Ehlers-Danlos syndrome

Symptoms & causes Diagnosis & treatment Doctors & departments Care at Mayo Clinic

Overview

Ehlers-Danlos syndrome is a group of inherited disorders that affect your connective tissues — primarily your skin, joints and blood vessel walls. Connective tissue is a complex mixture of proteins and other substances that provide strength and elasticity to the underlying structures in your body.

People who have Ehlers-Danlos syndrome usually have overly flexible joints and stretchy, fragile skin. This can become a problem if you have a wound that requires stitches, because the skin often isn't strong enough to hold them.

A more severe form of the disorder, called Ehlers-Danlos syndrome, vascular type, can cause the walls of your blood vessels, intestines or uterus to rupture. Because Ehlers-Danlos syndrome, vascular type, can have serious potential complications in pregnancy, you may want to talk to a genetic counselor before starting a family.

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Symptoms

Classic Ehlers-Danlos syndrome

Signs and symptoms of the most common form of Ehlers-Danlos syndrome include:

- **Overly flexible joints.** Because the connective tissue that holds joints together is looser, your joints can move far past the normal range of motion. Joint pain and dislocations are common.
- **Stretchy skin.** Weakened connective tissue allows your skin to stretch much more than usual. You may be able to pull a pinch of skin up away from your flesh, but it will snap right back into place when you let go. Your skin might also feel exceptionally soft and velvety.
- **Fragile skin.** Damaged skin often doesn't heal well. For example, the stitches used to close a wound often will tear out and leave a gaping scar. These scars may look thin and crinkly.

Symptom severity can vary from person to person. Some people with Ehlers-Danlos syndrome will have overly flexible joints, but few or none of the skin symptoms.

Ehlers-Danlos syndrome, vascular type

People who have Ehlers-Danlos syndrome, vascular type, often share distinctive facial features of a thin nose, thin upper lip, small earlobes and prominent eyes. They also have thin, translucent skin that bruises very easily. In fair-skinned people, the underlying blood vessels are very visible through the skin.

Ehlers-Danlos syndrome, vascular type, can weaken your heart's largest artery (aorta), as well as the arteries to other regions of your body. A rupture of any of these larger blood vessels can be fatal. The vascular type can also weaken the walls of the uterus or large intestines — which also may rupture.

Causes

Different types of Ehlers-Danlos syndrome are associated with a variety of genetic causes, some of which are inherited and passed on from parent to child. If you have the most common varieties of Ehlers-Danlos syndrome, there's a 50 percent chance that you'll pass on the gene to each of your children.

Complications

Complications depend on the types of signs and symptoms you have. For example, overly flexible joints can result in joint dislocations and early-onset arthritis. Fragile skin may develop prominent scarring.

People who have Ehlers-Danlos syndrome, vascular type, are at risk of often fatal ruptures of major blood vessels. Some organs, such as the uterus and intestines, also may rupture. Pregnancy can increase these risks.

Prevention

If you have a personal or family history of Ehlers-Danlos syndrome and you're thinking about starting a family, you may benefit from talking to a genetic counselor — a health care professional trained to assess the risk of inherited disorders. Genetic counseling can help you understand the inheritance pattern of the type of Ehlers-Danlos syndrome that affects you and the risks it poses for your children.

By Mayo Clinic Staff

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