ScienceDirect

Available online at www.indianjrheumatol.com and www.sciencedirect.com

Review Article

Benign joint hypermobility syndrome

Able Lawrence

Department of Clinical Immunology, SGPGIMS, Lucknow, India

Keywords:
Benign joint hypermobility syndrome
Therapeutic exercise

ABSTRACT

Benign joint hypermobility syndrome is the presence of musculoskeletal symptoms in subjects with joint hypermobility in the absence of demonstrable systemic rheumatic disease. Unlike the heritable disorders of connective tissue with which it shares considerable overlap in manifestations, most subjects with hypermobility remain asymptomatic. Prevalence of hypermobility varies considerably with age, gender and ethnicity. Muscle weakness and decreased proprioception contribute to recurrent micro trauma and joint pains in this otherwise benign condition. Supervised exercises designed to improve joint stability and proprioception remain the mainstay of treatment.

1. Introduction

Exaggerated flexibility of joints beyond the normal range occurs in certain individuals and had been recognised from ancient times.[1] While joint hypermobility is a feature of several inherited diseases of connective tissue such as Osteogenesis imperfecta, Marfan's syndrome and Ehlers Danlos syndrome,[2] it is also present in a subset of otherwise normal individuals. Joint hypermobility in such normal subjects is termed benign to distinguish it from the more serious heritable disorders of connective tissue.[3] Presence of musculoskeletal symptoms in subjects with hypermobility in the absence of demonstrable systemic disease is termed benign joint hypermobility syndrome. However, hypermobile type Ehlers Danlos syndrome can overlap and may be indistinguishable from benign hypermobility syndrome.[3]

Prevalence of joint hypermobility varies based on age, sex and ethnicity. It is common in children and decreases with age.[4,5] Men have less hypermobility than women.[6] There are wide variations between different ethnic groups across and within regions. While Caucasian boys and girls have 12.9% and 40.5% prevalence of hypermobility, among adults it is only 10%. While Nigerian undergraduate students had hypermobility of 12.9% (8% in males and 17% in females),[7] the prevalence was higher among the Yoruba population in south western Nigeria at 43% (35% in males and 57% in females).[8] In a study among children aged 3 to 19 from Mumbai, 58% had hypermobility[5] while 91% medical students from Kerala had Beighton score of 4/9 or more[1] underscoring the wide variation in the prevalence of hypermobility between different communities. Despite the widely varying prevalence of hypermobility as a trait in different communities, and the higher rates of musculoskeletal pains, the vast majority are asymptomatic[5] highlighting the benign nature of the condition.

Unlike heritable disorders of connective tissue (HDCT), the biochemical aetiology of benign joint hypermobility syndrome (BJHS) is unknown. Despite its benign nature in comparison with the HDCT, there is considerable clinical overlap.[9] While the vascular type Ehlers Danlos syndrome due to Tenascin X deficiency is autosomal recessive, heterozygous carriers have variable amount of joint hypermobility indistinguishable from

E-mail address: abledoc@gmail.com.
http://dx.doi.org/10.1016/j.injr.2014.09.009

benign hypermobility.[10] However, defects in Tenascin X have not been demonstrated in the vast majority of patients with benign joint hypermobility. Prolidase is an enzyme involved in collagen synthesis and catabolism of collagen and is primarily involved in Proline recycling.[11] Although patients with benign hypermobility were demonstrated to have lower serum prolidase activity,[11] it is not clear whether the relative deficiency is the cause or a result of decreased collagen and connective tissues. Unlike patients with heritable disorders of connective tissue, subjects with benign hypermobility lack demonstrable structural abnormalities in tendons and ligaments.[12] Most patients with symptomatic joint hypermobility have generalized or localized decrease in muscle bulk. Many patients recall onset of symptoms after a systemic illness, suggesting that loss of muscle mass and deconditioning might be contributing to the clinical syndrome. Abnormalities of joint proprioception are found in patients with symptomatic joint hypermobility. Joint hyperlaxity and diminished joint proprioception coupled with poor muscle bulk and tone may contribute to the risk of recurrent microtrauma and joint pains.

2. Clinical manifestations

Patients with BJHS present with chronic or recurrent pain in one more joints. In community studies, subjects with joint hypermobility are more likely to have joint pains.[5] Typically patients with joint hypermobility have recurrent episodes of joint pains or have joints pains precipitated by physical activity.[13] The joint pains are a result of unrecognized microtrauma. Besides joint pains, patients present with enthesitis, bursitis, tenosynovitis, chondromalacia patellae, rotator cuff problems and mechanical back pain. Among patients with soft tissue rheumatism complaints, presence of joint hypermobility has been linked to younger age at presentation and recurrent or multiple lesions.[14] Patients are at increased risk of recurrent joint dislocations, subluxations and sprains as a result of joint instability. Some patients may develop correctible deformities of joints without ever suffering from arthritis[1] and sometimes correctible deformities can result from transient and self-limited arthritis. Hypermobility is associated with higher risk of postural or mechanical back pain in professions that require prolonged sitting or standing while it is protective for those who have to frequently change positions.[15] Some patients may develop chronic low grade synovitis as a consequence of recurrent low grade trauma which may be misinterpreted as inflammatory arthritis.[16]

Several studies have noted the association of joint hypermobility with primary fibromyalgia. Patients with fibromyalgia have higher prevalence of hypermobility in comparison to controls besides having higher mean Beighton scores.[17,18] Interestingly the prevalence of hypermobility was higher among patients who were diagnosed as fibromyalgia[19] but did not fulfil ACR 1990 criteria,[20] suggesting that there might be considerable overlap between the two conditions. Patient with fibromyalgia may also be at increased risk of overuse syndromes. Among professional dancers, musculoskeletal complaints were more among those with hypermobility than others despite equivalent training.[21] Hypermobile subjects have lower bone mineral density compared to controls,[22] which predispose them to fractures. Besides bones and joints, these patients may have other extra articular manifestations such as mitral valve prolapse,[23] hernias[24] and prolapse of rectum[25] and uterus. Diminished stiffness of vasculature predisposes to risk of varicose veins[26] and eye involvement may lead to high myopia.

3. Diagnosis

Joint hypermobility is assessed at nine genetically determined sites for the modified Beighton score (Table 1).[1,27] A diagnosis of BJHS is made when the patient has pain in multiple joints along with generalized hypermobility (Beighton > 3) and other secondary causes of joint pain are excluded. A revised (Brighton 1998) criteria[1,28] has been proposed for the classification of BJHS (Table 2). Evaluation should be directed at exclusion of other heritable disorders of connective tissues that are associated with hypermobility including Ehlers Danlos syndrome, Osteogenesis imperfecta and Marfan's syndrome[2] besides other systemic diseases associated with joint pains including malignancies or rheumatological diseases like fibromyalgia.[13] Although a score of 4/9 or more is considered significant, some patients may have symptoms with fewer joints involved. However since hypermobility trait is very common in several populations including India, other causes should be actively sought before attributing the symptoms to joint hypermobility.

4. Management of BJHS

Most patients with benign joint hypermobility are concerned and apprehensive about their illness and would be relieved to know that they do not suffer from any systemic illness requiring therapy. Sympathetic counselling and reassurance goes a long way towards relieving anxiety and reducing maladaptive behaviour. Avoidance of physical activity and exercise leads to further decreased muscle mass and deconditioning. Targeted exercise therapy is therefore the mainstay of management of BJHS. Patients with hypermobility syndrome have decreased joint proprioception compared to normal population.[29] The muscles around joints can be classified into stabilizers close to the inside responsible for proper alignment and stability and prime movers providing the strength for different active movements. Weakness of stabilizer muscles is a common finding among

Table 1 – Modified Beighton score. Hypermobility present if total score >3.

Assessment site	Right	Left
Hyperextension of elbow >10°	1	1
Thumb touching the forearm	1	1
Hyperextension of 5th MCP joint >90°	1	1
Hyperextension of knee joint >10°	1	1
Palm of hands touching flat on the ground with knees extended	1	