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## Hypermobility can lead to musculoskeletal deformities and be associated with other serious conditions

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When a child presents in clinic with a clubfoot, hip dysplasia or scoliosis, the condition may be just as it appears — a singular musculoskeletal deformity. Yet, in each of these cases, an underlying cause of joint hypermobility, or hyperlaxity, can be easily overlooked. Similarly, in young children who are not yet walking by 18 months of age, joint hypermobility may be mistaken for developmental delay, hypotonia or even cerebral palsy.

When a joint hypermobility disorder goes undiagnosed, the result can be painful early-onset arthritis in adulthood, orthopaedic deformities or, in rare cases, life-threatening complications such as a ruptured bowel or uterus.

Recognizing generalized joint hypermobility is most effectively achieved using criteria described by Carter-Wilkinson,<sup>1</sup> with modifications by Beighton et al.<sup>2</sup> These criteria require a minimum of three types of hyperextension for a diagnosis of hypermobility (see table 1).

### Table 1 Exam criteria for joint hypermobility

Three of the following must be present for a positive diagnosis:

1. Passive opposition of the thumb to the flexor aspect of the forearm (see fig. 1).
2. Passive hyperextension of the fingers until parallel with the extensor aspect of the forearm.
3. Hyperextension of the elbows beyond 10 degrees.
4. Hyperextension of the knees beyond 10 degrees.
5. Forward trunk flexion maintaining straight knees with the palms of the hands on the floor.

Various studies have shown that the incidence of hypermobility varies with respect to age, gender and race. If there are no other associated symptoms, joint hypermobility is usually considered benign and is often familial. The condition is much more common in infants and young children than in adults, and is known to occur in a high percentage of Asian children. (Randomly ascertained data on school-age children found that 50 percent of Chinese-Asian subjects had joint hypermobility<sup>3</sup> as opposed to 7 percent of English-Caucasians.<sup>1</sup>) It is also more common in females than males.

Joint hypermobility becomes worrisome, however, when accompanied by musculoskeletal problems or other symptoms such as skin hyperelasticity, which results from defective collagen molecules in the skin.

Disorders associated with non-benign types of joint hypermobility include Ehlers-Danlos syndrome, osteogenesis imperfecta, Marfan syndrome and Larsen syndrome. Of these, Ehlers-Danlos (EDS) is the most common, (two times as common as OI), and consists of nine known genetically distinct disorders.

## The patient with EDS

By definition, EDS is a heterogeneous group of inherited connective tissue disorders characterized by:

- Joint hypermobility
- Hyperextensible, doughy skin
- Atrophic scars
- Connective tissue fragility
- Easy bruising



**fig. 1** Passive opposition of the thumb to the flexor aspect of the forearm

The diagnosis for most types of EDS is based on clinical exam and patient history, although a few types can now be diagnosed by means of skin biopsy and other lab tests. Despite the fact that EDS is an inherited disorder, family history cannot be relied upon for accurate diagnosis. A significant number of EDS patients are the first in their family to have the syndrome. Because diagnosis of EDS and other hyperlaxity syndromes can be complex, referral to a pediatric orthopaedist is prudent whenever a child appears to have joint hypermobility, or associated musculoskeletal problems (see table 2).

To understand the importance of diagnosing EDS early in a child's life, one need only consider the complications that can arise within the various subtypes of the disease. As mentioned earlier, there are nine subtypes within EDS, each with distinct clinical features (see table 3).

While there are currently no medical therapies for EDS, identification of the subtype the patient has is crucial to avoiding complications and to ensuring appropriate medical management. For example, patients with types I, IV and VII experience a high rate of hip dislocation. Fifty percent of those with EDS type III will eventually develop scoliosis, a condition that is also a hallmark of type VI. Most patients with EDS probably have types I, II or III.

## Table 2

Conditions associated with joint hypermobility and EDS:

Young children	<ul style="list-style-type: none"> <li>• hip dysplasia</li> <li>• scoliosis</li> <li>• delayed walking</li> <li>• clubfeet</li> </ul>
Older children and adolescents	<ul style="list-style-type: none"> <li>• patellar or shoulder dislocations</li> <li>• scoliosis</li> <li>• frequent joint injuries associated with hypermobility</li> <li>• easy bruising</li> <li>• excessive scarring after minor injuries</li> <li>• significantly flat feet</li> </ul>

Although rare, type IV EDS is the most worrisome. The skin elasticity is normal, but abnormal collagen compromises the integrity of the internal organs and leaves them vulnerable to rupture. Sudden, spontaneous rupture of the bowel, arteries or pregnant uterus can lead to death. Twenty percent of females with this type of EDS die during pregnancy. Thirty percent of type IV patients have a relative who died of complications from the same disorder.

Typically, a pediatric orthopaedist is the first to identify EDS, and oversees the patient's orthopaedic concerns. However, effective medical management of patients with more severe forms of EDS requires a multidisciplinary approach. No one specialist can appropriately address all of the patient's complex needs. In addition to the child's primary care provider, other key members of the care team may include a geneticist, a hematologist, a cardiologist, an ophthalmologist and a dentist.

**Table 3<sup>(4)</sup>**  
**Clinical features of EDS subtypes I-VIII and X.**  
 (Type IX is no longer considered EDS.)

<b>EDS Subtypes</b>	<b>Clinical Features</b>
I – Gravis	Hyperextensible skin, wide scars, bruising, striking joint hypermobility, joint subluxation and dislocation, joint effusions, joint pain, flat feet (in 52 %), problems with wound healing
II – Mitis	Similar to type I, less severe
III – Familial hypermobility	Soft skin, no scarring, hypermobility, scoliosis, patellar subluxation/dislocation
IV – Arterial	Translucent skin, visible veins, arterial/bowel/uterine ruptures, normal skin extensibility, cardiac manifestations, problems with wound healing and excessive bleeding
V – x-linked	Same as type II
VI – Ocular	Hyperextensible skin, small joint hypermobility only, scoliosis, ocular fragility
VII – A&B	Hip dislocation, joint hypermobility, soft skin with normal scarring
C	Very fragile skin, joint hypermobility
VIII – Periodontal	Periodontitis, skin similar to type II
IX – Fibronectin defect	Similar to type II

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