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Aplasia Cutis Congenita

Aplasia cutis congenita (ACC) is a rare congenital disorder characterized by the absence of skin in certain areas of the body, most commonly over the scalp. This condition affects approximately 3 in 10,000 births and presents as localized skin defects, typically involving the midline of the vertex of the scalp. Although these defects are often isolated, they may also occur in multiple locations, including the face, trunk, and limbs.

Clinical Features

ACC lesions are typically non-inflammatory, well-demarcated, and vary in size, ranging from 0.5-10 cm. The lesions are most commonly seen as single defects over the scalp, particularly in the midline, but can also present symmetrically or in multiple areas across the body. The lesions may have a circular, oval, linear, or stellate shape, and the degree of skin involvement can vary.

- *Superficial Lesions*: In cases where only the epidermis is involved, the lesion is generally shallow and often heals with scarring before birth, without significant complications.
- *Deeper Lesions*: Lesions that extend into the dermis or subcutaneous tissue, or occasionally the skull, may present with ulceration and increased risk of complications such as infection.
- *Membranous Aplasia Cutis*: This variant of ACC presents with an underlying flat, white membrane that covers a defect in the skull. This form can be associated with neural tube defects such as encephaloceles or meningoceles, with ultrasound findings revealing misplaced brain tissue outside the skull. The hair collar sign, which is a ring of dark, long hair encircling the lesion, is a notable feature.
- *Bullous Aplasia Cutis Congenita*: This type presents with fibrovascular or edematous connective tissue and is often associated with neural tube defects, supporting the hypothesis that this variant may represent an incomplete neural tube closure defect.
- *Preauricular Dermal Dysplasia*: A less common form of ACC, occurring in front of the ear, which is usually not associated with other extracutaneous anomalies.
- *SCALP Syndrome*: This syndrome is a complex phenotype that includes nevus sebaceous, central nervous system malformations, aplasia cutis congenita, limbal dermoids, and a giant congenital pigmented melanocytic nevus with neurocutaneous melanosis.

Etiology and Pathophysiology

The pathogenesis of ACC is multifactorial, with a variety of mechanisms proposed to contribute to its development. The condition may arise from:

- *Genetic Factors*: Genetic mutations or inherited conditions may predispose individuals to ACC, though specific genetic loci have not been consistently identified across all cases. Chromosome analysis may be recommended if ACC is associated with other congenital anomalies, suggesting a genetic syndrome.
- *Teratogens*: Exposure to teratogenic agents during pregnancy has been implicated in ACC. Notable teratogens include methimazole, carbimazole, misoprostol, and valproic acid. These agents can disrupt normal fetal development, leading to defects in skin formation.
- *Compromised Skin Vasculature*: Vascular disruption during embryonic development can lead to areas of skin necrosis and failure to form properly.
- *Trauma*: In some cases, mechanical trauma during fetal development may result in skin defects.
- *Fetus Papyraceous*: A rare association between fetus papyraceous and bilaterally symmetric ACC has been noted, wherein the compromised twin's developmental disturbances lead to skin defects in the surviving twin.

A hypothesis posits that scalp ACC may be linked to the scalp hair whorl, which is located at the point of maximum tensile force during the period of rapid brain growth. The disruption of skin in this area may occur around 10–15 weeks of gestation, during which time hair direction and patterning are established, coinciding with brain development.

In membranous ACC, amniotic band formation resulting from early rupture of the amniotic membranes may lead to constriction and skin defects in the affected area.

Diagnostic Workup

While no specific laboratory abnormalities are consistently associated with ACC, diagnostic workup may include:

- *Chromosomal Analysis*: This may be considered in cases where ACC presents with other systemic anomalies suggestive of a genetic syndrome.
- *Ultrasound*: This imaging modality is essential in detecting underlying neural tube defects, particularly in membranous ACC cases associated with encephaloceles or meningoceles.
- *Histological Examination*: In some cases, histological examination may reveal distinct features that support the diagnosis of ACC, particularly in the bullous variant, which shares characteristics with encephalocele and meningocele tissue.

Treatment and Management

Management of ACC is largely based on the size, depth, and location of the lesion, as well as the presence of associated abnormalities. Treatment strategies may include:

- ***Surgical Intervention***: Surgical repair may be indicated for large or ulcerated lesions, especially if the defect involves deeper layers of skin or underlying structures. Skin grafting may be required in some cases for large defects.
- ***Wound Care***: For superficial lesions that are expected to heal spontaneously, wound care involves keeping the area clean and monitoring for infection.
- ***Management of Associated Anomalies***: If ACC is associated with neural tube defects or other congenital malformations, appropriate referral to specialists (e.g., neurologists, geneticists) for management of these conditions is crucial.
- ***Multidisciplinary Approach***: For complex cases involving SCALP syndrome or other multi-organ anomalies, a multidisciplinary approach involving dermatologists, pediatric surgeons, and neurologists is essential for comprehensive care.

Conclusion

Aplasia cutis congenita is a rare but distinct condition characterized by congenital skin defects, most commonly on the scalp. The etiology is multifactorial, with genetic, environmental, and mechanical factors contributing to its development. The clinical management of ACC is based on lesion characteristics and associated anomalies, with surgical intervention reserved for more severe cases. Early detection and management of associated conditions are essential for improving outcomes, particularly in the context of neural tube defects or other syndromic presentations.

References

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