CASE STUDIES

Benign Joint Hypermobility Syndrome – not so 'Benign'

Yathish GC[1*], Canchi Balakrishnan[2], Mangat Gurmeet[2], Taral Parikh[1], Sagdeo Parikshit[1], Girish Kakade[1]

[1*]Resident in Rheumatology, P.D Hinduja Hospital and Research Centre, Mumbai

[2] Consultant Rheumatologist, P.D Hinduja Hospital and Research Centre, Mumbai

Abstract

Benign joint hypermobility syndrome (BJHS) was earlier considered as a mild or trivial entity, but nowadays it is reported as a multisystem hereditary connective tissue disorder with serious morbidities. In fact, the term 'Benign' has been removed, renaming the disease as 'Joint Hypermobility Syndrome' (JHS). In addition to the well-known musculoskeletal consequences of joint pain and instability, it can cause chronic widespread pain, gastrointestinal dysmotility, anxiety, phobic states and dysautonomia. We present here the case of a 53-year-old lady who presented with history of irritable bowel syndrome, elbow dislocation, repeated knee injuries, and instability resulting in severe secondary osteoarthritis (OA).

Case report

A 53-year-old lady presented to the OPD with a 16-year history of instability while walking, repeated knee injuries, and buckling and give away sensation at both knee joints. She had a previous episode of left elbow dislocation following a fall. She also experienced multiple joint and soft tissue pain while performing household activities. She denied back pain, swelling or morning stiffness in any of the joints. She was treated for irritable bowel syndrome by a gastroenterologist as she showed symptoms of repeated loose motions, abdominal pain and bloating sensation. She didn't have any co-morbidities or previous episodes of fractures.

Physical examination revealed hyperextensibility of joints as evidenced by ability to oppose the right thumb to the volar aspect of the forearm and passive dorsiflexion of the fifth metacarpophalangeal joint to ≥90°. She had bilateral knee valgus, genu recurvatum, ankle valgus, and pes planus deformities in lower limb. Both knees were unstable and valgus test was positive. In spite of severe knee deformities, range of motion was well preserved and painless. She had Marfanoid habitus in the form of soft elastic skin, high arched palate, thumb sign (Steinberg's sign), and an increased arm span to height ratio (Fig. 1 and 2). She didn't have any other systemic features suggestive of Marfan's syndrome.

Her hematological and biochemical investigations including inflammatory markers were within normal limits. Knee X-ray (AP view) showed bilateral valgus deformity, subluxation, loss of lateral tibiofemoral compartment joint space, osteophytes, subchondral sclerosis, and cysts suggestive of secondary OA (Fig 3).

Discussion

JHS is a common disorder originally defined as 'the occurrence of symptoms in otherwise healthy hypermobile individuals'.[1] It is now considered as a complex genetic disorder with manifestations that permeate well beyond the confinement of the musculoskeletal system. Hypermobility syndrome is diagnosed using the 1998 Brighton criteria, which includes relevant clinical symptoms, Beighton score, and other phenotypic features (Table 1 and 2).[2]

Musculoskeletal symptoms due to laxity of joints are the common presentation of JHS (Table 3). Skin may be visibly transparent, soft or silky, and palpably reduced in thickness with increased stretching. Scar formation is impaired, resulting in characteristically paper-thin scars. Visceral complications occur due to weakness affecting supporting structures such as the diaphragm, abdominal

Fig. 1: Soft, hyperelastic and visibly transparent skin

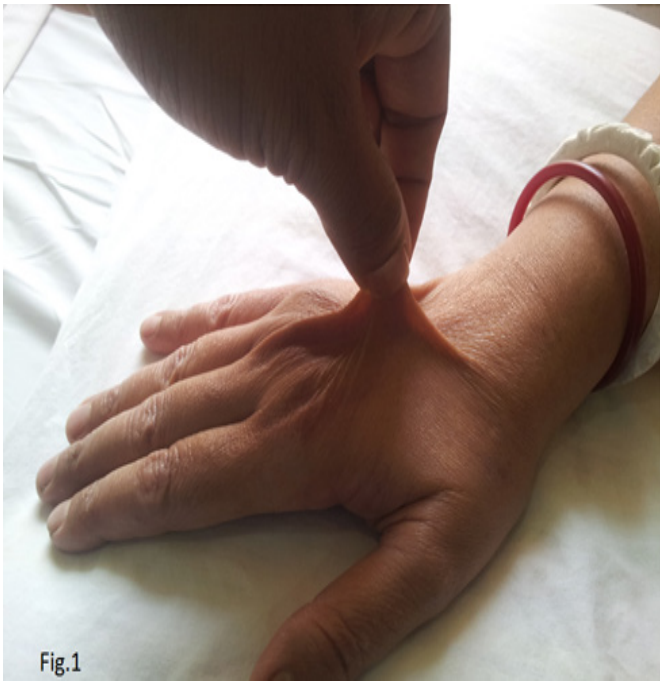


Fig. 2: Thumb sign (Steinberg sign)

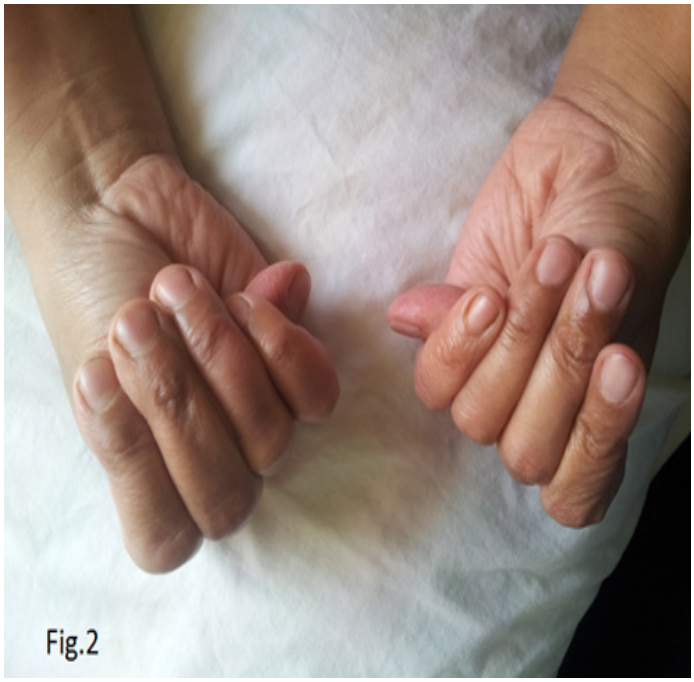
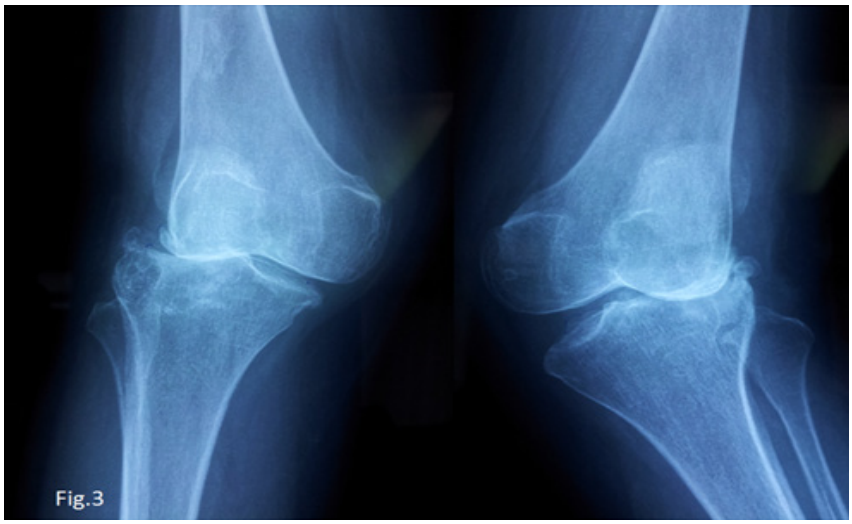


Fig 3: AP view of knee showing bilateral valgus deformity, subluxation, and severe lateral tibiofemoral compartment OA



wall, and pelvic floor. Complications include hiatal hernias, abdominal wall hernias, uterine or rectal prolapse, rectocele, and cystocele. Varicose veins are also a common occurrence in JHS. Mitral valve prolapse and spontaneous pneumothorax are rarer manifestations. Depression, anxiety, fibromyalgia and irritable bowel syndrome are frequent accompaniments of the chronic pain in JHS.[3, 4] Symptoms suggestive of an autonomic disturbance, including presyncope, syncope, and palpitations, are reported in around 60% of patients with JHS.[5, 6]

Treatment is often challenging in patients with severe symptoms. Agents like simple analgesics and anti-inflammatory drugs are the initial choices for both acute and chronic pain resulting from injuries. Majority of patients, however, report that these agents are of little benefit in

Table 1. Nine-point Beighton hypermobility score

Ability to		
1. Passively dorsiflex the fifth metacarpo phalangeal joint to ≥90°	1	1
2. Oppose the thumb to the volar aspect of the ipsilateral forearm	1	1
3. Hyperextend the elbow to ≥10°	1	1
4. Hyperextend the knee to ≥10°	1	1
5. Place the hands flat on the floor without bending the knees	1	
Total	9	

Table 2: 1998 Brighton revised diagnostic criteria for BJHS

Major criteria

1. A Beighton score of 4 of 9 or greater (either currently or historically)
2. Arthralgia for longer than 3 months in four or more joints

Minor criteria

1. A Beighton score of 1, 2, or 3 of 9 (0, 1, 2, or 3 if older than 50 years)
2. Arthralgia in one to three joints or back pain, spondylosis, or spondylolysis/ spondylolisthesis
3. Dislocation in more than one joint or in one joint on more than one occasion
4. Three or more soft tissue lesions (e.g., epicondylitis, tenosynovitis, bursitis)
5. Marfanoid habitus (tall, slim, span greater than height; upper segment–lower segment ratio <0.89, arachnodactyly)
6. Skin striae, hyperextensibility, thin skin, or abnormal scarring
7. Eye signs: drooping eyelids, myopia, or antimongoloid slant
8. Varicose veins, hernia, or uterine/rectal prolapsed

Requirement for diagnosis

Any one of the following:
Two major criteria
One major plus two minor criteria
Four minor criteria
Two minor criteria and unequivocally affected first degree relative

Source: The revised (Brighton 1998) criteria for the diagnosis of benign joint hypermobility syndrome (BJHS)

Table 3: Common musculoskeletal presentations

1. Soft tissue injury or overuse lesion affecting a ligament, tendon, muscle, enthesis or joint
2. Recurrent joint instability, subluxation or dislocation
3. Chronic non-inflammatory joint or spinal pain without apparent structural abnormality
4. Secondary osteoarthritic changes in peripheral joints and spondylotic changes in spinal joints

reducing chronic pain. Serotonergic and noradrenergic agents are used for treating fibromyalgia and chronic widespread pain. Physiotherapy, occupational therapy, and podiatry are the important components of treatment for this not so 'benign' condition.

Competing interests

The authors declare that they have no competing interests.

Disclosure

None

Citation

Yathish GC, Balakrishnan C, Gurmeet M, Parikh T, Parikshit S, Girish Kakade. Benign Joint Hypermobility Syndrome – not so 'Benign'. IJRCI. 2015;2(1):CS4.

Received: 5 April 2015, **Accepted:** 30 April 2015, **Published:** 21 May 2015

*Correspondence: Dr. Yathish GC, Department of Rheumatology, P.D

Hinduja Hospital and Research Centre, Mumbai, India
yathishgc45@gmail.com

References

1. Kirk JA, Ansell BM, Bywaters EG. The hypermobility syndrome. Musculoskeletal complaints associated with generalized joint hypermobility. Annals of the Rheumatic Diseases. 1967;26(5):419–25
2. Grahame R, Bird HA, Child A, et al. The revised (Brighton 1998) criteria for the diagnosis of benign joint hypermobility syndrome (BJHS). Journal of Rheumatology. 2000;27(7):1777-9.
3. Gratacòs M, Nadal M, Martín-Santos R, Pujana MA, Gago J, Peral B, et al. A polymorphic genomic duplication on human chromosome 15 is a susceptibility factor for panic and phobic disorders. Cell. 2001;106(3):367–79.
4. Farmer AD, Zarate-Lopez N, Mohammed SD, et al. Functional gastrointestinal symptoms and joint hypermobility: is connective tissue the missing link? [abstract 217]. Rheumatology. 2009;48.
5. Gazit Y, Nahir AM, Grahame R, Jacob G. Dysautonomia in the joint hypermobility syndrome. The American Journal of Medicine. 115(1):33–40.
6. Hakim AJ, Grahame R. Non-musculoskeletal symptoms in joint hypermobility syndrome: indirect evidence for autonomic dysfunction. Rheumatology. 2004;43: 1194-5.