BENIGN HYPERMOBILITY SYNDROME: A CASE REPORT

Gaurav Solanki

Jodhpur National University, Jodhpur-324003, (Rajasthan) India

Corresponding author*: drgauravsolanki@yahoo.com

Abstracts
A case of Benign Hypermobility Syndrome (BHS) is discussed. The case was a young female aged 24 with musculoskeletal symptoms since 5 years. The 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 doctors is warranted as some patients may be erroneously diagnosed as RA/SLE and may be put on DMARDs and steroids.

Keywords: Hypermobility, syndrome**,** rheumatic

1. 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) and hence the prefix benign to distinguish from the latter. There are many common features between EDS and Benign Hypermobility Syndrome (BHS).

The term hypermobility syndrome was coined by Kirke et al to denote the presence of rheumatic symptoms in otherwise healthy subjects in who have generalized joint laxity is the only observed abnormality. However; the condition had been first described by Hippocrtaes in Scythian warriors, who were warrior tribesmen from Central Asia who invaded India and established their kingdoms. Detailed history, careful clinical examination and the modified Beighton score aid in the accurate diagnosis[2].

2. Case Report:

A 30 year old female presented with multiple joint pains and low backache since 3 years. The pain occurred at any time of the day or night, sometimes disabling. There were 2-7 attacks of severe pain in a month, lasting for 2-3 hours, responding to analgesics and rest. Pain did not occur daily and some days were better than the others. Backache was not aggravated by bending forwards, coughing, sneezing or straining. She had been diagnosed as Spondyloarthropathy and had been on various NSAID's and DMARD's (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. 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 BHS was made. The DMARD's were omitted, she was counseled, advised supervised physiotherapy and she is doing well.

3. Discussion:

Diagnosis of BHS is one of exclusion. When patients present with pain in multiple joints with hypermobility in multiple joints; systemic rheumatic diseases should first be excluded. Women generally have more hypermobility than men; it is more common in childhood and decreases with age. Some patients also have localized hypermobility, which can make the

diagnosis difficult. There is also considerable variation between different ethnic groups. Modified Beighton score is widely accepted as a measure of joint hypermobility and assesses nine genetically determined sites. The testing involves 5 simple bedside clinical maneuvers and giving a score. A Beighton score of 4 or more is indicative of general hypermobility. Revised Brighton criteria, Bulbena criteria are some others. Careful history and clinical examination is essential as many patients with hypermobility have positive rheumatoid factor or antinuclear antibodies and are misdiagnosed as rheumatoid arthritis or SLE[3]

BHS can present withwith a variety of musculoskeletal complaints. The most common symptom is joint pain, which often develops after physical activities or sports during which the affected joint(s) is/are used repeatedly; like an over-use syndrome. Pain may be localized to one or several joints or it may be generalized and symmetric. Although pain most commonly involves the knee, any joint including those of the spine, can be affected. The pain is usually self-limited in duration, but can recur with activity. Less commonly, the patient may experience joint stiffness, myalgias, muscle cramps, and non-articular limb pain. In some cases, the onset of symptoms is preceded by growth spurt, and affected females often report premenstrual exacerbations. Since they are related to activity, the symptoms tend to occur late in the day. Morning stiffness is an uncommon finding. The pain is related to recurrent unrecognizable trauma. 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. Because joints stiffen with age, the natural history of BHS is typically one of improvement, with progressively lessening degrees of joint laxity and associated musculoskeletal symptoms. Many affected children outgrow their symptoms during adolescence or adulthood, and women may experience fewer symptoms following menopause. However, over many years, joint hypermobility can predispose to osteoarthritis, even prematurely, in the 4^{th} or 5^{th} decade followed by eventual chondrocalcinosis. BHS can also be associated with fibromyalgia syndrome[4].

Examination may also reveal extra-articular manifestations like hyperextensile skin leading to scarring and striae, mitral valve prolapse, weakness of the abdominal wall and pelvic floor leading to hernia and organ prolapse, decreased stiffness of the vasculature causing increased risk of varicose veins, eye involvement leading to myopia, etc. In a study of 87 patients, visiting Rheumatology clinic at Guys Hospital in London, it was reported that there was an increased prevalence of MVP, reduced upper to lower segment ratio, reduced skin thickness, spinal anomalies and history of fractures among subjects with hypermobility. These findings suggest that these patients may have a collagen defect that is manifested not only in joint laxity but also in abnormalities in heart valves, skin and bone. In fact, BHS is considered a former fruste of inherited disorders of connective tissue. After more serious disorders are excluded and hypermobility is diagnosed, clinical management is straightforward. 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, NSAID's may be prescribed. Regarding chronic management, 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. Moderate exercise is extremely beneficial, by maximizing the muscle support around the hyper mobile joint. Exercise therapy should aim at improving muscle tone, joint stability and proprioception. But it should be under the supervision of a physiotherapist familiar with the disorder, tailored to suit the patient's 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. In these exercises, the exercising limb, visualized 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.

Improved muscle tone and coordination and joint proprioception helps in avoiding injuries. Patients with back pain benefit from back strengthening exercises and improved posture. 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-emphasisesd. Nevertheless, some patients may develop inflammatory disease later, so regular follow up is required[5].

Conclusion:

From our case, 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 hyper extensile 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 BHS 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:

1. Everman DB, Robin NH. Hypermobility Syndrome. *Paediatrics in Review*. 1998; 19: 111 - 7.
2. Bulbena A, Duro JC, Porta M, Faus S, Vallescar R, Martin R. Clinical assessment of hypermobility of joints: assembling criteria. *J Rheumatol*. 1992; 19: 115 - 22.
3. Grahame R, Edwards JC, Pitcher D, Gabell A, Harvey W. A clinical and echocardiographic study of patients with the hypermobility syndrome. *Ann Rhem Dis*. 1981; 40: 541 - 6.
4. Zweers MC, Dean WB, van Kuppevelt TH, Bristo WJ, Schalkwijk J. Elastic fiber abnormalities in hypermobility-type of Ehlers-Danlos syndrome patients with tenascin-X mutations. *Clin Genet*. 2005; 67: 330 - 4.
5. Ferrell WR, Tennant N, Sturrock RD, Ashton L, Creeed G, Brydson G, Rafferty D. Amelioration of symptoms by enhancement of proprioception in patients with joint hypermobility syndrome. *Rthritis rheum*. 2004; 50: 33 - 8.