

MEDICAL BREAKTHROUGHS

RESEARCH SUMMARY

TOPIC: EHLERS-DANLOS SYNDROME: SUPER FLEXIBLE, OR LIFE-THREATENING?
REPORT: **MB #4948**

BACKGROUND: Ehlers-Danlos syndrome is a group of inherited disorders that affect connective tissues primarily 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 the body. People who have Ehlers-Danlos syndrome usually have overly flexible joints and stretchy, fragile skin. This can become a problem if someone has a wound that requires stitches, because the skin often isn't strong enough to hold them. A more severe form of the disorder, called vascular Ehlers-Danlos syndrome, can cause the walls of blood vessels, intestines or uterus to rupture. Because vascular Ehlers-Danlos syndrome can have serious potential complications in pregnancy, a patient may want to talk to a genetic counselor before starting a family.

(Source: <https://www.mayoclinic.org/diseases-conditions/ehlers-danlos-syndrome/symptoms-causes/syc-20362125>)

DIAGNOSIS: Doctors may use a series of tests to diagnose EDS (except for hEDS) or rule out other similar conditions. These tests include genetic tests, skin biopsy, and echocardiogram. An echocardiogram uses sound waves to create moving images of the heart. This will show the doctor if there are any abnormalities present. A blood sample is taken from the arm and tested for mutations in certain genes. A skin biopsy is used to check for signs of abnormalities in collagen production. This involves removing a small sample of skin and checking it under a microscope. A DNA test can also confirm if a defective gene is present in an embryo. This form of testing is done when a woman's eggs are fertilized outside of her body (in vitro fertilization).

(Source: <https://www.healthline.com/health/ehlers-danlos-syndrome#diagnosis>)

CURRENT TREATMENT: Treatment for Ehlers-Danlos syndrome aims to prevent dangerous complications. It can also help protect the joints, skin, and other tissues from injuries. An individual's treatment depends on many factors, including the type of the disorder and symptoms. To protect the skin, doctors recommend using sunscreen and mild soaps. Taking extra Vitamin C can help reduce bruising. Physical therapy (exercises to strengthen the muscles supporting the joints) can help prevent joint injuries. Braces also help stabilize joints. Because blood vessels are fragile, doctors will monitor people with Ehlers-Danlos syndrome and may use medication to help keep blood pressure low and stable. Dislocated joints and other joint injuries are common among people with Ehlers-Danlos syndrome.

(Source: <https://my.clevelandclinic.org/health/diseases/17813-ehlers-danlos-syndrome>)

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If this story or any other Ivanhoe story has impacted your life or prompted you or someone you know to seek or change treatments, please let us know by contacting Marjorie Bekaert Thomas at mthomas@ivanhoe.com