

Benign joint hypermobility syndrome

Łagodny zespół nadmiernej ruchomości stawów

Iwona Słowińska¹, Lidia Rutkowska-Sak²

¹Department and Policlinic of Rheumorthopedy, Institute of Rheumatology, Warsaw

²Department of Developmental Age Rheumatology, Institute of Rheumatology, Warsaw

Key words: benign joint hypermobility syndrome, arthritis, joint pain.

Słowa kluczowe: łagodny zespół nadmiernej ruchomości stawów, zapalenie stawów, bóle stawów.

Summary

Benign joint hypermobility syndrome (BJHS), commonly known as loose ligament syndrome, is a non-inflammatory rheumatic condition. It is characterised by a greater than normal range of motion of the joints of the limbs and spine. The prevalence of the syndrome in preschool-age children is estimated to be between 2% and 30%, depending on ethnic background (with higher prevalence in Asian and African populations), occurring most often in families with a history of the condition and more frequently in girls. This paper presents a case report of a 12-year-old girl. A broad differential diagnostic approach to recurrent joint inflammation with joint effusion and pain made it possible to establish a diagnosis of benign joint hypermobility syndrome. The child met the Brighton criteria; her Beighton score was 7 out of 9. Patient education aimed at eliminating abnormal joint movement and an appropriate rehabilitation programme play key roles in the treatment of BJHS.

Streszczenie

Łagodny zespół nadmiernej ruchomości stawów (BJHS), zwany potocznie zespołem nadmiernej wiotkości więzadeł, jest zaliczany do niezapalnych chorób reumatycznych. Charakteryzuje się przekraczającą prawidłowy zakres amplitudą ruchu w stawach kończyn i kręgosłupa. W wieku przedszkolnym częstość występowania tego zespołu szacuje się na 2–30%, zależnie od przynależności etnicznej (częściej jest stwierdzany wśród Azjatów i Afrykańczyków), najczęściej w rodzinach obciążonych tą chorobą i częściej u dziewczynek. W pracy opisano przypadek 12-letniej dziewczynki, u której szeroka diagnostyka różnicowa nawracających objawów zapalenia stawów kolanowych z wysiękami i bólami tych stawów pozwoliła na rozpoznanie BJHS. Dziewczynka spełniała kryteria BRIGHTON, w tym 7 z 9 punktów w skali Beighton. W leczeniu tego zespołu kluczową rolę odgrywa edukacja chorego mająca na celu wyeliminowanie nieprawidłowych ruchów oraz właściwie prowadzona rehabilitacja.

Introduction

Benign joint hypermobility syndrome (BJHS), commonly known as loose ligament syndrome, is a non-inflammatory rheumatic condition. It is characterised by a greater than normal range of motion of the joints of the limbs and spine. The prevalence of the syndrome in preschool-age children is estimated to be between 2% and 30%, depending on ethnic background (with higher prevalence in Asian and African populations), occurring most often in families with a history of the condition and more frequently in girls [1, 2].

The pathogenesis of the disorder remains unclear. It is believed the disease has an underlying genetic basis, associated with the X chromosome, and develops as

a result of defects in genes encoding connective tissue matrix proteins such as type I, III and V collagen, elastin and fibrillin as well as the extracellular matrix glycoprotein tenascin. It is assumed that this leads to a loss of resistance to stretching of the tissues surrounding joints [3].

The first mention of BJHS was probably made in antiquity by Hippocrates and concerned Scythian horsemen [4]. Clearly, we have no way to know whether these horsemen, or many famous musicians, acrobats and dancers, had genetic defects that affected their connective tissue and made it possible for them to achieve a virtuoso level in their fields. According to some authors, characteristic morphological skeletal features of the three sisters portrayed by Rubens in “The Three

Address for correspondence

Prof. Lidia Rutkowska-Sak, MD, PhD, Department of Developmental Age Rheumatology, Institute of Rheumatology, Spartańska 1, 02-637 Warsaw, e-mail: lidia.rutkowska@ir.ids.pl

Submitted: 21.10.2014

Graces" (among them, Rubens' wife) support the familial occurrence of the syndrome [5].

BJHS was initially considered to be a form of Ehlers-Danlos syndrome. Years later it was recognised to be a separate entity. The syndrome was first described in a paper by Kirk et al. [6].

The first criteria used for diagnosing BJHS were developed by Carter and Wilkinson in 1964:

- passive apposition of the thumb to the forearm,
- passive hyperextension of the metacarpophalangeal and carpal joints so that the fingers lie parallel to the dorsal aspect of the forearm,
- hyperextension of the elbow joints,
- hyperextension of the knee joints,
- excessive flexion of the ankle joint and eversion of the foot.

In 1973, Beighton et al. developed a scoring system for assessing patients suspected of having BJHS:

- ability to place palms flat on the floor while performing forward flexion of the trunk (1 point),
- passive hyperextension of the fifth finger ≥ 90 degrees (1 point for each hand),
- passive apposition of the thumb to the volar aspect of the forearm (1 point for each side),
- hyperextension of the elbow joint > 10 degrees (1 point for each side),
- hyperextension of the knee joint > 10 degrees (1 point for each side).

A score of at least 4 points was considered sufficient to establish a diagnosis.

In 1998, the British Society for Rheumatology Special Interest Group on Heritable Disorders of Connective Tissue developed a new set of criteria (the 'Brighton criteria') utilising the Beighton scale. These are currently the most popular criteria used for diagnosing BJHS, including in children [7] (Table I).

Joint hypermobility syndrome is diagnosed in the presence of at least two "major" criteria, or one "major" criterion plus two "minor" criteria, or four "minor" criteria. Two

"minor" criteria are considered sufficient to establish a diagnosis of BJHS in cases where the patient has a first-degree relative unequivocally affected by the disease.

It is worth mentioning that some authors emphasise the value of a set of BJHS diagnostic criteria developed by Bulbena, also known as the Hospital del Mar criteria [8] (Table II).

As children age, excessively mobile joints tend to become less flexible; however, the damage and discomfort caused by the condition can continue and worsen over time. These are usually the result of excessive strain and mechanical injuries that are typical for childhood.

Case study

The case concerns a 12-year-old girl with young, healthy parents. She was born in good general condition of a full-term, unremarkable first pregnancy through spontaneous, normal delivery and received an Apgar score of 10 points.

The family history was negative for chronic diseases. Her psychomotor development was normal. She began to walk at 13 months. The patient had frequent upper respiratory tract infections during early childhood. Since starting school she has had four episodes of knee joint swelling and effusion. These episodes were linked to frequent falls, recreational activities in the form of intensive dance classes several times a week and tennis, which she had played for approximately 5 years. Twice, due to articular effusion, the patient required decompression by aspiration, and twice the condition resolved spontaneously after 2 weeks of bed rest. Basic laboratory tests performed at that time did not reveal any abnormalities. Ultrasound imaging examination of the knee joints revealed the presence of significant effusion but was otherwise unremarkable. Anteroposterior and lateral X-ray examination of both knee joints performed following 3 joint aspirations showed only patellar lateralization. From investigation of the patient's past history it was established that she had sprained her ankles on many occasions and often wore

Table I. Criteria used for diagnosing BJHS using Beighton scale [7]

MAJOR CRITERIA
1. A Beighton score of $\geq 4/9$ (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 in individuals aged 50+)
2. Arthralgia ≥ 3 months in 1 to 3 joints, or back pain ≥ 3 months, spondylosis, spondylolysis/spondylolisthesis
3. A history of dislocation or subluxation in more than one joint or in one joint on more than one occasion
4. Soft tissue rheumatism lesions ≥ 3 (such as epicondylitis, tenosynovitis, bursitis)
5. Marfanoid habitus, arachnodactyly (positive Steinberg/wrist signs)
6. Skin abnormalities – striae, hyperextensibility, thin skin, papyraceous scarring)
7. Eye signs: drooping eyelids, myopia, antimongoloid slant
8. Varicose veins, hernia, rectal or vaginal/uterine prolapse

Table II. Criteria Bulbena used for diagnosing BJHS [8]

Examination of the upper extremity
1. Passive apposition of the thumb to the volar aspect of the forearm up to a distance of 21 mm
2. Passive dorsiflexion of the fifth finger to an angle of 90 degrees with the palm placed flat on a table
3. Passive hyperextension of the elbow joint to an angle of 10 degrees
4. External rotation of the shoulder to an angle of 85 degrees from the sagittal axis with the arm touching the torso and the forearm flexed at an angle of 90 degrees
Lower extremities – examination in a supine position
5. Passive abduction of the hip joint to an angle of 85 degrees
6. Patella may be moved from side to side with ease using one hand with the proximal end of the tibia stabilised using the other hand
7. Excessive range of passive ankle dorsiflexion and eversion of the foot
8. Dorsiflexion of the first metatarsophalangeal joint up to an angle of 90 degrees
Lower extremities – examination in a prone position
9. Knee flexion allows the heel to make contact with the buttock
Extra-articular manifestations
10. Ecchymoses appear after minimal, often unnoticed trauma

elastic bandages to help stabilize these joints. Despite this, according to her parents, she had always been very nimble and had been regarded as a model of physical fitness. In addition, the patient complained of significant pain in her feet and, on occasion, in the joints of her upper limbs, particularly in the evenings or after physical education classes, dancing, or tennis. She also reported night-time awakenings due to joint pain. The patient was referred to the Paediatric Rheumatology Outpatient Clinic of the Institute of Rheumatology in Warsaw with suspicion of a locomotor system disorder. Her physical examination was significant for severe pes planus, right-lateral scoliosis, patellar lateralization and ballotement, ganglions in the vicinity of the left ankle joint and right wrist as well as generalized joint hypermobility. The child's Beighton score was 7 out of the possible 9 points. The blood tests did not reveal any elevation in acute inflammation marker levels or immune response parameters. Calcium and phosphorus metabolism was normal. Infectious and reactive arthritis were excluded. MRI imaging of the knee joints detected discreet signs of bursitis and the presence of effusion in the left knee joint as well as bilateral patellar lateralization. The patient was referred to the Orthopaedic Outpatient Clinic with suspicion of BJHS.

The consultant orthopaedic rheumatologist confirmed the above-mentioned findings of the patient's physical examination. Scintigraphic imaging of bones excluded foci of infection and cancer.

Diagnostic arthrocentesis was performed, and the decision to refrain from intra-articular administration of anti-inflammatory medicine was made. Synovial fluid culture was sterile. The diagnosis of benign joint hypermobility syndrome was confirmed. The patient

met all of the currently accepted criteria for this syndrome.

Discussion

A child may be diagnosed with BJHS having a Beighton score of 7 out of 9 points. Recent works demonstrate the usefulness of the Beighton score for assessment of joint hypermobility in children of school age [9]. Our patient met that condition. She did not present any clinical manifestations characteristic for the Marfan syndrome or Ehlers-Danlos syndrome.

Many authors believe that a thorough investigation of the medical history and physical examination are sufficient for diagnosis [10].

In 2003, Hakim et al. stated that BJHS may be suspected based solely on a patient's history – specifically, on a set of 5 questions – particularly in the case of older patients [11]:

1. Can you now (or could you ever) bend forward and place your hands flat on the floor without bending your knees?
2. Can you now (or could you ever) bend your thumb to touch your forearm?
3. As a child, did you amaze people you knew by contorting your body into strange positions or could you do the splits?
4. As a child, or teenager, did your kneecap dislocate on more than one occasion?
5. Do you feel "double jointed"?

All these attributes, confirming an excessive range of movement in joints, were observed by us in our patient.

However, the process of establishing BJHS in children must always involve a broad differential diagnosis and

exclusion of the large number of disorders characterised by musculoskeletal system pain. In the case of our patient, whose disease was complicated by knee joint inflammation, the differential diagnosis was essential.

Benign joint hypermobility syndrome can lead to the development of pes planus, subluxations of the small joints of the hands and the patellae, bone fractures, and ligament rupture. Secondary tenosynovitis and arthritis with, on occasion, concomitant joint effusion (as in the case of our patient) and other signs of inflammation are recurrent in nature and may evolve into a chronic condition. Even young people can develop discopathy, degenerative disease, and osteoporosis [12, 13].

Joint hypermobility is present in approximately 30–40% of children below the age of 10 years. This fact is worth keeping in mind to avoid misdiagnosing BJHS [14]. Joint hypermobility may also occur as a result of many years of training and exercises. Correct diagnosis of BJHS is very important for the implementation of appropriate treatment. Treatment involves use of kinesiotherapy to build muscle strength as well as modern manual techniques and physiotherapy to improve proprioception, which is often impaired. Patient education plays an invaluable role in eliminating abnormal joint movement. Patients may also receive psychotherapy and, on occasion, pharmacotherapy – particularly patients with severe pain [15–17].

The authors declare no conflict of interests.

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