Benign joint hypermobility syndrome (BJHS) is a connective tissue disorder with hypermobility in which musculoskeletal symptoms occur in the absence of systemic rheumatologic disease. Although BJHS has been well recognized in the rheumatology and orthopedic literature, it has not been discussed in the family medicine literature. Because most patients with musculoskeletal complaints are first seen by family physicians, it behooves primary care physicians to be familiar with recognizing and diagnosing BJHS. 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. Use of the Brighton criteria facilitates the diagnosis of BJHS. Treatment modalities include patient education, activity modification, stretching and strengthening exercises for the affected joint, and osteopathic manipulative treatment.

Benign joint hypermobility syndrome (BJHS) is the occurrence of musculoskeletal symptoms in hypermobile individuals in the absence of systemic rheumatologic disease. This syndrome is thought to be an inherited connective tissue disorder. The primary clinical manifestations of BJHS are hypermobility and pain in multiple joints. This syndrome is different from other disorders that cause local joint hypermobility and generalized joint laxity, such as Marfan syndrome and Ehlers–Danlos syndrome (EDS).

The Brighton criteria are used to diagnose BJHS, and laboratory tests are used to distinguish BJHS from other systemic diseases. Other criteria have been proposed for diagnosis, including Bulbena’s criteria; however, the criteria defined by Brighton are the ones most frequently cited in the literature. Generalized joint laxity is commonly seen in healthy individuals who do not have complaints. Hypermobility that is not associated with systemic disease occurs in 4% to 13% of the population. Hypermobility diminishes as one ages, and it also appears to be related to sex and race. In general, women have greater joint laxity than men, and up to 5% of healthy women have symptomatic joint hypermobility compared with 0.6% of men. People of African, Asian, and Middle Eastern descent also have increased joint laxity.

Among studies examining the prevalence of generalized hypermobility in patients referred to rheumatologists, one study found that hypermobility occurred in 66% of school children with arthralgia of unknown etiology. Another study showed a similar prevalence of hypermobility in children; however, there was no association between hypermobility and the development of arthralgia. These data suggest that generalized hypermobility exists without joint pain and it does not necessarily lead to arthralgia. Furthermore, patients with hypermobility often lead normal lives and do not have BJHS or another connective tissue disorder.

Hypermobility may occur in several different connective tissue disorders including Marfan syndrome, EDS, and osteogenesis imperfecta. It may also be found in chromosomal and genetic disorders such as Down syndrome and in metabolic disorders such as homocystinuria and hyperlysinemia. Recurrent dislocation of the shoulder and patella as well as other orthopedic abnormalities are associated with joint laxity. Juvenile rheumatoid arthritis may also be associated with hypermobility, but many times, systemic symptoms are involved. Patients with BJHS have generalized hypermobility as well as chronic joint pain and other neuromusculoskeletal signs related to a defect in collagen.

Benign joint hypermobility syndrome has a strong genetic component with an autosomal dominant pattern. First–degree relatives with the disorder can be identified in as many as 50% of cases. The syndrome appears to be due to an abnormality in collagen or the ratio of collagen subtypes. Mutations in the fibrillin gene have also been identified in families with BJHS.

So, why do patients with BJHS present mainly with joint pain? It is thought that excessive joint laxity leads to wear and tear on joint surfaces and strains or fatigues the soft tissue...
Mobility. The onset of symptoms can occur at any age, and five simple questions (Table 1) can aid in recognizing hypermobility. 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. Primary care physicians can use the five simple questions (Figure 1) to aid in recognizing hypermobility. The onset of symptoms can occur at any age, and many patients have been referred to specialists in orthopedics, rheumatology, or physiatry. 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 nonarticular limb pain. Patients with BJHS often say that they are "double-jointed" or that they can contort their bodies into strange shapes (ie, voluntary subluxation) or do the splits. Such an admission, however, is not necessary for including BJHS in the differential diagnosis. Patients with BJHS may also have a history of shoulder or patellar dislocation.

Patients with BJHS may have a family history of "double-jointedness" or recurrent dislocations. Other presentations include easy bruising, ligament or tendon rupture, congenital hip dysplasia, and temporomandibular joint dysfunction.

Findings of the physical examination vary based on the joint affected. Pain in response to manipulation of the joint is common. Mild effusions are not common but may be present. Clinically significant tenderness along with redness, swelling, fever, or warmth suggests inflammation and is not present in patients with BJHS.

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 (Figure 2). The association of BJHS with mitral valve prolapse (MVP) is a subject of controversy. Early studies showed an association between MVP and BJHS, but later studies have questioned this association because of stricter echocardiographic criteria for MVP. Primary care physicians should refer patients with hypermobility in whom cardiac findings suggest MVP to a cardiologist for further evaluation to rule out more serious cardiac abnormalities and connective tissue disorders.

Diagnosis

Determining the Beighton score (Figure 3) is essential for making the diagnosis. The first step is to calculate the Beighton score, which is a measure of generalized joint laxity. Physicians calculate this score by doing five simple maneuvers (Figure 4) that can be completed in 45 to 60 seconds. A Beighton score of 4 or more points is considered indicative of generalized joint laxity. Most patients with BJHS have symmetric joint laxity. The Beighton criteria were developed to establish diagnostic criteria for BJHS. Using these criteria helps physicians to distinguish BJHS from other connective tissue disorders.

Diagnosis of BJHS is one of exclusion. For patients with painful or swollen joints, it is important to rule out inflammatory, infectious, and autoimmune causes. Workup may

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**Patient Interview Questions to Ask**

- Can you now (or could you ever) place your hands flat on the floor without bending your knees?
- Can you now (or could you ever) bend your thumb to touch your forearm?
- As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
- As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
- Do you consider yourself double-jointed?

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**Figure 1. Questions physicians should ask patients to detect benign joint hypermobility syndrome. (Reproduced with permission from Hakim AJ, Cherkas LF, Grahame R, Spector TD, MacGregor AJ. The genetic epidemiology of joint hypermobility: A population study of female twins. Arthritis Rheum. 2004;50:2640-2644.)**

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Benign joint hypermobility syndrome needs to be distinguished from other disorders that share many common features, such as Marfan syndrome, EDS, and osteogenesis imperfecta. Generalized hypermobility is a common feature in all these hereditary connective tissue disorders and many features overlap, but often distinguishing features are present that enable differentiating these disorders.

**Ehlers–Danlos Syndrome**

Ehlers–Danlos syndrome comprises a group of connective tissue disorders that have gross joint laxity and may have purple papyraceous scars, skin hyperelasticity, and skin fragility that leads to easy bruising. Similarly to BJHS, EDS is inherited in an autosomal dominant fashion and is due to a defect in collagen. Of the many different types of EDS that exist, the most common include a complete blood cell count, erythrocyte sedimentation rate, rheumatoid factor, antinuclear antibody test, serum complement (C3, C4, CH50) levels, and serum immunoglobulin (IgG, IgM, IgA) levels. Any of these test results that are not within the normal reference range suggest an alternate diagnosis. Sometimes, patients with BJHS have an effusion, but the joint aspirate shows a noninflammatory pattern from meniscal and cartilage irritation.

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**Brighton Criteria**

- **Major Criteria**
  - Beighton score of ≥4 (Figure 4)
  - Arthralgia for longer than 3 months in 4 or more joints

- **Minor Criteria**
  - Beighton score of 1, 2, or 3 (Figure 4)
  - Arthralgia (>3-month duration) in one to three joints or back pain (>3-month duration) or spondylosis, spondylolysis/spondylolisthesis
  - Dislocation or subluxation in more than one joint, or in one joint on more than one occasion
  - 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 first-degree relative in family history

*Figure 3. Brighton criteria, based on determination of the Beighton score (Figure 4), is used to make the diagnosis of benign joint hypermobility syndrome. (Note: Readers should not be confused by the similarity of these two names. “Beighton” is the correct spelling of the name of the score, and “Brighton,” the correct spelling of the name of the criteria.)*
Figure 4. Maneuvers used to calculate the Beighton score. Four of the five maneuvers are each done on the right and left sides, and 1 point is awarded for each positive result. The photographs show how to perform each maneuver but do not necessarily represent a positive result. The highest possible score is 9. A score of 4 or higher meets the Brighton major criteria for hypermobility. (Note: Readers should not be confused by the similarity of these two names. “Beighton” is the correct spelling of the name of the score, and “Brighton,” the correct spelling of the name of the criteria.)
common are types I, II, and III. Benign joint hypermobility syndrome is thought to be a mild variation of EDS and most closely resembles EDS type III (hypermobility type), which consists of joint pain, marked hypermobility, mild extra-articular involvement, and mild skin changes without scarring. Researchers have suggested that BJHS lies on a continuum with EDS and may be its mildest form because of their overlapping features.

Marfan Syndrome
Patients with Marfan syndrome often have a family history of the syndrome and tend to have cardiac and ocular features. Marfan syndrome is an autosomal dominant disorder in patients with a tall, thin body habitus (marfanoid habitus), generalized joint hypermobility, elongated fingers (arachnodactyly), myopia, and lens dislocation. Osteogenesis imperfecta is also characterized by a defect in collagen. Patients have excessive joint laxity, thin blue sclera, and bone fragility leading to multiple fractures and bony deformities.

Juvenile Rheumatoid Arthritis
Juvenile rheumatoid arthritis may be considered in children with hypermobility and joint pain, but its diagnosis requires the onset of arthritis before the age of 16 years, inflammation of one or more joints, and the exclusion of other rheumatic disorders.

Management
The first step in managing BJHS is to emphasize to patients that this syndrome is a nonprogressive, noninflammatory connective tissue disorder. Effective treatment may be accomplished with lifestyle modification, altering the patient’s exercise regimen, joint protection, and proper body mechanics.

For acute symptoms, nonsteroidal anti-inflammatory drugs (NSAIDs) or acetaminophen have often been used for pain control. Joint complaints in these patients are not thought to be due to inflammation, so the use of NSAIDs for anything other than pain is disputed. For moderate or severe pain, rest and abstinence from aggravating activities may improve symptoms. Physical therapy and joint protection may also help.

Long-term management of BJHS typically focuses on modification of activities, especially if they induce symptoms. Excessive joint movement is associated with the development of symptoms in patients with BJHS. Often, vigorous and repetitive activities have been targeted as aggravating factors. Overtraining, poor pacing, too many performances or athletic competitions, and focusing on joint flexibility rather than stability may all increase joint pain and the risk of injury. If avoidance of these activities is not an acceptable option for patients, physicians may try other approaches. NSAIDs taken before competition often may reduce symptoms. Also, starting a strengthening program to provide muscular stability and stabilization to the joint may be beneficial. Stretching techniques that are targeted to isolate tight muscles without stressing the surrounding joints may reduce symptoms by improving balance and control.

Strength training should consist of a combination of both open kinetic (distal extremity moves freely) and closed kinetic chain (distal extremity meets resistance) exercises. Closed kinetic chain exercises often simulate functional demands of an extremity, while open kinetic chain activities are better for more targeted strength training.

Focusing on improving the proprioception of a joint may improve symptoms (eg, using a wobble board). Sometimes, supportive splints along with appropriate footwear protect the joint, and supportive joint taping improves joint proprioception. Focused exercises to improve muscle strength, balance, and coordination may help improve joint stability and proprioception. Improvement of proprioception may reduce strain to the ligaments surrounding the joint and avoid further injury.

Along with exercise therapy, OMT is a useful adjunctive treatment modality for BJHS. Thrust treatment techniques applying high velocity/low amplitude forces are the most widely used, but because of the increased tissue fragility seen in BJHS and weak supporting structures of the joint, gentler techniques like facilitated positional release and counterstrain are good alternatives. 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. It is thought that OMT can lead to hypermobility; however, not enough research has been conducted to confirm this hypothesis, and it is currently recommended that OMT be limited to no more than three times per week. To help patients return to their activities while decreasing their symptoms and joint stress, osteopathic physicians should consider all these factors.

Prognosis
The prognosis for patients with BJHS is generally good owing to the syndrome’s nonprogressive nature and decreased joint laxity and symptoms that occur with age. However, patients need to be aware of the potential sequelae that have an increased frequency associated with BJHS. These 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. Altering their exercise regimen may avoid chronic joint pain. Good training strategies include slow, disciplined training, correct biomechanics, and effective proprioception.

Given the many sequelae that may develop and the poten-
tial impact of BJHS on quality of life, some experts are questioning why BJHS is called a “benign” disorder; instead, they refer to the disorder as “joint hypermobility syndrome.” However, most rheumatologists still refer to it as BJHS. In the future, more rheumatologists may use the changed name—joint hypermobility syndrome—because of the disorder’s potential effects on quality of life.

The potential complications of BJHS underscore the importance of making an early diagnosis and educating the patient.

Comment
Osteopathic physicians predominantly practice primary care medicine, and most patients with musculoskeletal complaints first see a primary care physician and are then referred to a rheumatologist. Hypermobility is a common cause of unexplained joint pain, yet it is often misdiagnosed in primary care. According to one source, primary care providers recognize generalized hypermobility in less than 10% of patients with generalized hypermobility who were referred to rheumatologists. Therefore, physicians need to be aware of the clinical presentation of BJHS to enhance their diagnostic acumen.

Benign joint hypermobility syndrome is the presence of musculoskeletal complaints in hypermobile individuals in the absence of systemic rheumatologic disease. It is a connective tissue disorder with a defect in collagen. The Brighton criteria are used to make the diagnosis. Education, activity modification, and exercise therapy to improve muscular stability and proprioception at specific joints are essential to symptom management. And, OMT is a useful adjunctive treatment modality to help reduce pain and improve proprioception.

Finally, primary care physicians are the best resource to educate patients with BJHS about their illness, potential complications, and prognosis. Furthermore, a prompt diagnosis improves pain control and decreases disruptions in these patients’ physical activities, school, work, and quality of life. Thus, by being the first medical contact for most of their patients with BJHS, physicians in primary care have the opportunity to diagnose and address BJHS and reduce the incidence of potential long-term sequelae, chronic pain, and traumatic injuries.

References