Case Report

Benign Joint Hypermobility Syndrome

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Abstract: Hypermobility or joint laxity is not a disease. Benign joint hypermobility syndrome (BJHS) is a disease entity in which symptoms are produced by hypermobility. The diagnosis of BJHS relies on the exclusion of other differential diagnoses of polyarthralgia and connective tissue diseases such as primary osteoarthrosis, rheumatoid arthritis, Sjogren’s syndrome and systemic lupus erythematosus and spondyloarthropathy. A patient with BJHS is presented here to illustrate the typical clinical presentation if the condition. The diagnostic criteria and mechanisms of pain in BJHS are reviewed and discussed.

Keywords: Benign joint hypermobility syndrome, polyarthralgia, pain

Introduction

Collagen gives the body and its moving parts intrinsic strength. When it is weakened, the "moving parts", i.e. the joints, muscles, tendons and ligaments, become lax and fragile. Joint laxity or hypermobility predisposes individuals to injury. A minority of people with hypermobile joints develop symptoms which lead to the hypermobility syndrome. This syndrome probably has a genetic basis because it may run in family and affects more than one member. At one end of the spectrum of conditions that give rise to joint hypermobility are diseases with potentially serious complications such as the Marfan syndrome or Ehlers-Danlos syndrome (EDS) vascular type (formally named EDS type IV). At the other end of the spectrum is what is now called Benign joint hypermobility syndrome (BJHS) and Ehlers-Danlos hypermobile type (formerly EDS type III). BJHS may cause troublesome symptoms and persistent problems, but does not affect vital organ function or pose a serious threat to life. Polyarthralgia in middle age women is not uncommon and the diagnosis of BJHS should be considered after excluding other differential diagnosis. A patient with BJHS is hereby presented to illustrate the typical clinical presentation of this entity.

Case Presentation

A 45-year-old woman presented to our clinic with intermittent polyarthralgia for 5 years, with increasing frequency of attack in the recent 4 months. The polyarthralgia was symmetrical, with the involvement of the proximal phalangeal joints (IPJ), metacarpo-phalangeal joints (MCPJ), wrists, shoulders and knees and toes. She had subjective feeling of hotness in these joints during an attack. However, there was no significant early morning stiffness or symptoms suggestive of enthesitis and inflammatory back pain. There were no clinical features of systemic lupus erythematosus or Sjogren’s syndrome.

She enjoyed good health all along and was in fact a swimming coach of an international school. Her job required high physical demand and large volume high intensity swimming training. She swam 6 days a week, three to four thousand meters every day. Her father was also an elite swimmer and suffered similar problems since teenage.

Physical examination revealed a tall and slim woman with a normal ratio of arm span to body. There were no features of Marfan syndrome. The skin texture was normal without abnormal laxity. There were no particular signs for Ehlers-Danlos syndrome (EDS). Other systems were essentially
normal. She scored 5 out of 9 in the Beighton score for joint hypermobility (Appendix 1) and fulfilled 2 major criteria for the diagnosis of the BJHS according to the Beighton's criteria1 (Appendix 2).

Discussion

The frequency of BJHS varies with sex, age and ethnic background. Girls tend to have more mobility of the joints than boys of the same age. It remains unclear why some people with joint hypermobility develop joint discomfort, pain and swelling while others with equally hypermobile joints do not have symptoms. In patients with BJHS, there is often a family history of "loose-jointedness". There may occasionally be positive personal or family history of congenital hip dislocation, scoliosis, elbow, patellar or shoulder dislocation, or frequent ankle or wrist sprains. Hypermobility per se is a state, not a disease, but it may lead to generalised arthralgia or localised symptoms (frequent ankle sprains, knee effusions, dislocations of the shoulders and recurrent episodes of back pain). Pain can occur even after minor strains, especially in young women.2 Patients with BJHS, besides having hypermobile joints, often have decreased sense of joint position, predisposing them to minor and repeated injuries to the musculoskeletal system. Reduced sensory feedback may lead to biomechanically unsound limb positions and adoption of strange postures. This mechanism may lead to acceleration of joint degeneration and may account for the increased prevalence of degenerative arthritis in BJHS patients.3 Patients with BJHS are more likely to have osteoarthritis, increased nerve compression disorders,4 chondromalacia patellae, excessive anterior mandibular movement,5 mitral valve prolapses,6 uterine prolapses, and varicose veins.2 Larsson et al7 described that patients with BJHS who had a sedentary job had increased incidence of back pain. The mechanism may be due to combination of weak ligamentous support and weak core muscle strength. The stability of the lumbar spine relies on the supporting muscles and ligaments. This 'active' support comes from four mechanisms: (1) tension from thoracolumbar fascia; (2) intra-abdominal pressure; (3) the paraspinal muscles; and (4) the deep lumbar extensors. This fascia-muscular system is often disrupted in people with sedentary lifestyle and hypermobile joints. Weak core muscles jeopardise lumbar stability, which further promotes back injuries and symptoms of back pain.

There is a new criteria for the diagnosis of BJHS, i.e. the revised Beighton criteria, which has been validated in adults but not yet in adolescents / children below the age of 16 years. BJHS is present when 2 major criteria, or one major plus 2 minor, or 4 minor criteria are fulfilled and Marfan / Ehlers-Danlos syndromes (other than the EDS hypermobility type (formerly EDS type III) are excluded. The Beighton criteria have high sensitivity and specificity, (93% each). The Beighton score is merely a measure of generalised hypermobility or joint laxity. Having high Beighton score does not equate to the BJHS. Other symptoms and signs are necessary to qualify the diagnosis of BJHS.

Conclusion

Hypermobility syndrome, the new nomenclature BJHS, is not a very common condition. However, under-diagnosis is bound to occur when the index of suspicious is not high enough. Rheumatologists should be aware of this entity in their clinical practice. Treatment is simple and straightforward if diagnosis can be made early and accurately. Patient education is extremely important in the management of pain in this group of patients. Although there is still much to learn, the understanding of the condition is advancing and better treatment advice will be available.

References

BENIGN JOINT HYPERMOBILITY SYNDROME

Appendix 1

Beighton score

1. More than 10° hyperextension of the elbows = 1 score for each side
2. Passively touch the forearm with the thumb, while flexing the wrist = 1 score for each side
3. Passive extension of the fingers or a 90° or more extension of the fifth finger (Gorling's sign). (This is used as a "Screen Test") = 1 score for each side
4. Knees hyperextension greater than or equal to 10° (genu-recurvatum) = 1 score for each side
5. Touching the floor with the palms of the hands when reaching down without bending the knees = 1 score

Total score: 9

Hypermobility is present when score ≥4/9

Appendix 2

Revised diagnostic criteria for the benign joint hypermobility syndrome (BJHS)

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

Minor Criteria
1. A Beighton score of 1, 2 or 3/9 (0, 1, 2 or 3 if aged 50+)
2. Arthralgia (>3 months) in one to three joints or back pain (>3 months), spondylosis, spondylolysis / spondylolisthesis.
3. Dislocation / subluxation in more than one joint, or in one joint on more than one occasion.
4. Soft tissue rheumatism. >3 lesions (e.g. epicondylitis, tenosynovitis, bursitis).
5. Marfanoid habitus (tall, slim, span/height ratio >1.03, upper: lower segment ratio less than 0.89, arachnodactyly [positive Steinberg/wrist signs]).
6. Abnormal skin: striae, hyperextensibility, thin skin, papyraceous scarring.
7. Eye signs: drooping eyelids or myopia or antimongoloid slant.
8. Varicose veins or hernia or uterine/rectal prolapse.
   i. The BJHS is diagnosed in the presence 2 major criteria, or 1 major and 2 minor criteria, or 4 minor criteria.
   ii. BJHS is excluded by presence of Marfan or Ehlers-Danlos syndromes (other than the EDS Hypermobility type [formerly EDS III] as defined by the Ghent 1996 (8) and the Villefranche 1998 (9) criteria respectively). Criteria Major 1 and Minor 1 are mutually exclusive as are Major 2 and Minor 2.