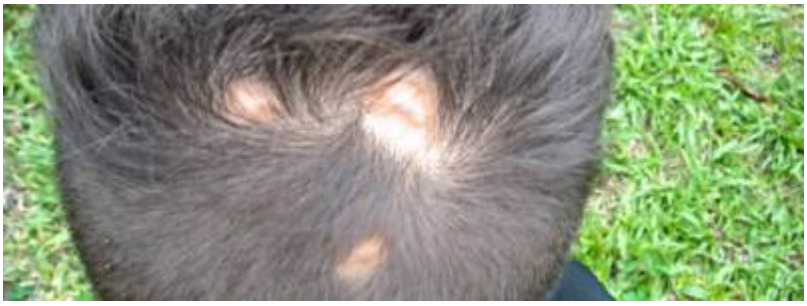
Aplasia Cutis Congenita



Aplasia cutis congenita (ACC) is a rare congenital disorder characterized by noninflammatory, well-demarcated defects of all skin layers, subcutaneous tissue, with possible co-defects in muscles, periosteum, bone and dura

Although the etiology is still uncertain, a variety of possible causes such as genetic syndromes, teratogens, intrauterine infection with varicella zoster or herpes simplex viruses, fetal exposure to cocaine, heroin, alcohol or antithyroid drugs, infarction of the placenta, and amniotic pathologies are described in the literature.

The incidence is 1 to 3/2000 to 10,000 and 25% of the reported cases are familial with a vast majority (69%) showing an autosomal dominant inheritance or part of a syndrome.

Lesions can be multiple and on different surfaces of the body, but most of the cases have solitary scalp lesions (70%).

It may often occur on the parietal and occipital areas and rarely on extremities.

It can also be associated with other physical anomalies such as defects of eyes, extremities, limbs, gastrointestinal system, genito-urinary system, and central nervous system. [1].

Scalp

see Scalp in Aplasia Cutis Congenita

Split cord malformation

ACC has seldom been reported to be associated with SCMs [2) 3) 4)].

Case series

Kuemmet et al. investigated the risk of cranial central nervous system dysraphism in children presenting with aplasia cutis congenita of the head, who presented between 1/1/2000 and 6/15/2016. Inclusion criteria were subjects with aplasia cutis congenita of the head who received CT or MR imaging of the head.

They identified a total of 69 subjects with aplasia cutis congenita affecting the head and who received imaging. The most common location of the aplasia cutis congenita lesion was the vertex scalp (49.3%). The hair collar sign was present in 27.5% of patients. Twelve of 69 patients (17.4%) demonstrated abnormalities of the bone, vasculature, or brain on head imaging. Only one patient had

a diagnosis of encephalocele that required neurosurgical intervention. There was a statistical association between the hair collar sign and the presence of abnormal imaging findings (P = .029), with a negative predictive value of 89.4%.

The incidence of central nervous system dysraphism in patients with aplasia cutis congenita of the head appears to below, and it may not be necessary to image the head of each child presenting with this skin lesion. The hair collar sign may be a marker of underlying defects [5].

References

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