



Heritable Disorders of Connective Tissue

Points To Remember About Heritable Disorders of Connective Tissue

- There are more than 200 heritable disorders that can affect the tissues between the cells of your body that give tissues form and strength.
- All of these diseases are related to problems in genes that are responsible for building connective tissues.
- Some heritable disorders of connective tissue change the look and growth of skin, bones, joints, heart, blood vessels, lungs, eyes, and ears. Others change how these tissues work.
- Treatments can include regular check-ups with your doctor as well as medicines or nutritional supplements.
- A nutritious diet, exercise, and healthy lifestyle habits can also help.

What are heritable disorders of connective tissue?

There are more than 200 heritable disorders of connective tissue that can affect the tissues between the cells of your body that give tissues form and strength. All of these diseases are directly related to problems in genes that are responsible for building connective tissues. The disorders are called “heritable,” because they are passed on from parent to child.

Some heritable disorders of connective tissue change the look and growth of skin, bones, joints, heart, blood vessels, lungs, eyes, and ears. Others change how these tissues work. Many, but not all, are rare.

Common heritable disorders of connective tissue include:

- **Ehlers-Danlos syndrome** mostly affects the skin and joints. Connective tissue becomes weak, which can cause loose joints and fragile, sagging skin.
- [Epidermolysis bullosa](#) affects the skin, causing blisters.
- [Marfan syndrome](#) can affect the heart, blood vessels, lungs, eyes, bones, and ligaments.

People with this syndrome may be unusually tall and thin, with long arms and legs.

- **[Osteogenesis imperfecta](#)** causes bones to break easily. Sometimes they break for no obvious reason.

Who gets heritable disorders of connective tissue?

Heritable disorders of connective tissue can affect anyone. Some of these disorders are seen at birth. Others are seen later in life.

If you have a heritable disorder of connective tissue, it was passed down to you from one or both of your parents.

What are the symptoms of heritable disorders of connective tissue?

Each heritable disorder of connective tissue has its own symptoms. Some examples are:

- **Bone growth problems:** Bones may become brittle, too long, or too short.
- **Joint issues:** Joints may be too loose or too tight.
- **Skin problems:** Skin may be loose, hang in folds, or blister.
- **Blood vessel damage:** Blood vessels may be weak or become blocked.
- **Height issues:** Some types of disorders cause people to be unusually tall or short.
- **Head and facial structural problems:** Certain disorders can make the head and face look different from others.

Is there a test for heritable disorders of connective tissue?

To diagnose heritable disorders of connective tissue, doctors look at:

- Family history.
- Medical history.
- Results from a physical exam.
- Lab tests, which may confirm some disorders.

You may wish to seek genetic counseling if you wish to have a child. A genetic counselor can help you estimate the risk of having a child with the disease. The counselor can also give you information about tests that look for the problem gene in you or your child.

How are heritable disorders of connective tissue treated?

Heritable disorders of connective tissue are a wide range of disorders, each requiring a specific treatment. Treatments can include:

- **Regular check-ups** with your doctor to keep track of tissue changes.
- **Medicines or nutritional supplements** such as:
 - Vitamin B₆ to correct a liver enzyme problem.
 - Drugs to slow the widening of blood vessel coming from the heart.
 - Drugs to strengthen brittle bones.

Living with heritable disorders of connective tissue

Maintaining general health is important if you have a heritable disorder of connective tissue. You should talk to your doctor about a plan that includes:

- A nutritious diet.
- Exercise.
- Healthy lifestyle habits.

For more info

U.S. Food and Drug Administration

Toll free: 888-INFO-FDA (888-463-6332)

Website: <https://www.fda.gov>

Drugs@FDA at <https://www.accessdata.fda.gov/scripts/cder/daf>. Drugs@FDA is a searchable catalog of FDA-approved drug products.

Centers for Disease Control and Prevention, National Center for Health Statistics

Website: <https://www.cdc.gov/nchs>

American Academy of Orthopaedic Surgeons

Website: <https://www.aaos.org>

American Academy of Dermatology

Website: <https://www.aad.org>

Genetic Alliance

Website: <https://www.geneticalliance.org>

National Organization for Rare Disorders

Website: <https://www.rarediseases.org>

National Society of Genetic Counselors

Website: <https://www.nsgc.org>

Dystrophic Epidermolysis Bullosa Research Association of America, Inc.

Website: <https://www.debra.org>

Ehlers-Danlos National Foundation

Website: <https://www.ednf.org>

National Association for Pseudoxanthoma Elasticum, Inc.

Website: <http://napeusa.com>

Marfan Foundation

Website: <https://www.marfan.org>

Osteogenesis Imperfecta Foundation

Website: <http://www.oif.org>

PXE International

Website: <https://www.pxe.org>

If you need more information about available resources in your language or other languages, please visit our webpages below or contact the NIAMS Information Clearinghouse at NIAMSInfo@mail.nih.gov.

- [Asian Language Health Information](#)
- [Spanish Language Health Information](#)