**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) and 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.

**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
- Estimated to affect about 500,000 individuals worldwide
- Not contagious

There are 4 types of EB:

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

**What is EB?** EB is a genetic condition. What does that mean?

Want to learn more?

- **Sohana Research Fund**
- **EB Research Partnership (EBRP)**
- **DEBRA UK**
- **EB Research South Africa**
- **Sohana Research Fund**

Want to learn more?

- **Amaranthus Therapeutics, Inc.** Cranbury, N.J.
- **NP/NPS/01/0280/GLO916**

**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 have EB.

**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.

**How is EB inherited?**

- A child must inherit 2 copies of a recessive mutated gene to be affected.

- **Carriers**
  - **AFFECTED**
  - **UNAFFECTED**

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.

**A VISUAL GUIDE TO UNDERSTANDING EPIDERMOLYSIS BULLOSA**

**What do these words mean?**

- **Anemia**
  - A condition in which a person has fewer red blood cells or hemoglobin than normal, resulting in fatigue.
- **DNA**
  - The inner layer of skin.
- **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 understand EB:

- **Dystrophic Epidermolysis Bullosa Research Association (DEBRA) International**
  - debar-international.org
- **debra of America**
  - debra.org
- **DEBRA UK**
  - ebresearch.org
- **EB Research Partnership (EBRP)**
  - ebrsearch.org
- **Sohana Research Fund**
  - sohanaresearchfund.org

*Please note that the opinions expressed by the organizations above do not necessarily reflect those of Amicus. Amicus does not sponsor and is not responsible for the content of communications for the listed organizations or their websites.*
How does EB affect daily living?

- **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?**
  - Follow your doctor’s recommendations for managing symptoms.
  - Keep skin cool by avoiding exposure to hot temperatures.
  - Keep skin moisturized to minimize itching, reduce friction, and prevent skin from cracking.
  - Treat blisters when they appear as recommended by your doctor, and use nonadhesive bandages and dressings.
  - 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.
  - Pad eyeglasses to protect the nose and ears.
  - Avoid tight clothing, hard shoes, internal seams, and tags to reduce friction.
  - Protect vulnerable skin sites by wearing gloves and padding around elbows and knees.
  - Follow your doctor’s recommendations for managing wounds.

- **IMPACT**
  - Acute and chronic pain
  - Loss or malnutrition
  - Gastrointestinal issues and malnutrition
  - Excessive tear formation, or discharge
  - 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
  - Narrowing of the esophagus due to scar tissue
  - Increased risk of squamous cell carcinoma

- **A VISUAL GUIDE TO UNDERSTANDING EPIDERMOLYSIS BULLOSA (EB)**

- **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
    - Blister form on hands, elbows, knees, and feet
    - 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
  - Blister form on any layer of skin or internal organs
  - There is increased risk of squamous cell carcinoma inside the mouth

- **INDIVIDUALS WITH EB EXPERIENCE:**
  - Fragile skin
  - Blistering
  - Itching
  - Pain

- **How is EB treated?**
  - 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.

- **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.