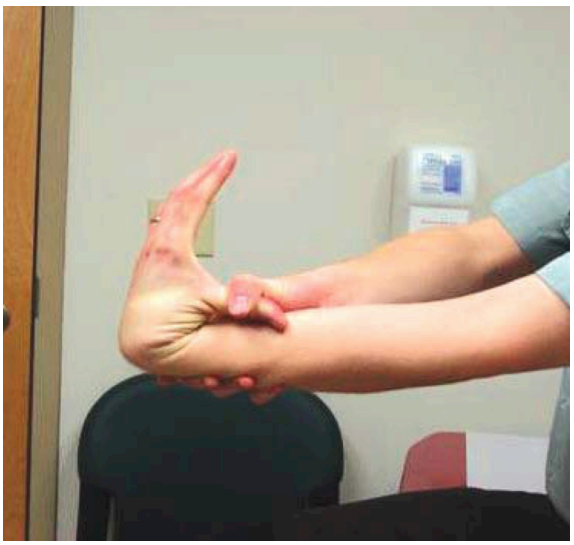
A 30 year old project manager, who is new to your general practice, presents with right anterior knee pain after slipping and landing on his knee three months ago. Imaging shows no abnormality, but he describes a long history of recurrent shoulder subluxation, and many soft tissue problems and joint pains, often after similarly trivial trauma, and he states that imaging and blood tests “for arthritis” have always been normal. He is hypermobile joint.



? DIAGNOSIS

HYPERLAXITY SYNDROME

Benign joint hypermobility syndrome (BJHS) is a connective tissue disorder with hypermobility in which musculoskeletal symptoms occur in the absence of systemic rheumatologic disease.

When patients with this syndrome are first seen by a physician, their chief complaint is joint pain, so BJHS can be easily overlooked and not considered in the differential diagnosis.

Symptoms

The signs and symptoms of BJHS are variable. Most commonly, the initial complaint in a hypermobile patient is joint pain, which may affect one or multiple joints and may be generalized or symmetric.

Typically, children have self-limited pain in multiple joints; however, pain can last for a prolonged time and may become constant in adulthood. Pain may involve any joint but most commonly involves the knee and ankle, presumably because they are weight–bearing joints. Physical activity or repetitive use of the affected joint often exacerbates the pain. Consequently, pain usually occurs later in the day and morning stiffness is uncommon. Less common symptoms are joint stiffness, myalgia, muscle cramps, and non-articular limb pain. Patients with BJHS often say that they are "double-jointed" or that they can contort their bodies into strange shapes

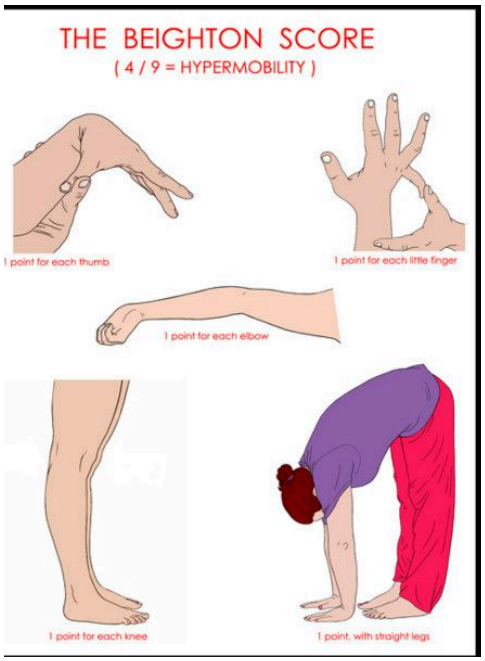
Questions to the patient

1. Can you now (or could you ever) 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 amuse your friends by contorting your body into strange shapes or could you do the splits?
4. As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
5. Do you consider yourself double-jointed?

Signs

1. **Acute or Traumatic**
   * Sprains: — recurrent ankle sprains
   * Meniscus tears
   * Acute or recurrent dislocations or subluxations of the: — shoulder, patella, temporomandibular joint
   * Traumatic arthritis
   * Bruising, " Fractures
2. **Chronic or Nontraumatic**
   * Soft tissue rheumatism: tendinitis, epicondylitis, rotator cuff syndrome, bursitis
   * Chondromalacia, Back pain, Scoliosis
   * Fibromyalgia
   * carpal tunnel syndrome, tarsal tunnel syndrome, thoracic outlet syndrome
   * Raynaud syndrome
   * Flat feet and sequelae
   * Unspecified arthralgia or effusion of affected joint(s)

Brighton's Criteria 9 points



Major Criteria
Beighton score of >4
Arthralgia >3 months in 4 or more joints
!
Minor Criteria
Beighton score of 1, 2, or 3
Arthralgia > 3-month in one to three joints or back pain
Dislocation or subluxation >one joint

Three or more soft tissue lesions (eg, epicondylitis, tenosynovitis, bursitis

Marfanoid habitus (tall, slim, span greater than height ("1.03 ratio), upper segment less than lower segment (<0.89 ratio), arachnodactyly)
Skin striae, hyperextensibility, thin skin, or abnormal scarring
Ocular signs: drooping eyelids, myopia, antimongoloid slant

" Varicose veins, hernia, or uterine or rectal prolapse
" Mitral valve prolapse

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 firstdegree relative in family history

Look for: Signs of a typical
connective tissue disorder may be present, including scoliosis, pes planus, genu valgum, lordosis, patellar subluxation or dislocation, marfanoid habitus, varicose veins, rectal or uterine prolapse, and thin skin

How common is it?
Occurs in 10-20% of populations of Western countries, and higher still in those in Indian, Chinese, and Middle Eastern groups. Hyperlaxity may diminish in severity after skeletal maturity and with advancing age, and it is more prevalent in young females3-5 and in Asian, African, and Middle Eastern individuals6-

It is important to distinguish between joint hypermobility and joint hypermobility syndrome. People who are hypermobile without symptoms are merely people with hypermobility. Patients with multidirectional shoulder instability may have specific patterns of collagen abnormality related to genetic inheritance.

Why is it missed?
In a recent survey suggested missed diagnosis, 52% of 251 patients waited over 10 years from the onset of symptoms to get a correct diagnosis.
Doctors may be unaware of the prevalence of the condition, its effect on quality of life, or its multisystemic nature and may not routinely look for hypermobility in the clinical examination

Why does this matter?
If joint hypermobility syndrome is missed, the following problems may arise:
Inappropriate and potentially harmful labeling or treatments may be applied on the basis of an erroneous diagnosis such as rheumatoid arthritis, hypochondriasis, or

somatization.
Over zealous physical manipulation may cause avoidable damage Inflicting rupture on ligaments, joint capsules, muscles, or tendons, or *(c)* precipitating pathological fractures in fragile bone.

How is it diagnosed?
Diagnosis is entirely clinical as currently no biological or imaging markers are available. The musculoskeletal symptoms mainly derive from a vulnerability to injury resulting from fragile collagenous tissues (tendon, ligament, muscle, bone, cartilage, and skin).
In patients with arthralgia or post-injury musculoskeletal pain, screening blood tests and/or appropriate imaging are needed to exclude conditions such as inflammatory arthritis and fractures.
Diagnosis requires the application of the 1998 Brighton criteria into which the Brighton score has been incorporated.

How is it managed?
The key players are the family doctor and a suitably trained physiotherapist.

Doctor's role
To establish an accurate diagnosis of joint hypermobility syndrome while being alert to the possibility of one of the rarer and more serious heritable disorders of connective tissue, such as Marfan's syndrome, or other forms of Ehlers-Danlos syndrome, such as vascular, or classical. A positive family history of sudden early death from aortic aneurysmal dissection and/or rupture should suggest the possibility of Marfan's syndrome, and a history of major spontaneous arterial rupture or uterine rupture in childbirth should raise suspicions of the vascular type of Ehlers-Danlos syndrome.

Physiotherapist's role:
This involves:
-Core and joint stabilising and proprioception enhancing exercises
-General fitness training to reverse the tendency for the body to lose condition
-The use of mobilising techniques to restore natural hypermobility to joints

D/D

Connective Tissue Disease	Inheritance	Genetic Mutation	Key Diagnostic Features
Hypermobility-type Ehlers-Danlos syndrome	Autosomal dominant	Unknown collagen gene defect	Skin hyperextensibility, tissue fragility
Marfan syndrome	Autosomal dominant	Fibrillin-1 gene	Marfanoid body habitus, skin hyperextensibility, ectopia lentis, aortic dilatation
Osteogenesis imperfecta	Autosomal dominant	Type-I collagen genes	Bone fragility, blue sclerae, short stature
Benign joint hypermobility syndrome	Autosomal dominant	Unknown collagen gene defect	Musculoskeletal symptoms in persons with hypermobility who are otherwise healthy

Acquired joint laxity syndrome
In patients with acquired shoulder joint laxity, repeated minor injuries (so-called microtrauma), or repetitive use during training and competition, stretch the normal capsuloligamentous restraints. Symptoms are commonly unilateral in the dominant shoulder, and other joints are often normal. The acronym AMBRI was subsequently used by Matsen to describe instability that is atraumatic multidirectional, and

bilateral, responds to rehabilitation, and occasionally requires an inferior capsular shift

Attempt to answer the following questions:
1. Are the patient's symptoms attributable to instability?
2. In which directions does the patient have symptomatic instability?
3. What is the cause of any instability?
a. How severe is the hyperlaxity associated with instability?
b. Is there evidence of shoulder soft-tissue or osseous structural abnormalities?
c. Is there evidence of voluntary control, muscle patterning, or social factors?
4. Are there other contributory causes for the symptoms, such as rotator cuff impingement?

Treatment
1. Assurance: Nonprogressive, noninflmatory
2. Life style modification; avoid repeated activity
3. Alter patients exercise regimen, joint protection and proper body mechanics
4. Judicious use of NSAID
5. Open and closed kinetic exercise in moderation: to improve strength and coordination
6. Appropriate foot wear
7. Few sessions of osteopathic treatment: Osteopathic manipulative treatment helps induce articular release resulting in increased joint motion, and reduced pain as well as improved blood flow, lymphatic drainage, and proprioception.
8. Prolotherapy could contribute to the treatment of hypermobility disorders also by preventing the development of precocious osteoarthritis. It has long been known that individuals with joint hypermobility syndrome and EDS suffers with premature osteoarthritis in various joints and the amount of degeneration correlates with the extent of the individuals hypermobility. The combination of extreme hypermobility and repeated injury is presumed to be what leads to the early osteoarthritis.
9. Most patients with hyperlaxity have a reduction in instability symptoms after nonoperative treatment, including physical therapy, activity modification, and additional psychological support when necessary.
10. Operative treatment provides reproducibly good results for patients with hyperlaxity who do not respond to a prolonged program of nonoperative measures.

Multidirectional instability of the shoulder: Open inferior capsular shift remains the gold standard of operative treatment, although arthroscopic capsular shift and plication procedures are now producing comparable results.
Thermal capsulorrhaphy is associated with unacceptably high failure rates and postoperative complications and cannot be recommended as a treatment.

Prognosis
The syndrome's nonprogressive nature; decreased joint laxity symptoms that occur with age. However, patients need to be aware of the potential sequelae: include acute ligament and soft tissue injury, overuse injury, joint instability, possible increase in fractures and scoliosis, and increased frequency of uterine and rectal prolapse.

In addition, these patients may be predisposed to osteoarthritis from years of

excessive joint motion. Also, an association between BJHS and panic disorder has been shown. Despite these sequelae, patients should remain as active as possible.

References

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