions to a child’s joint range of motion may come from both paternal and maternal sides of the family, as so often appears to be the case in the clinical situation.

As early as birth, specific problems have been associated with hypermobility, including congenital dislocation of the hip. Congenital (benign) hypotonia or ‘floppy infant syndrome’ and hypermobility of the joints have been recognized by a number of researchers. Delayed motor development, which may be persistent, and laxity of ligaments have been documented in several studies, but some researchers have not found such a link [16–19]. This may relate to other research suggesting diminished proprioception in individuals with joint hypermobility. Benign nocturnal leg pains, or ‘growing pains’, are a common pain syndrome of childhood and have been linked to underlying joint hypermobility in some children. We would postulate that unusual or excessive exercise leads to minor injury or repetitive strain to musculoskeletal or ligamentous structures in the lower limbs, which are noticed when children are at rest in the evenings. The characteristic age of prevalence, between 3 and 6 yr, is not the age of most rapid growth in childhood, but may represent a critical time of changes in physical demands and activities, coincident with important changes in body morphology, such as muscle power, balance and ligamentous support. Underlying hypermobility may contribute, therefore, to the appearance of symptoms. One of the most common complaints seen in paediatric rheumatology clinics is recurrent foot/ankle and knee pains that are believed to be related to hypermobility, and it is not uncommon to obtain a history from the parents during early childhood of excessively flat or pronated feet or abnormal gait patterns due to femoral anteverision (‘knock-knees’). Generalized hypermobility of the joints may often be seen in such children, though symptoms as such may be quite variable. Some children may relate such symptoms to specific physical activities but others do not. Parents and children often report brief episodes of joint swelling, which usually occur after activities and resolve within a few days. This pattern of symptoms has been referred to as juvenile episodic arthralgia/ arthritis by some authors. As children become older, recurrent injuries related to sporting activities increase. It is not uncommon to gain a history of a child who has been proficient at sport, gymnastics or ballet who has had to give up participation because of musculoskeletal symptoms or problems increasing at the time of increased demands of training and frequency of competition. Still other children often report being clumsy in early childhood, and having difficulties in any participation in physical or sporting activities. Subluxation of the radial head from the annular ligament, the so-called pulled elbow, has been linked with joint hypermobility in one study [20].

As children progress through primary school with increased demands on handwriting skills, children who are hypermobile develop upper limb problems. Not infrequently, pain and fatigue in the hand, wrist or lower arm are noted, particularly by teachers, who may also note that unusual or bizarre hand postures are assumed for writing tasks, with the relatively less stable hand in hypermobility. Parents may report symptoms when the child returns from school in the afternoon, and there may be a history of difficulty with tasks such as music lessons in some children as the tasks become increasingly complex with age.

Back pain has been linked to hypermobility in childhood, particularly during the adolescent years. A number of studies have suggested a causal link between chronic or recurrent back pain and generalized hypermobility [21, 22]. The symptoms become more frequent from 10 yr of age and older, perhaps coincident with increasing physical and sporting activities. The pain is usually lumbar and often enough to limit activities at home and school. The typical adolescent has an exaggerated lumbar lordosis and compensated thoracic kyphosis, giving the typical round-shouldered appearance. In some patients spondylolysis or spondylolisthesis may be seen, and is associated with more severe disabling back pain. Hip pain can be seen in this age group, particularly in those involved in gymnastics or dance, and may be due to subluxation or actual dislocation episodes in some. The authors have also noted, in a cohort of adolescent patients with reflex sympathetic dystrophy, an increased prevalence (25%) of generalized hypermobility (K. J. Murray, personal communication). The joint laxity is presumed to be an important predisposing factor in a young person who may be predisposed to this condition for other reasons.

Other chronic musculoskeletal pain syndromes have been linked to hypermobility in childhood. A number of authors have documented the co-occurrence of fibromyalgia or a fibromyalgia-type pain pattern with generalized joint hypermobility in schoolchildren. It is
perhaps understandable that chronic underlying arthralgia and musculoskeletal symptoms can be associated with sleep disturbance and the development of more generalized pain disorders in the vulnerable adolescent. Recurrent temporomandibular joint problems have been recognized in adolescents; these have been related to underlying hypermobility in at least two studies [23, 24].

The successful management of patients with BJHS includes early recognition of joint laxity as being related to the symptom complex with which the child may present at different ages, before the symptoms may become chronic and perhaps disabling. Education of the parents as to the nature of the condition and defusing anxiety about less benign conditions is important. Similarly, advice to schoolteachers and sporting coaches may be critical in improving symptoms and allowing more gradual rehabilitation and return to full activities or activities that are less likely to lead to recurrent joint injury. Appropriate intervention with physiotherapy and occupational therapy can be important for the optimum management of problems. The intervention may also include psychological support or counselling if the child has developed a chronic pain syndrome complex.

In summary, the BJHS is a common cause of musculoskeletal complaints in paediatric practice, with quite well-described patterns of symptoms and signs, and early recognition is likely to improve the outcome. Further understanding of the genetics of connective tissue integrity and laxity is likely to yield better understanding of the physiological and pathological variants that no doubt underlie the variable modes of presentation and phenotype seen in the children.

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References