Genetic Testing for Ehlers-Danlos Syndrome

Ehlers-Danlos Syndrome (EDS) is a group of connective tissue disorders that can occur in families. In this handout, you will learn about EDS and available genetic testing options, based on your type of EDS.

WHAT IS EHLCERS-DANLOS SYNDROME?

Ehlers-Danlos Syndrome (EDS) refers to conditions that affect the connective tissues in your body made mostly of collagen. Collagen is found in bones, muscles, tendons (which form our joints) blood vessels and the gut. Collagen also keeps skin strong and flexible.

WHAT ARE POSSIBLE SYMPTOMS OF EDS?

- Recurrent joint pain (joint pain that comes back)
- Joint hypermobility (loose or very flexible joints)
- Orthostatic hypotension (feeling lightheaded or dizzy when changing positions quickly)
- Easy bruising
- Fatigue
- Soft skin
- Stretch marks (striae)
- High and narrow roof of the mouth (palate)
- Irritable Bowel Syndrome (IBS) (a disorder that causes gas, belly pain, diarrhea and constipation)

HOW IS EDS PASSED DOWN IN FAMILIES?

With most types of EDS there is a 50% (1 out of 2) chance that each child of a parent with EDS will also have EDS. Not everyone with EDS is affected the same way, even in the same family. This is because each person experiences unique symptoms of EDS. The type of EDS does not change within the same family.

WHAT ARE THE DIFFERENT TYPES OF EDS?

There are many types of EDS, but here are some of the more common ones:

- **EDS Hypermobility (Type III).** This is the most common type of EDS. People with this type have joint pain and loose or very flexible joints.
- **EDS Classic (Type I and Type II).** This is less common than Type III. People with these types of EDS have all the symptoms of Type III, plus fragile stretchy skin, wide scars and may have heart problems.
- **EDS Vascular Type (Type IV).** This type of EDS is rare. People with Type IV have fragile skin, a higher risk of organ rupture (hurting their organs) and torn blood vessels.

HOW DO WE DIAGNOSE EDS?

Doctors diagnose EDS by your medical history and a physical exam. In other words, your geneticist (genetics doctor) will do a physical exam and check your symptoms, including your joints and skin and how flexible they are. The geneticist will also go over your history and also look at your family’s medical history.

WHAT ARE GENETIC TESTING OPTIONS FOR EDS?

Whether genetic testing is available for EDS depends on the type of EDS your geneticist thinks you might have. If testing is available, it might not be covered by health insurance. Talk with your geneticist or call your health insurance to ask if genetic testing is covered:

- **If you have EDS Type III,** genetic testing is not done because doctors base this diagnosis on a physical exam. We don’t know which gene (piece of your DNA) causes most instances of this type of EDS at this time.
- **If you have EDS Type I or Type II,** genetic testing is usually available through a blood test. But, the genetic test only finds about 50% (1 out of every 2) of cases. Your geneticist can also diagnose this type of EDS without testing.
- **If we think you might have EDS Type IV,** we will talk with you about genetic testing for the COL3A1 gene that causes this type of EDS.

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