



## Points To Remember About Epidermolysis Bullosa

- Epidermolysis bullosa is a group of rare diseases that cause fragile skin that leads to blistering and tearing.
- The disease is caused by one or more mutated (changed) genes you received from your parents.
- Although there is no cure for the disease, your doctor treats the symptoms. Treatments may include managing pain and itch, treating wounds caused by the blisters and tears, and helping you cope with the disease.
- Living with epidermolysis bullosa can be hard; however, you can take steps to care for your skin to help prevent blisters from forming and get help to cope.

## What is epidermolysis bullosa?

Epidermolysis bullosa is a group of rare diseases that cause fragile skin that leads to blisters and tearing. Tears, sores, and blisters in the skin happen when something rubs or bumps the skin. They can appear anywhere on the body. In severe cases, blisters may also develop inside the body.

The symptoms of the disease usually begin at birth or during infancy and range from mild to severe.

## Who gets epidermolysis bullosa?

Anyone can get epidermolysis bullosa. It occurs in all racial and ethnic groups and affects males and females equally.

What are the types of epidermolysis bullosa?

There are four major types of epidermolysis bullosa. Doctors determine the type of epidermolysis bullosa based on:

- The place on your body where the skin changes.
- The gene mutation that may have caused the disease.

The symptoms of epidermolysis bullosa vary depending on the type you have. Everyone with the disease has fragile skin that blisters and tears easily. The skin changes and blisters can cause pain and itching. Other symptoms can include:

- Thick skin on the palms of the hands and soles of the feet.
- Rough, thick, or missing fingernails or toenails.
- Blisters inside the mouth.
- Changes in the color of the skin.
- Scarring, which can cause tightening of your skin, muscles, or other tissue in your body.

People who have blisters inside their bodies may have problems with nutrition.

What causes epidermolysis bullosa?

Mutations (changes) to genes that you inherit from your parents cause most forms of epidermolysis bullosa. Genes carry information that determines which features are passed to you from your parents. We have two copies of most of our genes—one from each parent. People with the disease have one or more genes that carry the incorrect instructions to make certain proteins in the skin. You may get this changed gene from one or both parents.

## Is there a test for epidermolysis bullosa?

There is no one test for epidermolysis bullosa. To see if you have the disease, doctors may:

- Ask about your family and medical history, because most types of epidermolysis bullosa are passed down in families.
- Do a physical exam and look at the skin closely, which can help doctors identify where the skin is separating to form blisters.
- Perform a skin biopsy, which helps doctors identify which layers of the skin are affected and determine the type of epidermolysis bullosa you have.
- Order genetic testing to identify specifically which gene mutations you may have.

## How is epidermolysis bullosa treated?

There is no cure for epidermolysis bullosa. The goals of treatment are to prevent and control symptoms by:

- Managing pain and itch with medications.

- Protecting skin and caring for blisters and wounds, such as by using appropriate bandages and changing them when needed.
- Treating and preventing infection by using antibiotics and washing your hands before caring for your skin or changing bandages.
- Maintaining or improving your ability to move areas of your body that may have tightened.
- Finding recipes and foods that are easy to chew, swallow, and digest.

## Who treats epidermolysis bullosa?

You may see one of the following types of doctors:

- Dermatologists, who specialize in conditions of the skin, hair, and nails.
- Clinical geneticists, who diagnose and treat children and adults with genetic disorders.
- Nurse educators, who specialize in helping people understand their overall condition and set up their treatment plans.
- Occupational therapists, who teach how to perform activities of daily living safely.
- Pediatricians, who diagnose and treat children.
- Physical therapists, who teach ways to build muscle strength while keeping the skin protected.
- Primary care physicians, who diagnose and treat adults.
- Registered dietitians, who teach about nutrition and meal planning.

## Living with epidermolysis bullosa

Living with epidermolysis bullosa can be hard; however, you can take steps to care for your skin to help prevent blisters from forming and get help to cope.

Your doctor may recommend the following:

- Keep your skin cool. Never apply anything hot to the skin, and avoid using water higher than your body temperature when bathing.
- Wear loose fitting, soft clothing to avoid rubbing against the skin.
- Keep rooms at a cool, even temperature.
- Apply lotion to the skin to reduce rubbing and keep the skin moist.
- Use sheepskin on car seats and other hard surfaces.
- Wear mittens at bedtime to help prevent scratching while asleep.

You may find it helpful to find a community or online support group. Some people may find it helpful to speak to a mental health professional about coping with the disease.

## 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>

### **National Society of Genetic Counselors**

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

### **American Academy of Dermatology**

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

### **The Dystrophic Epidermolysis Bullosa Research Association of America, Inc.**

Website: <http://www.debra.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](mailto:NIAMSInfo@mail.nih.gov).

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