CONDITION

Bullosa hereditaria

Learn about bullosa hereditaria, an older term for inherited epidermolysis bullosa, including symptoms, genetic causes, diagnosis, wound care, treatment, complications, and follow-up.

WHAT IT IS
Group of inherited skin-fragility conditions

BODY SYSTEM
Skin

MAIN SYMPTOMS
Blisters, open wounds, pain, scarring

WHAT CAUSES IT
Inherited or new genetic changes

HOW IT IS DIAGNOSED
Genetic testing or specialized skin biopsy

MAIN TREATMENT
Wound care and skin protection

CAN IT BE CURED?
No, care manages wounds and complications

IS IT AN EMERGENCY?
Sometimes, with trouble breathing or uncontrolled bleeding

LONG-TERM OUTLOOK
Widely variable with possible lifelong care

Overview

Bullosa hereditaria is an older term usually used for inherited epidermolysis bullosa , often shortened to EB. EB is a group of rare genetic conditions in which the skin is unusually fragile. Friction, pressure, heat, or minor injuries can cause blisters, wounds, and skin loss.

The term "bullosa hereditaria" does not identify one exact form of EB. If it appears in a medical record, ask which current diagnosis and subtype it refers to. The main inherited EB types are epidermolysis bullosa simplex, junctional epidermolysis bullosa, dystrophic epidermolysis bullosa, and Kindler epidermolysis bullosa.

These types differ according to where the skin separates and which protein is affected. Some mainly cause blisters on the hands and feet. Others affect large areas of skin and tissues inside

the mouth, throat, digestive tract, eyes, or urinary system.

EB is not contagious and cannot spread through touch. It is not caused by poor hygiene or something a parent did during pregnancy. Because the condition is genetic, it may be inherited from one or both parents, or it may result from a new genetic change in the affected person.

Severity varies widely. Some people develop fewer blisters with age and lead active lives with practical skin protection. People with more extensive disease may need lifelong wound care, nutrition support, pain control, rehabilitation, and monitoring for complications.

Symptoms

Symptoms often begin at birth or during infancy, but milder forms may not become clear until a child starts crawling, walking, or taking part in activities that create friction. In some people, the condition is not recognized until later in life.

Skin findings may include:

Blisters may also develop inside the body because the mouth, esophagus, eyes, and other surfaces are lined with fragile tissue. Possible symptoms include mouth sores, painful swallowing, food sticking in the esophagus, hoarseness, constipation, eye discomfort, or pain with urination.

Repeated injury and scarring can reduce joint movement. In severe dystrophic EB, fingers or toes may gradually become joined by scar tissue. Dental enamel may be poorly formed in some junctional forms, increasing the risk of tooth decay.

Symptoms alone cannot determine the exact EB type. Several forms can look similar, especially in newborns and young children.

- Blisters after rubbing, pressure, heat, or minor injury
- Open wounds, crusting, bleeding, or slow healing
- Blisters on the hands and feet after walking or physical activity
- Scarring or small white bumps called milia
- Thickened skin on the palms and soles
- Changes in the fingernails or toenails
- Itching, pain, or increased sensitivity

Causes and risk factors

EB is caused by disease-related changes in genes that make proteins responsible for skin strength. These proteins connect skin cells to one another or attach the outer layer of skin to deeper tissue. When a protein is missing or does not work properly, the skin layers can separate under minor stress.

The four main groups are defined by the level of separation:

More than one inheritance pattern is possible. In autosomal dominant EB, a change in one copy of a gene can cause the condition. An affected parent may pass that change to a child. In autosomal recessive EB, both copies of a gene are affected. The parents are often healthy carriers, meaning they each have one altered copy.

A person may also have EB because of a new, or de novo, genetic change that was not found in either parent. The inheritance pattern and chance of passing EB to a child depend on the specific gene variant. A genetic counselor can explain these issues using the family's test results.

Ordinary contact, food, infection, and vaccination do not cause inherited EB. Heat and friction can worsen blistering, but they do not create the underlying condition.

- Epidermolysis bullosa simplex: Separation usually occurs within the outer skin layer.
- Junctional epidermolysis bullosa: Separation occurs near the junction between the outer and deeper skin layers.
- Dystrophic epidermolysis bullosa: Separation occurs below that junction and usually involves abnormal type VII collagen.
- Kindler epidermolysis bullosa: Separation can occur at several levels and may be associated with sensitivity to sunlight.

Diagnosis

A clinician may suspect EB from the pattern of blistering, the age when symptoms began, wound healing, scarring, nail changes, and family history. Evaluation is often coordinated by a dermatologist or an EB specialty center.

Genetic testing is important because it can identify the affected gene and often confirm the EB subtype. A blood or saliva sample may be used. Results can guide follow-up, clarify inheritance, and help determine whether a person may qualify for a targeted treatment or clinical trial.

A skin biopsy may also be needed. A small sample is taken from the edge of a newly induced blister or from carefully selected skin. Specialized methods can show where the skin layers

separate and whether certain structural proteins are present. These methods include immunofluorescence mapping and, less commonly, electron microscopy.

Other assessments depend on the person's symptoms. They may include blood tests for anemia and nutrition, growth checks, dental and eye examinations, or evaluation of swallowing. Tests should be chosen carefully because adhesive devices and routine procedures can injure fragile skin.

When the genetic cause is known, people planning a pregnancy may discuss reproductive options with a genetic counselor. These can include testing during pregnancy or testing embryos created through in vitro fertilization. These choices are personal and require specialist counseling.

Treatment and management

Management depends on the EB type, age, wound pattern, pain, nutrition, mobility, and organs involved. Care is often provided by a team that may include dermatology, nursing, primary care, pain management, nutrition, dentistry, rehabilitation, gastroenterology, ophthalmology, surgery, and genetic counseling.

Wound care aims to protect the skin, support healing, control drainage, and reduce infection. Dressings are usually soft, nonstick, and secured without ordinary adhesive tape. A wound-care clinician can demonstrate how to remove dressings gently and how to protect areas exposed to pressure.

Intact blisters may continue to enlarge. An EB team may recommend draining selected blisters while leaving the blister roof in place as a natural cover. Patients and caregivers should receive hands-on instruction before doing this at home.

Daily prevention may include reducing friction, keeping rooms comfortably cool, choosing soft clothing, padding pressure points, and modifying shoes. Babies with EB need special handling techniques. Lifting under the arms can damage skin, so caregivers may be taught to support the head, trunk, and buttocks instead.

Pain and itching should be assessed directly. Options may include medicines, cooling measures, protective dressings, sleep support, and techniques to reduce distress during dressing changes. Treatment is individualized because pain may come from open wounds, inflammation, infection, nerve irritation, or scar tissue.

Nutrition is especially important when wound healing increases calorie and protein needs or when mouth and esophageal problems make eating difficult. A dietitian may suggest changes in food texture or nutrient intake. Some people need treatment for iron-deficiency anemia,

vitamin or mineral deficiencies, constipation, or poor growth. Feeding support may be considered when eating by mouth is not enough.

Physical and occupational therapy can help preserve movement and independence. Splints, stretches, activity adjustments, and surgery may be used for contractures or severe hand scarring. Dental care should begin early, with techniques adapted to fragile mouth tissue.

Some targeted treatments are available for particular forms of EB. In the United States, topical gene therapy is approved for wounds in people with dystrophic EB who have mutations in the COL7A1 gene. A topical birch triterpene treatment is also approved for wounds associated with dystrophic and junctional EB. Eligibility, availability, expected benefits, and possible adverse effects should be discussed with an EB specialist. These treatments do not replace protection of the skin or broader medical care.

Clinical trials are studying additional gene, cell, protein, and wound-healing treatments. Participation is optional and should include a clear discussion of potential benefits, burdens, and uncertainty.

Complications

Complications depend strongly on the EB subtype and severity. Open skin can become colonized with bacteria without causing an infection. A true infection is more likely when there is increasing pain, warmth, swelling, spreading redness, pus, an unpleasant new odor, fever, or a sudden change in the wound.

Repeated wounds can cause scarring, joint tightening, reduced mobility, and changes in the hands and feet. Chronic inflammation and blood loss may contribute to anemia. Extensive healing needs, mouth pain, dental disease, swallowing problems, and constipation can affect nutrition and growth.

Scarring in the esophagus can narrow the passage from the mouth to the stomach. This may cause food to stick or make swallowing painful. Eye injury can cause light sensitivity, tearing, or pain. Certain EB subtypes can also affect the kidneys, urinary tract, breathing passages, heart muscle, or bones.

People with severe recessive dystrophic EB have an increased risk of aggressive cutaneous squamous cell carcinoma, a type of skin cancer. Regular skin examinations and prompt assessment of suspicious areas are important. Warning changes can include a wound that behaves differently from usual, develops a raised or firm area, repeatedly bleeds, becomes more painful, or does not heal as expected.

The emotional effects also matter. Visible wounds, lengthy dressing changes, sleep disruption,

pain, and limits on school, work, or relationships can create significant stress. Psychological and social support can be included as part of EB care.

Follow up

Routine follow-up should be based on the EB subtype and individual needs. Visits may review wounds, pain, itching, nutrition, growth, movement, dental health, emotional well-being, and supplies. People at increased skin-cancer risk need regular full skin examinations on a schedule set by their specialist.

Contact the care team promptly for a wound that changes from its usual pattern, worsening swallowing, reduced food or fluid intake, new eye symptoms, increasing weakness, or signs of anemia. Fever, spreading redness, pus, increasing warmth, or rapidly worsening pain should receive same-day medical advice because they may indicate infection.

Seek urgent assessment for severe eye pain, sudden vision changes, inability to swallow liquids, repeated vomiting, signs of dehydration, or uncontrolled bleeding.

Call emergency services for trouble breathing, blue or gray lips, severe chest symptoms, fainting, confusion, or a rapidly worsening illness. These are emergency concerns rather than routine EB follow-up.

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