Hypermobility Syndrome

K Krishna*, AG Diwan**, Navtej Singh***, Rishikesh Desai***

Abstract

Three cases of Hypermobility Syndrome (HS) are discussed. The first case was a young female, aged 24, with musculoskeletal symptoms since 5 years. Second was an 18 year old male with similar symptoms since 3 years. The third was an elderly female who presented with knee joint osteoarthritis (OA) and apparent but reducible deformities of hands mimicking rheumatoid arthritis (RA). All three patients had hypermobility of joints in absence of demonstrable systemic rheumatic disease. The prevalence, clinical features and management of the entity is discussed. An increased awareness of this condition among physicians is warranted as some patients may be erroneously diagnosed as RA/SLE and may be put on DMARDs and steroids.

Introduction

Generalised hypermobility in the absence of systemic disease is a common condition that has a prevalence of 4-13% in the general population. Children have higher laxity of joints which diminishes gradually during adolescence and adulthood. At any given age, females have a greater degree of joint laxity. It also varies among ethnic groups – more common in Africans, Asians and Middle-East descent.1 Hypermobility of joints is also seen in Marfan’s syndrome and Ehler Danlos syndrome (EDS).

The term “hypermobility syndrome” (HS) was coined by Kirke et al to denote the presence of rheumatic symptoms in otherwise healthy subjects in whom generalised joint laxity is the only observed abnormality.1 However, the condition had been first described by Hippocratica in Scythian warriors, who were warrior tribesmen from Central Asia who invaded India and established their kingdoms.2 Detailed history, careful clinical examination and the modified Beighton score (Table 2) and Brighton criteria (Table 3) aid in the accurate diagnosis.2,3

Case Report

Case 1

A 24 year old female student, presented with multiple joint pains and low backache since 5 years. The pain occurred at any time of the day or night, sometimes disabling. There were 3-10 attacks of severe pain in a month, lasting for 2-4 hours, responding to analgesics and rest. Pain did not occur daily, and some days were better than the others. She had been diagnosed as spondyloarthropathy and had been on various NSAIDs and DMARDs (hydroxychloroquine, sulphasalazine), steroids and calcium supplements, with no relief. On examination of the musculoskeletal system, there was no joint swelling, tenderness, deformity or restriction of movement; no tender points for fibromyalgia were 16 out of 18. Complete back flexion was possible; Schoeber’s test was negative. There was no tenderness of sacroiliac joints. There was hypermobility of many joints and the Beighton score was 8/9 (Figures 1 and 2). Investigations revealed an ESR of 15 mm, negative rheumatoid factor and anti-CCP antibodies, CRP 0.5 mg/l, HLA B27 negative. Haemogram and biochemical tests were within normal limits. All radiographs of various joints, X-Ray and MRI of sacroiliac joint were normal. On the
basis of clinical findings, Beighton score of 8/9 and normal investigations, a diagnosis of HS was made. The DMARDs were omitted, she was counselled, advised supervised physiotherapy and she is doing well.

Case 2

A male aged 18 years presented with pain in elbows, fingers and knees since last 3 years and low backache since 1 year. Pain was more after he played badminton or football. The attacks were for a few days every fortnight and prevented him from playing for 4-5 days. He also had been investigated extensively and despite normal reports was put on Sulphasalazine and Indomethacin, without any relief. On examination, he had hyperextensibility of all joints; no swelling, tenderness or deformity. Beighton score was 8/9, and 2 major Brighton criteria. He was also diagnosed to have BHS, counselled and advised physiotherapy, besides precautions during sports activities.

Case 3

A 65 year old lady presented with pain in both knee joints since 10 years and in small joints of the hands since 5 years. She had been diagnosed as Rheumatoid arthritis, put on Methotrexate 7.5 mg once a week, since the last 3 years, without any relief. On examination of outstretched hands, she appeared to have hyperextension of PIP and flexion of DIP joints, like swan neck deformities, but these were completely reducible; there was no swelling, tenderness or restriction of movement. On the contrary, there was hypermobility of all the joints, excepting knees, quite amazing for her age; a Beighton score of 7/9 (Figure 3), and 2 major Brighton criteria. Knees showed varus deformity and crepitus, due to prolonged osteoarthritis. Radiographs of the hands showed just mild osteopenia and of the knees were suggestive of osteoarthritis. She was diagnosed to have BHS, with osteoarthritis of the knee joints, advised calcium supplements, physiotherapy. Unlike the previous 2 cases, this patient never complained of any joint pain until 10 years before and that was because of osteoarthritis.

Discussion

Hypermobility syndrome can present with a variety of musculoskeletal complaints. The most common symptom is joint pain, especially knee joint which often develops after physical activities or sports during which the affected joint(s) is/are used.
Table 1: Five part questionnaire for recognising hypermobility

- Can you now (or could you ever) place your hands flat on the floor without bending your knees?
- Can you now (or could you ever) bend your thumb to touch your forearm?
- As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
- As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
- Do you consider yourself double jointed?

Table 2: Modified Beighton Score

<table>
<thead>
<tr>
<th>Assessment site</th>
<th>Right</th>
<th>Left</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hyperextension of elbow &gt; 10°</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Thumb touching the forearm</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Hyperextension of 5th MCP joint</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Hyperextension of knee joint &gt; 10°</td>
<td>1</td>
<td>1</td>
</tr>
<tr>
<td>Hands touching the floor with full palms with knees extended (forward flexion of the trunk)</td>
<td>9</td>
<td></td>
</tr>
</tbody>
</table>

Maximum possible score 9

repeatedly; like an over-use syndrome, as in our 2nd patient. Pain may be localised to one or several joints or it may be generalised and symmetric. Primary care physicians can use the 5 part questionnaire as an aid in recognising hypermobility (Table 1). In some cases, the onset of symptoms is preceded by growth spurt, and affected females often report premenstrual exacerbations. Many affected children outgrow their symptoms during adolescence or adulthood, and women may experience fewer symptoms following menopause. Since they are related to activity, the symptoms tend to occur late in the day. Morning stiffness is an uncommon finding. The ligament laxity and joint instability leads to recurrent dislocations, subluxations and sprains. Some develop correctable deformities, including swan – neck deformities of fingers, hallux valgus and varus deformities of feet in absence of arthritis, but can be mistaken as rheumatoid arthritis like our 3rd case. Because joints stiffen with age, the natural history of HS is typically one of improvement, with progressively lessening degrees of joint laxity and associated musculoskeletal symptoms. However, over many years, joint hypermobility can predispose to osteoarthritis (as our 3rd case), even prematurely, in the 4th or 5th decade followed by eventual chondrocalcinosis. HS can also be associated with fibromyalgia syndrome, as our 1st patient.

Determining Beighton score (Table 2) is essential for making a diagnosis. This can be calculated by doing 5 simple manoeuvres that can be completed in 45-60 seconds. A Beighton score of 4 or more points is considered indicative of generalised joint laxity. But this score is too insensitive for diagnosing BHS. So the Brighton Criteria (Table 3) were developed to establish diagnostic criteria for BHS; into this the Beighton score has been incorporated. BHS is diagnosed in presence of 2 major criteria; 1 major criterion plus 2 minor criteria. Two minor criteria will suffice where there is an unequivocally affected 1st degree relative. The syndrome is excluded by the presence of Marfan’s or EDS (other than the hypermobility type of EDS) as defined by Ghent 1996 and Villerfranche 1998 criteria respectively. Examination may also reveal various extra-articular manifestations of skin, heart, vessels, etc. In fact, HS is considered a forme fruste of inherited disorders of connective tissue. However, no mutation in any collagen tissue has been found in HS. There are also reports of autonomic dysfunction and decreased analgesia with topical lidocaine. Postural orthostatic tachycardia syndrome (POTS), has been associated with BHS. Patients with BHS and POTS appear to become symptomatic much earlier and have significantly higher incidence of syncope and migraine; increase in joint laxity pooling with secondary hyperadrenergic state or receptor dysregulation predisposing to autonomic dysregulation has been postulated to be the cause.

After other systemic rheumatic disorders are excluded and hypermobility is diagnosed, clinical management is multifaceted, involving the family physician, physiotherapist, and podiatrist. First and foremost is reassurance to the patient and family that hypermobility is relatively common and benign condition and does not have potentially disabling or life-threatening sequelae of other rheumatologic or connective tissue disorders. For acute symptoms, NSAIDs may be prescribed. For patients with chronic pain, a pain management programme based on cognitive behavioural techniques and delivered by a specially trained pain psychologist may reduce pain catastrophising, anxiety, and interference of pain with daily life. Patients should be advised to identify the activities that precipitate symptoms and modify their lifestyles accordingly. While on one hand, the precipitating and repetitive activities such as certain sports or hobbies should be avoided, by no
means inactivity should be encouraged. If avoidance of certain activities is not an acceptable option, then NSAIDs taken before the competitions/ sports may reduce symptoms, as in our 2nd patient. Moderate exercise is extremely beneficial, by maximising the muscle support around the hypermobile joint.

Exercise therapy should aim at improving muscle tone, joint stability and proprioception, under the supervision of a physiotherapist familiar with the disorder, tailored to suit the patients needs, as inappropriate use can exacerbate the symptoms. Isometric exercises should be advised that improve muscle tone without increasing the joint instability. Because the knee joint is commonly involved, quadriceps exercises can be especially helpful. Closed kinetic chain exercises improve proprioception and avoid injuries. In these exercises, the exercising limb, visualised as a chain of jointed segments, is the kinetic chain. In closed kinetic chain, the ends of the chain are fixed, while the intervening segments make small controlled movements. Along with exercise therapy, osteopathic manipulative treatment (OMT) is a useful adjunctive treatment modality. OMT helps induce articular release resulting in increased joint motion, and reduced pain as well as improved blood flow, lymphatic drainage, and proprioception. Therapy for autonomic disturbances and POTS is aimed at correcting the hypovolaemia and autonomic imbalance. Low dose beta-adrenergic antagonists and vasoconstrictors such as midodrine (an alpha – 1 agonist) may be useful. Patients with back pain benefit from back strengthening exercises and improved posture.

There remains much to be understood of this common and debilitating condition. Identification of causative genes will lead to greater understanding of the pathogenesis of the condition. Neurophysiological mechanisms leading to impaired proprioception, autonomic disturbance, local anaesthetic failure and pain amplification also warrant further investigation. At therapeutic level, randomised controlled trials of pharmacological, physical and psychology techniques are lacking for which we first need to identify good outcome measures. Most patients need sympathetic counselling and explanation. Reassurance that they are not suffering from serious diseases like RA and SLE will alleviate considerable suffering. Nearly 5% of normal individuals are positive for R factor; it often leads to misdiagnosis and unnecessary treatment. Awareness of high prevalence of the condition, a good history, clinical examination needs to be over and re-emphasised. Nevertheless, some patients may develop inflammatory disease later, so regular follow up is required.

From our 3 cases, we learnt that while some patients of BHS can be relatively asymptomatic throughout life and present later with degenerative osteoarthritis, some can present at a much younger age, with so much pain so as to affect a person’s day to day activity. Another point to note is that the hands with hyperextensible joints may mimic deformities of RA, but they are completely reducible deformities and there is no stiffness, swelling or tenderness. As physicians, we need to understand the impact that HS may have on many patients for whom each day is a struggle against pain and injury. Reassure the patients we must, but also accept the fact that we, the medical community have not really been able to provide effective cure for them.

References