CONDITION

Acanthosis bullosa

Learn about acanthosis bullosa, an older term associated with epidermolytic ichthyosis, including its symptoms, genetic causes, diagnosis, skin care, treatment, and warning signs.

WHAT IT IS
Rare inherited skin disorder

BODY SYSTEM
Skin

MAIN SYMPTOMS
Blisters, peeling, thick scaly skin, redness

WHAT CAUSES IT
Inherited genetic changes

HOW IT IS DIAGNOSED
Skin biopsy, genetic testing

MAIN TREATMENT
Skin care and symptom management

CAN IT BE CURED?
No, genetic change cannot be removed

IS IT AN EMERGENCY?
Sometimes, severe dehydration or extensive skin loss

LONG-TERM OUTLOOK
Lifelong with widely varying severity

Overview

Acanthosis bullosa is an older and uncommon name associated with epidermolytic ichthyosis , a rare inherited skin disorder. Modern medical records are more likely to use the terms epidermolytic ichthyosis, epidermolytic hyperkeratosis, or bullous congenital ichthyosiform erythroderma.

Because older terminology can be inconsistent, ask a dermatologist to confirm what "acanthosis bullosa" means in a particular medical record. It should not be confused with acanthosis nigricans , which causes dark, thickened areas of skin and may be related to insulin resistance. It is also different from the group of conditions called epidermolysis bullosa , although both can cause fragile skin and blisters.

Epidermolytic ichthyosis affects keratin proteins that help give the outer skin layer strength.

When these proteins do not work normally, skin cells can break apart under minor stress. Infants may therefore have widespread redness, blisters, and raw areas. As a child grows, blistering often becomes less prominent while thick, scaly skin becomes more noticeable.

The condition is lifelong, but its severity varies widely. Some people have limited thickening and occasional blisters. Others need regular wound care, infection monitoring, and treatment for extensive scaling. The disorder is not contagious and cannot be passed to another person through touch.

Medical care matters because the skin is a protective barrier. When that barrier is damaged, especially in a newborn, the body may lose fluid and heat more easily. Broken skin can also become infected. Long-term treatment focuses on protecting the skin, reducing excessive scale, treating symptoms, and supporting daily life.

Symptoms

Symptoms can change with age. A newborn may have red, tender-looking skin with superficial blisters or peeling. Blisters can form after pressure, rubbing, heat, or handling, although they may also appear without an obvious trigger.

During childhood and adulthood, the skin often becomes increasingly thick and scaly. This thickening is called hyperkeratosis , meaning that too much keratin builds up in the outer skin layer. It may be widespread or especially noticeable in skin folds, over joints, or in areas exposed to friction.

Possible features include:

Warm weather, sweating, tight clothing, and repeated friction may worsen blistering. Thick scale in skin folds can hold moisture and create an environment where bacteria or yeast grow more easily. Odor does not always mean that there is a serious infection, but new pain, swelling, drainage, or rapidly increasing redness should be assessed.

Hair, teeth, and internal organs are not usually the main areas affected. However, severe skin disease can interfere with sleep, mobility, school, work, exercise, and emotional well-being.

- Blisters, peeling, or shallow raw areas
- Thick, rough, or ridged scales
- Redness and skin tenderness
- Itching, burning, or pain
- Cracking around joints or other areas that move

- Increased skin odor caused by trapped moisture and bacterial growth
- Thickened skin on the palms and soles in some genetic forms
- Difficulty with movement when thick skin builds up around joints

Causes and risk factors

Epidermolytic ichthyosis is usually caused by a harmful change, called a pathogenic variant , in either the `KRT1` or `KRT10` gene. These genes provide instructions for making keratin 1 and keratin 10. Both proteins help create a strong framework inside cells in the outer layer of skin.

When one of these keratins is altered, skin cells are less able to tolerate friction and pressure. This explains the blistering seen early in life. Over time, the skin responds by producing a thicker outer layer, leading to scaling and hyperkeratosis.

The condition usually follows an autosomal dominant inheritance pattern. This means that a person generally needs one altered copy of the gene to have the disorder. An affected parent can pass the altered gene to a child, but the severity in the child cannot always be predicted from the parent's symptoms.

Many affected people are the first person in their family to have the condition. In these cases, the genetic change may have occurred spontaneously in the egg, sperm, or early embryo. It was not caused by something a parent did during pregnancy.

Some genetic changes occur after an embryo has started developing. This is called mosaicism , meaning that only some cells carry the variant. Mosaic disease may appear as localized bands or patches of thickened skin, sometimes called an epidermal nevus. In some circumstances, a person with mosaicism may have reproductive cells carrying the variant and could have a child with more widespread disease.

A family history raises the likelihood of an inherited form, but having no family history does not rule it out. The condition is not caused by poor hygiene, food choices, allergies, or an infection.

Diagnosis

Diagnosis often begins with a dermatologist's examination and a detailed history. The doctor may ask when symptoms began, whether blisters were present at birth, what triggers skin breakdown, and whether relatives have similar skin findings.

A skin biopsy may be performed. During this procedure, a small sample of numbed skin is removed and examined under a microscope. Epidermolytic ichthyosis has a characteristic

pattern called epidermolytic hyperkeratosis. This describes breakdown of cells in the upper epidermis together with thickening of the outer skin layer.

This microscopic pattern can also occur in other skin conditions, so biopsy results must be considered along with the person's symptoms and distribution of affected skin.

Genetic testing can look for variants in `KRT1`, `KRT10`, or a broader panel of genes associated with inherited ichthyosis. Testing may confirm the diagnosis, clarify inheritance, and help distinguish epidermolytic ichthyosis from similar disorders. However, a negative test does not always exclude the condition.

A clinician may need to consider other causes of newborn blistering or lifelong scaling, including:

Families considering genetic testing may benefit from genetic counseling. A genetic counselor can explain possible results, family implications, and reproductive testing options without directing a family toward a particular choice.

- Other inherited forms of ichthyosis
- Epidermolysis bullosa
- Infections that cause blisters
- Staphylococcal scalded skin syndrome
- Other genetic or inflammatory skin disorders

Treatment and management

There is no single treatment that removes the underlying genetic change. Management is adjusted to age, symptoms, skin area involved, and how easily the skin blisters. A dermatologist familiar with inherited skin disorders can help create a practical care plan.

For newborns with extensive skin loss, treatment may initially take place in a hospital or specialist nursery. Priorities include gentle handling, temperature control, fluid balance, nutrition, wound care, pain relief, and monitoring for infection. Adhesive products may damage fragile skin and are generally avoided unless a specialist recommends a suitable option.

Regular use of bland moisturizers or ointments helps support the skin barrier and reduce cracking. Products are usually applied gently rather than rubbed in. A clinician can help select products that do not sting or irritate open skin.

Bathing can soften thick scale and make gentle removal easier. The skin should not be aggressively scrubbed or peeled, because trauma may cause new blisters. Some people

benefit from prescribed creams that loosen scale, called keratolytics . These products can irritate broken skin and may not be appropriate over large areas, particularly in infants or young children, unless a clinician provides guidance.

Practical measures can reduce friction and overheating:

Blisters may need nonstick dressings and careful wound care. A clinician can demonstrate whether intact blisters should be left alone or managed in another way. Repeatedly puncturing or cutting blisters without medical instruction can introduce infection.

Antiseptic washes or antimicrobial treatments are sometimes used for recurrent bacterial growth, odor, or infection. Antibiotics or antifungal medicines are appropriate only when a clinician identifies or strongly suspects an infection. Routine, unsupervised antibiotic use can cause side effects and contribute to antibiotic resistance.

Oral retinoids, medicines related to vitamin A, may reduce severe skin thickening. They can also increase tenderness or blistering in some people. Retinoids require specialist supervision because they can cause significant side effects, require laboratory monitoring, and can cause severe harm to a developing fetus. Treatment decisions should consider both skin improvement and effects on daily life.

Pain, itching, sleep problems, and emotional distress deserve direct attention. Physical therapy may help when thick or cracked skin limits joint movement. Support from a psychologist, social worker, school team, or patient organization may also be useful.

- Choose soft, breathable clothing and well-fitting shoes.
- Keep living and sleeping areas comfortably cool.
- Use padding or protective dressings on frequently injured areas.
- Change damp clothing when practical.
- Avoid picking or forcefully removing thick scale.

Complications

Infants with widespread blistering or peeling can develop dehydration, difficulty maintaining body temperature, electrolyte problems, or infection. These risks are greatest when large areas of the skin barrier are disrupted.

Later complications may include recurrent bacterial or fungal skin infections, painful cracks, chronic odor, limited movement, and sleep disruption. Thick skin on the soles can make walking uncomfortable. Repeated inflammation may also change skin color.

Seek prompt medical advice for increasing pain, warmth, swelling, pus, foul drainage, fever, or redness that is spreading beyond the usual affected area. These symptoms may indicate an infection that needs treatment.

Emergency care is appropriate for trouble breathing, unusual sleepiness or confusion, fainting, signs of severe dehydration, or a rapidly worsening illness with extensive skin loss. A young infant with fever, poor feeding, fewer wet diapers, or marked changes in alertness should receive urgent medical assessment.

The visible nature of the condition can affect confidence, relationships, and participation in activities. These effects are medical concerns, not personal failings, and can be discussed during routine care.

Follow up

Routine follow-up is usually coordinated by a dermatologist. Visits may review blister frequency, scaling, pain, itching, infections, mobility, and the effect of treatment on school, work, or sleep. Children may need growth, nutrition, and developmental monitoring if skin disease is extensive.

People using prescription treatments may need laboratory tests or other safety monitoring. Follow-up is also a chance to adjust skin-care routines as weather, activity, age, or symptoms change.

Contact the care team sooner than planned if there are frequent new infections, rapidly increasing blistering, poorly controlled pain, difficulty walking, or treatment side effects. Genetic counseling can be revisited when a person is planning a pregnancy or wants updated information about testing.

Sources

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