CONDITION

Epidermolysis bullosa

Epidermolysis bullosa is a group of genetic conditions that makes the skin unusually fragile and prone to blisters.

WHAT IT IS
Inherited conditions causing extremely fragile skin

BODY SYSTEM
Skin and sometimes internal body linings

MAIN SYMPTOMS
Blisters, wounds, pain, fragile skin

WHAT CAUSES IT
Inherited changes in skin-supporting genes

HOW IT IS DIAGNOSED
Genetic testing, sometimes a specialised skin biopsy

MAIN TREATMENT
Wound care, symptom control, specialist therapies

CAN IT BE CURED?
No - current treatments manage damage

IS IT AN EMERGENCY?
Sometimes - breathing trouble or severe infection

LONG-TERM OUTLOOK
Mild to life-limiting, according to subtype

Overview

Epidermolysis bullosa, often shortened to EB, is the name for a group of rare genetic conditions that make the skin unusually fragile. Everyday friction, pressure or minor injuries can cause blisters and open wounds.

Some forms mainly affect the hands and feet. Others cause widespread wounds and can affect the mouth, throat, eyes, digestive tract or urinary tract. Nails, hair and teeth may also be affected.

EB happens when proteins that normally hold layers of skin together are missing or do not work properly. The depth at which the skin separates helps define the main types:

Each type has several subtypes. Severity can differ greatly, including among people with the

same broad type.

EB is not contagious. You cannot catch it through touch, shared objects or contact with wounds. This article focuses on inherited EB. Epidermolysis bullosa acquisita is a separate, acquired autoimmune condition.

- EB simplex , where separation usually occurs within the outer skin layer
- Junctional EB , where separation occurs at the junction between skin layers
- Dystrophic EB , where separation occurs below that junction
- Kindler EB , which can cause separation at different levels

Symptoms

Blisters and skin wounds are the main features. They may be present at birth, appear during infancy or become noticeable later when a child starts crawling or walking. Milder forms may not be recognised until repeated rubbing from shoes, sports or manual work causes symptoms.

Possible skin and nail symptoms include:

Repeated scarring can make fingers or toes join together and may restrict movement around joints. This is more common in some forms of dystrophic EB.

EB can also affect tissues inside the body. Blisters or scarring in the mouth and throat may make eating or swallowing difficult. Dental enamel may be weak in some types, increasing the risk of tooth decay.

Other possible symptoms include constipation, painful bowel movements, eye irritation, hoarse crying or breathing, and pain when passing urine. Poor food intake and ongoing wound healing can contribute to slow growth, low iron levels and tiredness.

Not everyone develops these problems. The symptoms and their effects are strongly linked to the genetic subtype.

- Blisters after friction, pressure, heat or minor injury
- Painful open wounds that heal slowly
- Scarring or tiny white bumps called milia
- Thickened skin on the palms and soles
- Nails that are thick, damaged or missing
- Itching, which may lead to scratching and further skin damage

Causes and risk factors

Inherited EB is caused by a change in a gene involved in making or organising the proteins that strengthen skin. When these proteins are faulty or missing, layers of skin can separate too easily.

The altered gene may be inherited in a dominant pattern, meaning one changed copy can cause the condition. It may instead follow a recessive pattern, meaning both copies must be changed. Parents of a child with recessive EB are often unaffected carriers.

Sometimes EB results from a new genetic change that was not inherited from either parent. Once the exact change is identified, a genetics specialist can explain what it may mean for siblings, future pregnancies and the affected person's own children.

EB is not caused by anything a parent did during pregnancy. Diet, hygiene and ordinary skin contact do not cause it.

Diagnosis

A specialist may suspect EB from the pattern of blistering, when it began, where it occurs and whether relatives have similar symptoms. Appearance alone cannot reliably identify every subtype.

Genetic testing can confirm the diagnosis by finding the relevant gene change. It often identifies the subtype and inheritance pattern, which can guide care and family planning.

A specialised skin biopsy may also be used. A tiny skin sample is examined with techniques such as immunofluorescence mapping or electron microscopy. These tests show where the skin layers separate and which structural proteins are affected. This is different from an ordinary biopsy used to examine a suspicious skin growth.

Testing may involve a dermatologist, geneticist and laboratory experienced in EB. Genetic counselling can help you understand the results and their implications for relatives.

Treatment and management

There is no general cure for inherited EB. Treatment aims to protect the skin, help wounds heal, control pain and itching, and prevent or treat complications. Care is usually coordinated through a specialist EB or dermatology service.

Dressings should protect wounds without sticking to them. Wound care may include gentle

cleansing, non-adhesive dressings and padding over areas exposed to pressure. Blisters are sometimes drained in a controlled way while leaving the blister roof in place, but families need practical training from their EB team before doing this.

Management may also include:

Regular movement and physiotherapy can help preserve joint movement and strength. Occupational therapy may suggest changes to clothing, shoes, school equipment, workspaces and daily activities.

For certain EB subtypes, wound-applied treatments are available in some countries. These include a topical gene therapy for wounds in dystrophic EB and a birch-bark-based gel for some wounds in dystrophic or junctional EB. Eligibility, access and expected benefits are specific to the treatment and region. These therapies do not remove the need for ongoing skin care.

Bone marrow or stem cell transplantation has been studied for severe forms but carries major risks. It is not routine care for most people with EB.

- Pain relief before dressing changes, bathing or procedures
- Treatment for itching and sleep disruption
- Antibiotics like amoxicillin or doxycycline when a bacterial infection is diagnosed
- Nutrition support for increased healing needs or swallowing problems
- Dental, eye, hand, foot and movement care
- Treatment for constipation, reflux or low iron levels
- Surgery for severe scarring, narrowed swallowing passages or other complications

Complications

Open skin can allow bacteria to enter. Local infection may cause increasing redness, warmth, swelling, pain, discharge or odour. Infection can occasionally spread into the bloodstream and become an emergency.

Ongoing wounds and blood loss can cause anaemia, meaning too few healthy red blood cells. Increased nutritional needs, mouth pain or swallowing difficulty may affect growth and energy. Some people need a feeding tube to support nutrition.

Scarring may narrow the oesophagus, the tube carrying food to the stomach. It can also limit hand movement, tighten joints or affect the eyelids and urinary tract.

People with some forms of severe dystrophic EB have a high risk of an aggressive skin cancer

called squamous cell carcinoma. A wound that changes, grows, becomes raised, bleeds more than usual or does not follow its normal healing pattern needs prompt specialist assessment. Routine skin checks are especially important for those at increased risk.

Severe subtypes may also affect breathing, kidneys, the heart or overall health. These problems are not expected in every person with EB.

Prevention

Inherited EB cannot currently be prevented after conception, because the genetic change is present from birth. It is not caused by avoidable behaviour.

Genetic counselling can explain inheritance and reproductive options when the family's gene change is known. This information supports personal decisions; it does not predict the exact severity of every possible case.

Although EB itself cannot be prevented, some skin damage can be reduced. Helpful measures may include keeping the environment comfortably cool, choosing soft clothing, avoiding adhesive products directly on fragile skin and padding areas exposed to rubbing. Complete avoidance of movement is not recommended because safe activity supports mobility, development and independence.

Living with it and what to expect

The course of EB is determined mainly by its subtype. Some people have blistering mostly on the hands and feet and remain active with practical adjustments. In certain forms of EB simplex, blistering may become less frequent with age, although heat and friction can remain triggers.

More severe forms cause lifelong wounds, scarring and effects on other parts of the body. Needs may change over time as a child grows, starts school or work, or develops complications. Regular follow-up helps monitor nutrition, mobility, teeth, eyes, swallowing and skin changes.

Daily wound care can take considerable time and may affect sleep, school, employment and family life. Pain and visible skin differences can also affect mood and confidence. Psychological support, peer networks and adjustments at school or work can be valuable parts of care.

Most people learn which materials, movements and routines protect their skin without unnecessarily limiting daily life. Care plans should support participation and independence,

rather than treating all activity as unsafe.

Follow-up schedules are individual. People at risk of scarring, nutritional problems or skin cancer usually need ongoing specialist review even when their condition appears stable.

When to get medical help

Arrange prompt medical advice for a new or changing wound, worsening pain, difficulty eating, poor weight gain, constipation that is not settling, eye pain or reduced movement. Increasing redness, warmth, swelling, pus, bad odour or fever may indicate infection.

A wound that becomes raised, unusually hard, repeatedly bleeds, grows quickly or does not heal in its usual way needs urgent assessment. This is particularly important for people with recessive dystrophic EB because of the risk of squamous cell carcinoma.

Seek emergency help for:

Routine blistering without these warning signs is usually managed through the established wound-care plan and specialist follow-up.

- Trouble breathing, noisy breathing or blue-grey lips
- Sudden inability to swallow liquids or saliva
- Severe weakness, confusion, fainting or difficulty waking
- Signs of a serious spreading infection, such as fever with rapid deterioration
- Bleeding that does not stop with gentle, appropriate pressure

Common questions

Is epidermolysis bullosa contagious?

No. Inherited EB is caused by genetic changes and cannot spread through skin contact, dressings, blood or shared household items.

Can epidermolysis bullosa get better with age?

Some milder forms, particularly certain types of EB simplex, may cause fewer blisters as the skin thickens with age. Other forms remain stable or develop more complications from repeated wounds and scarring. The genetic subtype gives the most useful guide, but it cannot predict every person's course.

Can people with EB go to school, work or exercise?

Many can, with adjustments based on skin fragility, pain and mobility. Soft clothing, padded equipment, planned dressing care and alternatives to high-friction activities may help. Safe movement is important for strength and joint flexibility.

Can someone with EB have children?

Yes. The chance of passing EB on depends on the exact gene change and whether inheritance is dominant or recessive. Genetic counselling can provide information based on the person's confirmed test result.

Why should adhesive bandages be avoided?

Ordinary adhesive dressings can pull away fragile skin when removed and create a larger wound. Specialist non-adhesive dressings and gentle securing methods are usually preferred.

Is there a cure for epidermolysis bullosa?

There is no cure that corrects all inherited EB throughout the body. Some targeted treatments can help selected wounds or subtypes, and research into gene, cell and protein-based treatments continues. Wound protection and complication monitoring remain essential.

Sources

- NHS. Epidermolysis bullosa
- Has C, Bauer JW, Bodemer C, et al. Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. British Journal of Dermatology . 2020;183:614-627
- GeneReviews. Dystrophic Epidermolysis Bullosa
- DEBRA International. Clinical Practice Guidelines
- Denyer J, Pillay E, Clapham J. Best practice guidelines for skin and wound care in epidermolysis bullosa. Wounds International; 2017
- European Medicines Agency. Filsuvez: European public assessment report