EHLERS-DANLOS SYNDROME HYPERMOBILITY TYPE

Ehlers-Danlos syndrome hypermobility type is a connective tissue disorder that mostly affects the bones and joints. People with this condition have loose joints and frequently have long-term joint pain.

Ehlers-Danlos syndrome hypermobility type is a connective tissue disorder that predominantly affects the skeletal system.

What other names do people use for Ehlers-Danlos syndrome hypermobility type?

Ehlers-Danlos syndrome hypermobility type is also known as Ehlers-Danlos syndrome Type III, Ehlers-Danlos Type 3, and EDS hypermobility type.

How prevalent is Ehlers-Danlos syndrome hypermobility type?

It is estimated that 1 in 5,000–20,000 people have Ehlers-Danlos syndrome hypermobility type.

What are the characteristics of Ehlers-Danlos syndrome hypermobility type?

Characteristics range from mild, such as loose joints, to severe, such as functional bowel disorders and incisional hernias. The joints and skin are most commonly affected. Joints may have a wide range of movement (hypermobility), be unstable, and tend to move out of place (dislocate) frequently. The shoulder, knee, and jaw are some of the joints that dislocate most often. The skin of a person with Ehlers-Danlos syndrome hypermobility type is often soft or velvety and may be easily stretched and overly flexible (hyperextensible), although this is variable.

What causes Ehlers-Danlos syndrome hypermobility type?

The exact cause of Ehlers-Danlos syndrome hypermobility type is not known; however, a small percentage of the time it is believed to be caused by a change (mutation) in the TNXB gene (tenascin-X).

Most people diagnosed with Ehlers-Danlos syndrome hypermobility type inherit it from a parent who also has the condition. It is an autosomal dominant condition; therefore, only one parent needs to have a change in the gene that causes the disorder in order to pass it on. With each pregnancy, there is a 50 percent chance of passing it to the child.

How is Ehlers-Danlos syndrome hypermobility type diagnosed?

Diagnosis is based entirely on a clinical evaluation and family history. The clinical evaluation is done by a medical professional who looks for the major and minor criteria required to make the diagnosis. Genetic testing and prenatal genetic testing are available on a limited basis for TNXB mutations, but only if the disease-causing mutation has been identified in a family member.

The major diagnostic criteria are:

- Joint hypermobility
    - Ability to bend your pinky finger backwards towards your wrist (passive dorsiflexion) greater than 90°
    - Ability to push your thumb to your wrist (passive apposition)
    - Hyperextension of each elbow greater than 10°
    - Hyperextension of each knee greater than 10°
    - Ability to place the palms on the floor with the knees fully extended
- Soft skin with normal or only slightly increased extensibility
- Absence of fragility or other significant skin or soft tissue abnormalities
- Fleshy, heaped-up growths or patches of skin that do not resemble the area that surrounds it (molluscoid pseudotumors), associated with scars over pressure points such as the elbows and knees
- Surgical complications, such as incisional hernia or wound dehiscence (sutures tearing through tissues and failing to hold)

The minor criteria include:

- Positive family history of Ehlers-Danlos syndrome hypermobility type (or family history of joint laxity), without significant skin or soft tissue weakness, in a pattern consistent with autosomal dominant inheritance
- Recurrent joint dislocations (joint moves out of place) or subluxations (restricted movement of the joint)
- Chronic joint, limb, and/or back pain
- Easy bruising

- Functional bowel disorders (functional gastritis, irritable bowel syndrome)
- A feeling of faintness after long periods of standing, being in a warm environment, immediately after exercise, after a large meal, or in stressful situations which is caused by low blood pressure (neurally mediated hypotension) or an excessive heart rate upon standing up (postural orthostatic tachycardia).
- High, narrow palate
- Dental crowding

How is Ehlers-Danlos syndrome hypermobility type managed?

Treatment is based on a person's specific set of symptoms. Options include:

- Physical therapy/rehabilitation
- Assistive devices, such as braces to improve joint stability; wheelchair or scooter to take stress off lower-extremity joints; and suitable mattress to improve sleep quality
- Pain medication to help relieve joint pain
- Surgical procedures should be considered with caution

What is the life expectancy of someone with Ehlers-Danlos syndrome hypermobility type?

Ehlers-Danlos syndrome hypermobility type does not affect life expectancy.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.