Ehlers-Danlos Syndrome

What is Ehlers-Danlos syndrome?

Ehlers-Danlos syndrome (EDS) is a group of rare genetic disorders. These disorders affect the way collagen is made or used in the body. Collagen is a type of protein. It adds strength and structure to organs and connective tissues. Problems with collagen can affect many parts of the body, including the skin, joints, blood vessels, bones, cartilage, and tendons.

There are 13 types of EDS. They are classified based on their symptoms, the gene affected, and the pattern of inheritance. The most common type of EDS, hypermobile EDS, is regarded as a genetic condition but the genetic cause is unknown as the responsible gene(s) has not been identified. All types are present at birth. But a person with EDS may not be diagnosed until later in life if they have mild symptoms. In some types, symptoms can be severe and even life-threatening.

What causes Ehlers-Danlos syndrome?

A change, or mutation, in a certain gene can lead to EDS. This change is often passed down in families. Or it may happen during development. Many different genes can affect the way collagen is made or used in the body. The different types of EDS are partly based on which gene is affected.

Who is at risk for Ehlers-Danlos syndrome?

People who have a family history of EDS are more at risk for it. The type of EDS in the family does not change, such that if a specific type of EDS is identified, then others in the family would be at risk for the same type.

What are the symptoms of Ehlers-Danlos syndrome?

The symptoms of EDS vary from person to person. They also depend on the type of EDS a person has. Some types can cause mild symptoms, while others can be life-threatening.

EDS mainly affects the skin and joints. Symptoms of the most common types of EDS include:

- Soft, thin, stretchy skin
- Abnormal scarring of the skin
- Joints that move too much and that can easily dislocate
- Joint and muscle pain
- Fatigue
- Muscle weakness
- Skin growths on the knees or elbows
- Hard bumps under the skin
- Easy bruising
- Wounds that take a long time to heal

In more severe types of EDS, a person may have:

- Heart and valve problems, such as mitral valve prolapse
- Tear or rupture of a blood vessel or an organ

- Hernia
- Eye problems, such as glaucoma or retinal detachment
- Dental problems like gum disease and tooth loss
- Spine problems, such as scoliosis
- Delay in growth
- Short stature

How is Ehlers-Danlos syndrome diagnosed?

Your healthcare provider will ask about your symptoms and past health. They will also talk with you about your family's health history. Because EDS is a genetic disorder, it can be passed down in families.

Each type of EDS has its own set of diagnostic criteria. Features that may be seen across all types include changes in your skin and joints. So your healthcare provider will do a physical exam. They will look closely at your skin and joints. They may test the stretchiness of your skin. You may also need some tests, such as:

- **Genetic blood test.** Some types of EDS have been linked to a specific gene. Testing your blood may be able to identify the type of EDS you have. The hypermobile type of EDS does not have a known genetic cause and would not be diagnosed by a genetic test.
- **Imaging tests.** Your healthcare provider may advise X-rays or a CT or MRI scan. These can find problems in parts of the body such as the bones, heart valves, or blood vessels.

How is Ehlers-Danlos syndrome treated?

There is no cure for EDS. Care is focused on easing symptoms, such as joint pain. Your healthcare providers will also talk with you about how you can prevent future health problems. These include joint dislocation and poor wound healing.

It's best to have a team of healthcare providers help you manage EDS. Your primary healthcare provider may refer you to a medical geneticist. This provider specializes in treating genetic disorders. You may also need to see other providers to manage certain aspects of the disease. For instance, if you have heart problems, you may need to see a cardiologist. You may also want to speak with a genetic counselor if you are planning to have a child.

To help you manage EDS, your healthcare providers may advise:

- **Skin protection.** Sunscreen can help protect thin skin from the sun. Bandages or pads on elbows, knees, and other trauma prone areas may help prevent slow-healing wounds.
- **Physical and occupational therapy.** These can improve joint strength and function.
- **Regular exercise.** Being active everyday can lessen joint and muscle pain. But don't do activities that can damage or dislocate your joints. These include contact sports, weight lifting, and running. Activities such as walking and swimming are best.
- **Mobility devices, such as a wheelchair or braces.** These can help if you have trouble walking.
- **Pain medicine.** To ease chronic pain, your provider may advise over-the-counter or prescription medicines like acetaminophen. You may not be able to take aspirin or nonsteroidal anti-inflammatory drugs (NSAIDs). These may increase bruising and bleeding.
- **Other pain management options.** Talk with your provider about other ways to ease pain. You may find meditation, massage, or heat therapy helpful.

For people who have severe symptoms, surgery or other treatments may be needed. This can include surgery for a dislocated joint or a ruptured blood vessel.

What are possible complications of Ehlers-Danlos syndrome?

EDS can cause complications, such as:

- Joint dislocation
- Early onset of arthritis
- Tear or rupture of an organ
- Heart valve or blood vessel problems
- Physical disability due to muscle and joint problems
- Poor wound healing
- Mental health problems like depression

Living with Ehlers-Danlos syndrome

EDS is a lifelong disease. But you can take steps to prevent problems linked to EDS. Make sure you see your healthcare team for regular checkups and tests. Keep in mind that poor wound healing can delay recovery after dental and medical procedures.

Living with a chronic disorder like EDS can be hard to handle. A support group can help you cope with the mental and physical problems of EDS. Talk with your healthcare team about support groups near you. Or reach out to the Ehlers-Danlos Society at www.ehlers-danlos.com.

When to call your healthcare provider

Call your healthcare provider right away if you have any of these:

- Fever of 100.4°F (38°C) or higher, or as directed by your provider
- Symptoms that don't get better or get worse
- New symptoms

When to call

Call right away if you have any of these symptoms:

- Sudden pain in the chest, belly, or back
- Sudden bleeding
- Trouble breathing
- Dizziness, lightheadedness, or fainting
- Vision loss
- Severe headache

Key points about Ehlers-Danlos syndrome

- EDS is a group of rare genetic disorders. These disorders affect the way collagen is made or used in the body.
- A change, or mutation, in a certain gene can lead to EDS.
- EDS mainly affects the skin and joints. Many types cause soft, thin skin and loose joints.

- To diagnose EDS, your healthcare provider will ask about your symptoms and family history. Some types of EDS can also be identified with a genetic blood test (except the hypermobile type of EDS).
- There is no cure for EDS. Care is focused on easing symptoms and preventing future health problems, such as joint dislocation.

Next steps

Tips to help you get the most from a visit to your healthcare provider:

- Know the reason for your visit and what you want to happen.
- Before your visit, write down questions you want answered.
- Bring someone with you to help you ask questions and remember what your provider tells you.
- At the visit, write down the name of a new diagnosis and any new medicines, treatments, or tests. Also write down any new instructions your provider gives you.
- Know why a new medicine or treatment is prescribed and how it will help you. Also know what the side effects are.
- Ask if your condition can be treated in other ways.
- Know why a test or procedure is recommended and what the results could mean.
- Know what to expect if you do not take the medicine or have the test or procedure.
- If you have a follow-up appointment, write down the date, time, and purpose for that visit.
- Know how you can contact your provider if you have questions or if an emergency occurs.