

Nonsyndromic aplasia cutis congenita

Description

Nonsyndromic aplasia cutis congenita is a condition in which babies are born with localized areas of missing skin (lesions). These areas resemble ulcers or open wounds, although they are sometimes already healed at birth. Lesions most commonly occur on the top of the head (skull vertex), although they can be found on the torso or limbs. In some cases, the bone and other tissues under the skin defect are also underdeveloped.

Most affected babies have a single lesion. The lesions vary in size and can be differently shaped: some are round or oval, others rectangular, and still others star-shaped. They usually leave a scar after they heal. When the scalp is involved, there may be an absence of hair growth (alopecia) in the affected area.

When the underlying bone and other tissues are involved, affected individuals are at higher risk of infections. If these severe defects occur on the head, the membrane that covers the brain (the dura mater) may be exposed, and life-threatening bleeding may occur from nearby vessels.

Skin lesions are typically the only feature of nonsyndromic aplasia cutis congenita, although other skin problems and abnormalities of the bones and other tissues occur rarely. However, the characteristic skin lesions can occur as one of many symptoms in other conditions, including Johanson-Blizzard syndrome and Adams-Oliver syndrome. These instances are described as syndromic aplasia cutis congenita.

Frequency

Aplasia cutis congenita affects approximately 1 in 10,000 newborns. The incidence of the nonsyndromic form is unknown.

Causes

Nonsyndromic aplasia cutis congenita can have different causes, and often the cause is unknown. Because the condition is sometimes found in multiple members of a family, it is thought to have a genetic component; however, the genetic factors are not fully understood. Researchers suggest that genes important for skin growth may be involved. It is thought that impairments of skin growth more commonly affect the skin at the top of the head because that region needs to be able to grow quickly to cover the fast-growing skull of a developing baby.

In some cases, nonsyndromic aplasia cutis congenita is caused by exposure to a drug called methimazole before birth. This medication is given to treat an overactive thyroid gland. Babies whose mothers take this medication during pregnancy are at increased risk of having the condition. In addition, certain viral infections in a pregnant mother can cause the baby to be born with the skin lesions characteristic of nonsyndromic aplasia cutis congenita. Other cases are thought to be caused by injury to the baby during development.

Inheritance

Most cases of nonsyndromic aplasia cutis congenita are sporadic, which means they occur in people with no history of the disorder in their family. When the condition runs in families, inheritance usually follows an autosomal dominant pattern, which means one copy of an altered gene in each cell is sufficient to cause the disorder. Rarely, the condition appears to follow an autosomal recessive pattern of inheritance, which means both copies of a gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- ACC
- Congenital absence of skin on scalp
- Congenital defect of the skull and scalp
- Congenital ulcer of the newborn
- Scalp defect congenital

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Aplasia cutis congenita (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0282160/>)

Genetic and Rare Diseases Information Center

- Aplasia cutis congenita (<https://rarediseases.info.nih.gov/diseases/5835/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Catalog of Genes and Diseases from OMIM

- APLASIA CUTIS CONGENITA, NONSYNDROMIC; ACC (<https://omim.org/entry/107600>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28aplasia+cutis+congenita%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>)

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