Read Item - Aplasia Cutis Congenita And Focal Dermal Hypoplasia

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Abstract: Doctor's resource on Aplasia Cutis Congenita And Focal Dermal Hypoplasia

Aplasia Cutis Congenita And Focal Dermal Hypoplasia

Definition
The focal absence of skin may occur anywhere on the body, but around 85% are found on the scalp. The depth of the ulcer is variable, and the defect may comprise epidermis alone, epidermis and dermis, and in some cases the cranial vault. Occasionally lesions heal completely in utero, and the child is born with a scar devoid of appendages.

Focal dermal hypoplasia is an hereditary ectodermal dysplasia, with a number of distinctive clinical features, while focal facial dermal hypoplasia tends to occur as an isolated abnormality.

Epidemiology
Aplasia cutis congenita (ACC) is a congenital anomaly seen in 0.03% of live births.

Aetiology
ACC can be sporadic, familial with an autosomal dominant inheritance or the hallmark of a syndrome.

Pathogenesis
Several hypotheses have been proposed to explain ACC. Most have emphasised mechanical, vascular or developmental factors. The intrauterine development of the scalp occurs from the periphery to the vertex, where the growing islands of skin fuse. Aplasia cutis occurs most commonly over the parietal whorl and usually represents a failure of skin fusion.

Clinical features
Aplasia cutis congenita may present at birth as a sharply circumscribed ulcer with a red raw base simulating a wound or it may have completely healed in utero and present as a scar . Lesions may be oval, linear, rhomboidal or stellate. In up to 20% of cases the aplasia includes the dura with lysis of the underlying skull . The defect is usually 1-2 cm in diameter, but may be much larger. The ulcer usually heal rapidly in the post-natal period to produce an atrophic, or rarely a hypertrophic scar. Most commonly the lesions are solitary (70-75% of cases), however in 20% there are double and in 8% triple lesions. Lesions that involve the calvaria pose a risk of meningitis or haemorrhage.

Focal dermal hypoplasia or Goltz syndrome is an X-linked dominant disorder of ectodermal and mesodermal tissues, lethal in utero in males. Sporadic cases have occurred in males with either an XXY karyotype, a gametic half chromatid mutation or a somatic mutation early in embryogenesis. Females demonstrate Lyonization*, due to random inactivation of the X chromosome, which manifests as streaks of dermal hypoplasia, hypopigmentation and, telangiectasia and herniations or hamartomas of fat following BlaschkoÕs lines. In some cases an autosomal dominant inheritance has been observed and the proposed gene for this variant mapped to chromosome 9.

Osteopathia striata is the radiological hallmark and represents Lyonization of chondroblasts. Other characteristics of this syndrome are an aged leonine facies; absent eyebrows; puckered periorbital skin; a rubbery texture to the nose and chin; and congenital symmetrical scar-like defects on each temple following BlaschkoÕs lines. These scar-like defects are seen in between 10 to 50% of cases and are hairless, pigmented, puckered areas above the eyebrows, that extend upwards and outwards.

Pathology
Histologically, there is atrophy of the dermal and subcutaneous tissues and loss of the pilosebaceous units, but the epidermis is normal. Striated muscle can be seen high in the dermis and may be responsible for the puckered appearance. Focal facial dermal hypoplasia produces very similar symmetrical temporal scar-like lesions, without any associated defects. It is inherited as an autosomal dominant trait.

Investigation
Nil usually required for aplasia cutis.

Associated Features
Aplasia cutis most commonly occur as an isolated defect, however in 20-25% of cases, associated developmental defects are present. The wide spectrum of associated disorders has been classified into nine different sub-types of ACC.

Prognosis
The patch of hair loss is fixed.

Treatment
Parents often suspect obstetric mismanagement to be the cause of the ACC and a detailed explanation of the condition is often required to placate them. If the lesion is deep or does not heal quickly, skin grafting is usually advocated to counter the risk of infection.

Once healed a small lesion requires no treatment. When the child is older, consideration may be given to surgical excision with primary closure to correct the alopecia. For larger areas tissue expanders may be required. Hair transplantation is a reasonable alternative.

Key Points
Congenital absence of skin over the vertex usually occurs as a developmental defect, rather than as a result of obstetric trauma. Healing usually occurs spontaneously to leave a patch of alopecia that can be corrected when the child is older. Associated developmental abnormalities should be sought.

Focal dermal hypoplasia is an unrelated condition that also produces scar-like lesions, but the epidermis is intact. It is an X linked dominant syndrome lethal in males. Focal facial dermal hypoplasia occurs as an isolated autosomal dominant defect.

* The Lyon hypothesis states that:
1. In the somatic cells of female mammals, only one X chromosome is active. The second X chromosome is condensed and inactive and appears in interphase cells as the sex chromatin (Barr body).
2. Inactivation occurs early in embryonic life (12th to 16th day post-fertilisation).
3. The inactive X can be either the paternal or the maternal X (XP or XM) in different cells of the same
individual; but after the decision as to which X will be inactivated has been made in a particular cell, all the
clonal descendants of that cell will abide by that decision and will have the same inactive X. Thus
inactivation is random but fixed. The relative proportions of XP to XM vary from female to female even in
identical twins.
4. If the mutation is present only in somatic cells, all the descendants of these cells will be affected, but
the abnormality will not be transmitted to subsequent generations. However if the mutation is also present

in the gametes then the abnormality is heritable.