CONDITION

Dowling-meara syndrome

Dowling-Meara syndrome is an inherited skin-fragility condition that causes widespread blistering from minor friction or heat.

WHAT IT IS
Severe inherited skin-fragility condition causing early blistering

BODY SYSTEM
Skin and sometimes mouth and throat

MAIN SYMPTOMS
Widespread blisters, skin pain, thickened palms and soles, nail changes

WHAT CAUSES IT
Inherited keratin gene changes, often newly arising

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

MAIN TREATMENT
Wound care, pain control, infection prevention, specialist support

CAN IT BE CURED?
No - current care controls symptoms

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

LONG-TERM OUTLOOK
Often improves with age, but skin fragility remains

Overview

Dowling-Meara syndrome is an older name for generalised severe epidermolysis bullosa simplex , or generalised severe EBS. It is an inherited condition that makes the skin unusually fragile. Minor rubbing, pressure, injury, or heat can cause painful blisters.

The condition affects the epidermis, the skin's outer layer. Proteins that normally help skin cells withstand stress do not work properly, so cells near the bottom of the epidermis can break apart.

Blistering usually begins at birth or in early infancy. It may affect large areas and can worsen during hot weather, illness, or increased activity. Blisters often appear in clusters.

Generalised severe EBS is lifelong, although blistering often becomes less severe as a child

grows. Thickened skin on the palms and soles may become more noticeable later.

This condition is one form of epidermolysis bullosa, or EB. Other forms involve different genes and skin layers and may require different care, so an accurate diagnosis is important.

Symptoms

Symptoms vary, even among relatives with the same genetic change. Common features include:

Blisters may develop on the hands, feet, limbs, and trunk. Skin beneath clothing, nappies, shoes, or medical equipment may be affected because of rubbing or pressure. Hot weather and fever can increase blistering.

Blisters sometimes form inside the mouth, making feeding or eating uncomfortable. Involvement of the throat or breathing passages is less common but requires prompt assessment.

Most blisters heal without the heavy scarring associated with some other forms of EB. Temporary skin-colour changes or small white bumps called milia may remain. Repeated blistering can still affect comfort, movement, sleep, and daily activities.

Babies with extensive skin damage may lose fluid and body heat. Pain and mouth blisters can also interfere with feeding.

- Blisters beginning at birth or in early infancy
- Widespread or grouped blisters, sometimes arranged in rings
- Blistering after friction, pressure, heat, sweating, or minor injury
- Pain, itching, and tenderness
- Thickened skin on the palms and soles
- Changes to the fingernails or toenails

Causes and risk factors

Dowling-Meara syndrome is usually caused by a harmful change in the KRT5 or KRT14 gene. These genes provide instructions for keratin 5 and keratin 14, proteins that form a strong framework inside skin cells.

When either protein does not work correctly, cells can break under mechanical stress, producing a blister within the lower epidermis.

The condition is usually inherited in an autosomal dominant pattern. One altered gene copy can cause it, and an affected person generally has a 1 in 2 chance of passing the change on in each pregnancy.

Sometimes the genetic change is new, or de novo, and neither parent has the condition. It may arise in an egg, sperm, or early embryo. It is not caused by anything a parent did during pregnancy.

Friction and heat trigger blisters but do not cause the underlying condition. EBS is not caused by infection, allergy, or poor skin care, and it is not contagious.

Diagnosis

A specialist may suspect generalised severe EBS from when blistering began, its appearance and triggers, and any family history of fragile skin.

Genetic testing can confirm the diagnosis by identifying a disease-causing change. A test may examine several genes linked to EB because different types can look similar in infancy.

A small skin biopsy may be needed if genetic testing is unavailable or inconclusive, or if doctors need to distinguish quickly between EB types. Specialised methods such as immunofluorescence mapping or electron microscopy can show where the skin separates and whether important proteins are present.

An ordinary biopsy examination may not classify EB accurately. Testing is best coordinated through a centre experienced in inherited skin-fragility conditions.

When the genetic change is known, genetic counselling can explain the result, inheritance pattern, testing options for relatives, and reproductive choices.

Treatment and management

There is no current cure that corrects generalised severe EBS throughout the skin. Treatment aims to protect the skin, support healing, control pain, and identify complications early.

A specialist EB or dermatology team usually plans wound care. Soft, non-stick dressings can protect wounds while limiting adhesive contact, pressure, and overheating.

Many teams teach families to drain larger blisters carefully while leaving the blister roof in place as a protective layer. The treating team should demonstrate the method and equipment. Pulling off intact skin can enlarge the wound and increase pain.

Pain and itching may come from new blisters, open wounds, dressing changes, or thickened

skin. Management can include age-appropriate medicines, cooling, suitable dressings, distraction, and changes to wound-care routines.

Antibiotics like amoxicillin or doxycycline are not needed for every wound. A clinician may prescribe them when bacterial infection is present. Unnecessary use can cause side effects and encourage antibiotic resistance.

Other care may include:

Research into treatments targeting the underlying genes or proteins continues. Treatments approved for other EB forms should not be assumed to work for EBS.

- Nutrition support if pain or mouth blisters affect eating
- Clothing and footwear adjustments to reduce friction
- Treatment for thickened palms and soles
- Physiotherapy or occupational therapy if pain limits movement
- Dental care adapted for fragile skin and mouth tissues
- Emotional, school, workplace, and social support

Complications

Open blisters can become infected. Warning signs include increasing redness, warmth, swelling, pain, pus, unpleasant odour, or fever. Infection can occasionally spread beyond the skin.

Extensive wounds may cause fluid, salt, or heat loss, especially in newborns and young infants. Mouth blisters may reduce feeding, contributing to dehydration, slow weight gain, or nutritional deficiencies.

Repeated blistering can cause painful thickening of the palms and soles, called palmoplantar keratoderma. This may make walking, writing, or using tools difficult. Sweating can worsen friction.

Nails may thicken or fall off, and skin-colour changes can occur. Pain, visible wounds, poor sleep, and time-consuming dressing routines may also affect mental health, education, work, and family life.

Severe infection or major fluid loss can be dangerous, particularly in infancy. Specialist follow-up helps identify and manage these risks early.

Prevention

The genetic change causing Dowling-Meara syndrome cannot be prevented through diet, lifestyle, or pregnancy care.

When a family-specific change is known, a genetics service can explain testing options before or during pregnancy. These are personal choices, and no option suits every family.

Some blisters may be prevented by reducing avoidable friction, pressure, and overheating. Soft clothing, well-fitting footwear, padded equipment, and a cooler environment may help. Plans should balance skin protection with normal movement, activity, and development.

Living with it and what to expect

The first months and years are often the most difficult because blistering may be widespread and infant skin is especially vulnerable. Feeding, dressing, holding, nappies, and medical equipment may require adjustments.

For many people, blistering becomes less frequent or widespread later in childhood or around puberty, although improvement is not guaranteed. The skin remains more fragile than usual, and heat, sweating, illness, prolonged walking, or repeated pressure may continue to trigger symptoms.

Thickened palms and soles can become more prominent with age, even if generalised blistering improves. Painful feet may affect sport, travel, or work involving prolonged standing. Changes to footwear, schedules, and equipment can help maintain independence.

Most people can participate in school, relationships, employment, and family life, although they may need accommodations and regular wound care.

Routine follow-up may review wounds, pain, nutrition, mobility, foot care, sleep, and emotional well-being. Report a new blistering pattern, persistent wounds, or reduced ability to manage daily activities rather than assuming these changes are unavoidable.

When to get medical help

Call emergency services if there is:

Seek urgent medical assessment for a baby or child who cannot drink, is repeatedly vomiting, has far fewer wet nappies than usual, or shows other signs of dehydration.

Prompt assessment is also needed for fever with rapidly spreading redness, severe swelling, worsening pain, pus, or marked illness. These may indicate a spreading infection.

Contact the EB or dermatology team soon if wounds are not healing as expected, mouth blisters limit food or fluids, pain is becoming harder to control, or thickened skin affects walking or hand use.

Routine appointments can cover dressings, footwear, nutrition, school or work adaptations, genetic counselling, and long-term symptom control.

- Difficulty breathing, noisy breathing, or rapidly worsening throat swelling
- Severe weakness, confusion, fainting, or unusual difficulty waking
- Heavy bleeding that does not stop with gentle pressure

Common questions

Is Dowling-Meara syndrome the same as epidermolysis bullosa?

It is one type of epidermolysis bullosa simplex, now called generalised severe EBS. Other EB forms involve different genes or skin layers and may cause different complications.

Is it contagious?

No. It is caused by a genetic change and cannot spread through touch, shared clothing, blood, or open blisters.

Will a child grow out of it?

The condition is lifelong. Blistering often improves with age, but the skin usually remains fragile. Thickened skin on the hands and feet may become more troublesome later.

Should EBS blisters be popped?

Larger blisters are often drained to prevent them from spreading, but the blister roof should usually remain in place. An EB team should demonstrate safe blister care to reduce infection and further injury.

Can I pass it on to my children?

With an autosomal dominant genetic change, each child generally has a 1 in 2 chance of inheriting it. Genetic testing and counselling can clarify the pattern for a particular family.

Can someone with this condition exercise?

Many people can be active, but friction, pressure, heat, and sweating may trigger blisters. Suitable footwear, clothing, padding, cooling, rest breaks, and gradual changes can help.

Sources



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