CONDITION

Aplasia cutis congenita

Learn how aplasia cutis congenita affects a newborn's skin, how it is evaluated, treatment options, possible complications, and when to seek urgent care.

WHAT IT IS
Rare congenital absence of skin

BODY SYSTEM
Skin

MAIN SYMPTOMS
Missing skin at birth, open wound, thin shiny membrane, smooth hairless scar

WHAT CAUSES IT
Usually unknown, sometimes inherited

HOW IT IS DIAGNOSED
Newborn physical examination

MAIN TREATMENT
Wound care, sometimes surgery

CAN IT BE CURED?
Sometimes - wounds heal but scars remain

IS IT AN EMERGENCY?
Sometimes - heavy or persistent bleeding

LONG-TERM OUTLOOK
Good for shallow isolated lesions, worse with deep defects or associated conditions

Overview

Aplasia cutis congenita is a rare condition in which a baby is born without skin over a specific area of the body. It is often shortened to ACC . The affected area developed before birth and may look like an open wound, a thin membrane, or a smooth scar.

ACC most often affects the scalp, especially near the top or back of the head. It can also occur on the trunk, arms, or legs. A baby may have one area or several areas of missing skin. The size and depth can vary widely.

Many cases involve only the upper layers of skin and heal with careful wound care. Deeper defects may extend through the tissue under the skin and, on the scalp, into the skull or the protective covering around the brain. These deeper areas need prompt specialist assessment because of the risk of bleeding, infection, or injury to structures beneath the wound.[1,2]

ACC is a description of a physical finding rather than one single disease. It may occur by itself, with another birth difference, or as part of a genetic condition. The letters "ACC" are also used for unrelated conditions, including adenoid cystic carcinoma and agenesis of the corpus callosum. Using the full name helps prevent confusion.

Symptoms

The affected area is present at birth. Its appearance depends on how much tissue is missing and whether healing began before delivery.

Common features include:

Small, shallow defects may not cause other symptoms. An open area may be tender and can ooze a small amount of clear fluid or blood. A deeper scalp defect may appear sunken or pulsate with the baby's heartbeat.

ACC affecting the arms or legs may occur with missing or underdeveloped fingers, toes, or nails. Fragile or blistering skin elsewhere can suggest an inherited skin disorder. Some babies have additional differences involving the limbs, heart, blood vessels, nervous system, or other organs.

Changes such as increasing redness, warmth, swelling, pus, an unpleasant odor, or worsening tenderness may indicate infection and need prompt medical review. Rapid or persistent bleeding requires emergency attention.

- A round, oval, or irregular area where skin is absent
- A moist red wound or shallow ulcer
- A thin, shiny, or partly transparent membrane
- A dry, smooth scar if the area healed before birth
- Little or no hair growing from a healed scalp lesion
- A ring of longer or darker hair around a scalp defect, sometimes called the "hair collar sign"
- One lesion or several lesions arranged in a line or pattern

Causes and risk factors

Skin begins forming early in pregnancy. ACC is thought to result when normal skin development is interrupted or when tissue that had formed is damaged before birth. In many

babies, the exact reason cannot be identified.

Possible causes or associated factors include:

One associated condition is Adams-Oliver syndrome , which can include scalp ACC and underdevelopment of the fingers, toes, hands, or feet. Some people with this syndrome also have heart, blood vessel, or nervous system differences.[4]

ACC can also occur with epidermolysis bullosa, a group of inherited disorders that make skin unusually fragile. When missing skin on the legs occurs with blistering or fragile skin, clinicians may consider a form sometimes called Bart syndrome.

Most parents could not have predicted or prevented ACC. A parent should not stop a medicine during pregnancy based only on an association. Medication changes should be discussed with the clinician treating the pregnancy and the condition for which the medicine was prescribed.

- Genetic changes. ACC may run in a family or be part of a genetic syndrome. Different forms can follow different inheritance patterns.
- Reduced blood flow. A disruption in blood supply to a developing area may damage the skin and nearby tissue.
- Amniotic bands. Strands from the membranes around the fetus can rarely interfere with tissue development.
- A multiple pregnancy complication. ACC has been reported when one fetus in a twin pregnancy dies and remains in the uterus, a situation called fetus papyraceus.
- Certain medicines or exposures during pregnancy. ACC has been associated with specific medicines, including the antithyroid medicines methimazole and carbimazole. An association does not prove that a medicine caused an individual baby's condition.[1,3]
- Other congenital conditions. ACC can occur with chromosome disorders or syndromes affecting the skin, limbs, or blood vessels.

Diagnosis

Clinicians usually recognize ACC by examining the baby after birth. The first goal is to determine the lesion's size, depth, location, and condition. The baby's healthcare team will also check whether deeper structures may be involved.

The evaluation may include:

Imaging is not necessary for every small, shallow lesion. It may be considered when a scalp defect is large, deep, near the middle of the scalp, covered by a membrane, pulsating, or associated with unusual hair growth or neurological findings. Ultrasound, magnetic resonance

imaging, or computed tomography may be used to assess the skull, blood vessels, and tissue around the brain. The choice depends on the baby's condition and the information needed.[2,3]

A skin biopsy is usually not needed when the appearance is typical. Removing tissue from a thin or deep scalp lesion could cause injury, so specialists weigh the risks carefully.

Genetic assessment may be recommended when there are multiple lesions, limb differences, fragile skin, developmental concerns, other organ findings, or a family history. Testing may help identify an associated syndrome and clarify whether relatives or future pregnancies could be affected. A normal genetic test does not rule out every possible cause.

- Examining the entire skin surface for other lesions, blisters, or areas of scarring
- Checking the skull, limbs, fingers, toes, and nails
- Looking for signs of infection or active bleeding
- Reviewing pregnancy history, medicines, prenatal imaging, and complications
- Asking about relatives with similar skin, limb, or nail findings
- Performing a general newborn examination for other birth differences

Treatment and management

Treatment depends on the defect's location, depth, size, and underlying structures. A neonatal clinician, dermatologist, plastic surgeon, neurosurgeon, wound-care specialist, or clinical geneticist may be involved.

Small, shallow defects often heal without surgery. Care usually focuses on protecting the wound while new skin grows. The clinical team may recommend gentle cleansing, a nonstick dressing, and a moisture-protecting ointment. Dressings should be changed as directed so they do not pull on delicate new tissue.

Parents and caregivers should wash their hands before touching the area. Products containing alcohol, peroxide, iodine, antibiotics like amoxicillin or doxycycline , or herbal ingredients should not be applied unless the baby's clinician recommends them. Some products can irritate healing tissue, and topical antibiotics are not needed for every wound.

Clinicians monitor open lesions for infection, fluid loss, and healthy tissue growth. They may also watch the baby's temperature and feeding, particularly when the affected area is extensive.

Surgery may be considered when a defect is large, is not healing safely, involves the skull or deeper tissue, or leaves important structures exposed. Options can include closing the wound

directly, using a skin graft, moving nearby skin over the area with a flap, or using tissue expansion later. A skull defect may need separate reconstruction, either during infancy or after the child has grown.[2,3]

There is no single procedure that is best for every baby. Surgery itself may carry risks, including bleeding, infection, wound separation, and problems with a graft or flap. For some large scalp lesions, careful nonsurgical management may still be appropriate if specialists believe the wound can be protected safely. Decisions are based on the individual anatomy and the experience of the treating team.

After healing, a scalp lesion often leaves a hairless scar. Reconstructive treatment may be discussed later if the scar is uncomfortable, restricts movement, breaks down, or causes cosmetic concerns. These decisions can usually be made over time rather than during the newborn period.

Complications

Most small, superficial areas heal well. Possible complications are more likely with open, deep, or extensive defects.

They may include:

A scalp defect over a major vein, such as the superior sagittal sinus, can lead to serious bleeding if the area is injured. Caregivers should not press hard, probe the wound, or try to remove tissue that appears attached. The medical team should give clear instructions for protecting the area at home.

The long-term outlook depends mainly on the defect's depth and whether other conditions are present. A child with an isolated, shallow lesion may need only wound and scar follow-up. A child with an associated syndrome may need monitoring from several specialties.

- Wound infection
- Infection spreading to the skull, tissues around the brain, or bloodstream
- Bleeding from a blood vessel beneath a scalp defect
- Fluid and heat loss through a large open area
- Delayed healing or repeated wound breakdown
- A permanent scar or an area of hair loss
- Incomplete skull closure
- Complications related to an associated genetic or congenital condition

Follow up

Routine follow-up checks whether the wound is closing, the dressing plan is working, and the baby is feeding and growing normally. The healthcare team may photograph or measure the lesion to track healing. Follow-up can also address scar care, hair loss, skull growth, and whether further imaging or surgery is needed.

Keep scheduled appointments even when the area appears better. A thin layer of new skin may remain easy to injure. Ask when bathing, tummy time, hats, car-seat head supports, and other routine activities are safe.

Contact the baby's healthcare team promptly for increasing redness, warmth, swelling, drainage, odor, tenderness, or wound opening. A fever in a young infant needs immediate medical advice. Clear fluid leaking from a deep scalp lesion, a new bulge or pulsation, unusual sleepiness, poor feeding, repeated vomiting, or seizure-like movements also require urgent assessment.

Call emergency services for heavy or rapidly recurring bleeding, bleeding that does not stop with gentle direct pressure as instructed, trouble breathing, unresponsiveness, or a seizure. These are emergency concerns rather than symptoms to wait on until a routine appointment.

Sources

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