

What is EB?

EB is a family of **rare genetic disorders involving the skin and skin-like tissues** that share a common way in which wounds are formed

epidermolysis bullosa

refers to the outer layer of skin (epidermis)

means breakdown

a kind of blister

People with EB have **extremely fragile skin** leading to constant blisters or skin tears that can cause wounds that are difficult to heal. These blisters and wounds are caused by **changes to proteins** that help hold layers of skin together and support skin structure

Blisters and wounds can occur from **friction**, such as something rubbing or scratching the skin or tissue. They can form anywhere on the skin, and sometimes inside the body, such as the lining of the mouth

There are 4 types of EB:

- 1 **EBS**
EB simplex
- 2 **JEB**
Junctional EB
- 3 **DEB**
Dystrophic EB, including Dominant DEB (**DDEB**) and Recessive DEB (**RDEB**)
- 4 **Kindler Syndrome**

EB is a genetic condition. What does that mean?



Genetic conditions are caused by mutations in genes



Normally, certain genes provide instructions to make proteins



In EB, mutated genes provide altered instructions that change the function or amount of protein being made



These protein changes impact the structure and strength of the skin

How is EB inherited?



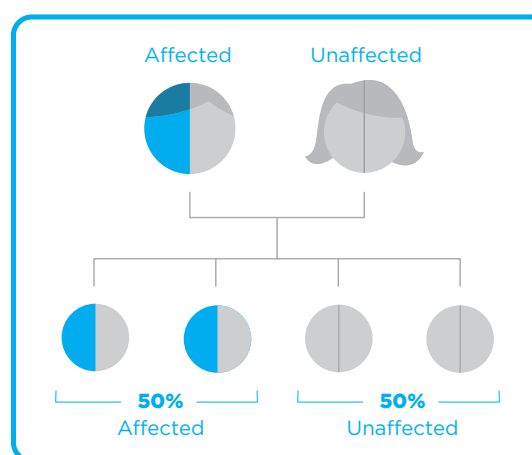
Sometimes, EB occurs **spontaneously** in an individual because of a new genetic mutation that his or her parents did not have



But usually, EB is **inherited** from one or both parents who either have EB or carry a mutated EB gene

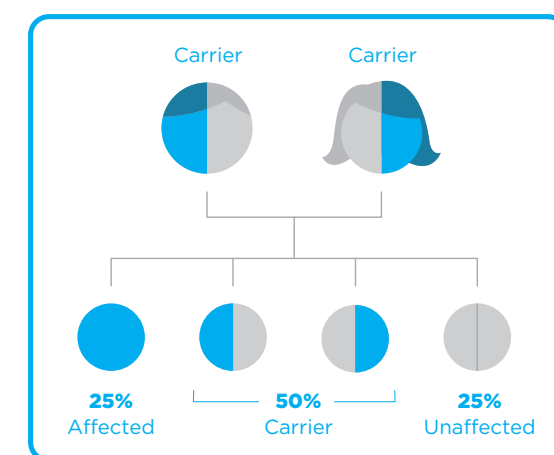
DOMINANT (EBS, DDEB)

- If 1 parent is affected by EB and 1 parent is not affected, there is about a 50% chance that each child born could have EB
- A child who does not inherit a mutated gene from an affected parent will not develop EB or pass it on to his or her children



RECESSIVE (JEB, RDEB, KINDLER SYNDROME)

- A child must inherit 2 copies of a recessive mutated gene to be affected
- If both parents are carriers of a mutated gene but are not affected by EB, there is about a 25% chance that each child born could have EB
- If a person only inherits 1 copy of a mutated gene, that person is a carrier



What are some key facts about EB?



a **chronic** disease that can worsen with **age**



affects **males and females** equally and can occur in all ethnicities



signs and symptoms vary **widely**



not contagious



estimated to affect about **500,000** individuals worldwide



causes pain and can **physically** and **emotionally** impact daily living

What do these words mean?

Anemia

A condition in which a person has fewer red blood cells or hemoglobin than normal, resulting in fatigue

Dermis

The inner layer of skin

DNA

Basic unit that allows for the transmission of genetic information from one generation to the next and contains instructions, or code, for making proteins

Epidermis

The outer layer of skin

Esophagus

The tube that leads from the mouth through the throat to the stomach

Gene

A part of a chromosome in a cell transferred from parent to offspring that influences inherited traits

Mutation

A permanent error in the DNA code

Reflux

A backward flow of the stomach contents into the esophagus

Squamous cell carcinoma

A type of skin cancer

Want to learn more?

Talk to your doctor or nurse. These additional resources can also provide support and information to help you to understand EB*:

→ **Dystrophic Epidermolysis Bullosa Research Association (DEBRA) International**
debra-international.org

→ **debra of America**
debra.org

→ **DEBRA UK**
debra.org.uk

→ **EB Research Partnership (EBRP)**
ebresearch.org

→ **Sohana Research Fund**
sohanaresearchfund.org

→ **National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS)**
niams.nih.gov

→ **National Organization for Rare Disorders (NORD)**
rarediseases.org

→ **European Organization for Rare Disorders (EURORDIS)**
eurordis.org

*Please note that the opinions expressed by the organizations above do not necessarily reflect those of Amicus. Amicus does not maintain and is not responsible for the content of communications for the listed organizations or their websites.

A VISUAL GUIDE

TO UNDERSTANDING

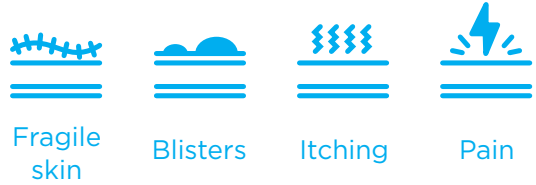
EPIDERMOLYSIS BULLOSA



INFORMATION FOR
PEOPLE WITH
EPIDERMOLYSIS BULLOSA
(EB) AND THEIR FAMILIES

What might someone with EB experience?

INDIVIDUALS WITH EB EXPERIENCE:



Symptoms usually first appear in babies and toddlers but sometimes don't appear until adolescence

Symptoms can range from mild to severe

All areas of the skin can be affected, both inside and outside the body

Signs and symptoms differ depending on the type of EB, but all types can cause acute and chronic pain

SKIN STRUCTURE: SITES OF PRIMARY BLISTER FORMATION

EBS

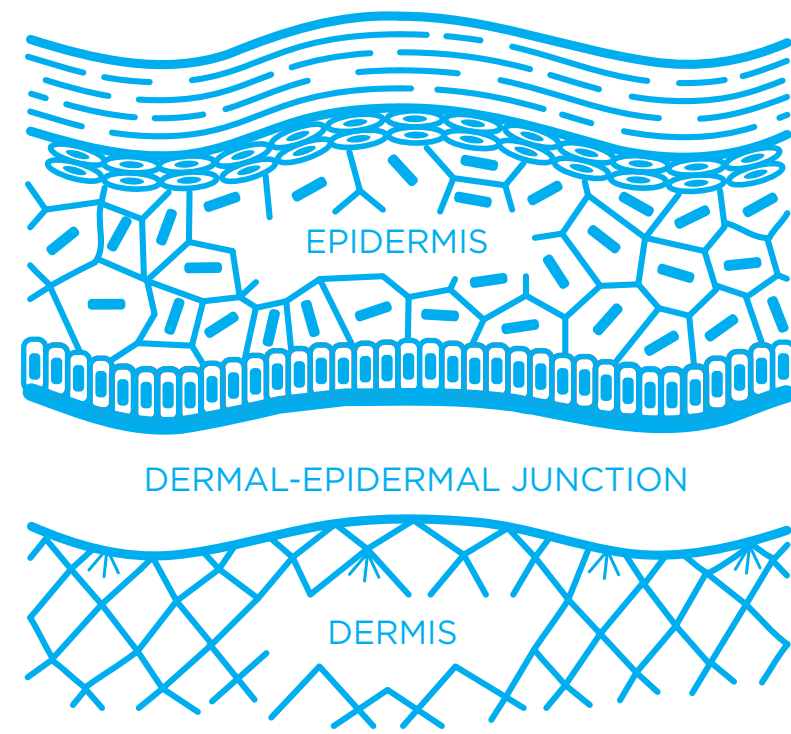
- Symptoms range from mild to severe
- Blisters form on the top layer of skin, known as the epidermis
- Blisters typically appear on the hands and feet but can be widespread over the entire skin

JEB

- Symptoms range from mild to severe
- Blisters form between the epidermis and dermis
- Blisters can occur on the skin and inside the body, such as in the linings of the mouth and esophagus

DEB

- Symptoms range from mild to severe
- Blisters form in the dermis
- There are 2 types of DEB:
 - 1 DOMINANT**
Wounds appear on hands, elbows, knees, and feet
 - 2 RECESSIVE**
Widespread blistering and scarring occur, and there is increased risk of a type of skin cancer called squamous cell carcinoma

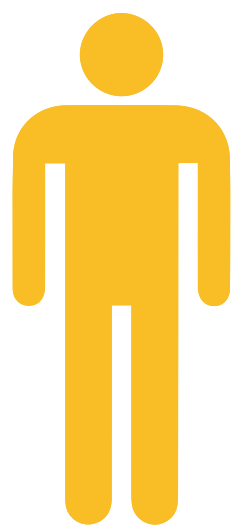


KINDLER SYNDROME

- A rare type of EB
- Skin sunburns easily
- Blisters form on any layer of skin or internal organs
- There is increased risk of squamous cell carcinoma inside the mouth

Adapted with permission from Dystrophic Epidermolysis Bullosa Research Association of America (debra of America).

How does EB affect daily living?



PHYSICAL IMPACT

- Daily wound care can be painful and time consuming and can include soaking baths and constant dressing changes
- Wounds and blisters can occur all over the body, which may make it difficult to perform daily activities
- Chronic wounds may cause scar tissue, which may lead to deformities of hands and feet that limit dexterity and mobility
- Itching is a common problem that may lead to disruption of sleep and the ability to focus. During sleep, scratching can cause or worsen wounds
- The clear outer layer of the eye (cornea) can become injured, which may cause pain, excessive tear formation, or discharge
- Gastrointestinal issues and malnutrition may result from difficulty swallowing, narrowing of the esophagus due to scar tissue, reflux, lactose intolerance, and constipation
- Anemia and fatigue, which can vary across EB types, can be due to having a chronic disease, or factors such as chronic blood loss or malnutrition
- Social isolation can result from fear of further trauma and physical limitations
- Stress and depression may be related to changes in appearance and limitations in daily activities



PSYCHOLOGICAL IMPACT

How is EB treated?



Symptoms are addressed with daily wound care and bandaging, along with prescription medication for itch and pain management



Prevention of infection, scarring, and rigid joints

Currently, there is no cure for EB; however, potential treatments are being investigated:



Wound care advancements



Protein replacement



Stem cell transplant



Gene therapy

What are some things that may help?

Although this is a serious, life-altering condition, these daily tips may help with managing symptoms



Follow your doctor's recommendations for managing wounds



Keep skin cool by avoiding exposure to hot temperatures



Protect vulnerable skin sites by wearing gloves and padding around elbows and knees



Keep skin moisturized to minimize itching, reduce friction, and prevent skin from cracking



Avoid tight clothing, hard shoes, internal seams, and tags to reduce friction



Treat blisters when they appear as recommended by your doctor, and use nonadhesive bandages and dressings



Pad eyeglasses to protect the nose and ears

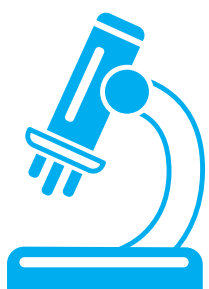


Consider hobbies and noncontact sports to avoid risk of skin trauma



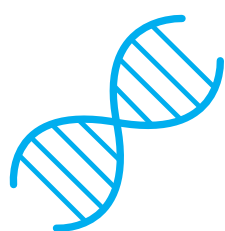
Maintain a healthy diet; additional calories and protein are needed to help with skin healing

A doctor suspects EB—what might happen next?



SKIN BIOPSY

A small sample of affected skin tissue from open or unhealed wounds is taken and examined to find protein deficiencies and structural flaws



GENETIC TESTING

A blood sample is taken to determine whether the condition was inherited from one parent or both parents



PRENATAL TESTING

When there is a family history of EB, prenatal testing may be performed