A case of Aplasia Cutis Congenita of scalp with calvarial defect and left parietal lobe cerebral infarction

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Abstract:
Aplasia Cutis Congenita of scalp with calvarial defect is a rare disorder. In large lesions with extensive calvarial defects, treatment in the neonatal period is controversial. Surgical and conservative management both have attending complications. Sagittal sinus thrombosis, bleeding, and meningitis have been reported during conservative approach. The baby presented here had an unusual clinical course of meningoencephalitis with left parietal lobe cerebral infarct. CSF leak and neurologic symptoms were absent and the condition was diagnosed incidentally during MRI scan.

Keyword: Aplasia Cutis Congenita, Calvarial defect, Meningoencephalitis, Cerebral infarct, MRI

Introduction:
Aplasia Cutis Congenita (ACC) a developmental defect of skin occurs with a frequency of 1 in 10,000 births. 500 cases have been reported so far in literature. Scalp lesions constitute 80% of cases. Non scalp defects occur in extremities and trunk. Majority of cases belong to group 1 in Frieden classification. The size of lesion in scalp vary from 0.05 cms to 10 cms in diameter. The scalp defects have calvarial defects in 20% babies. These babies have a predisposition for complications like sagittal sinus thrombosis, haemorrhage, and meningitis. Mortality is as high as 50% in large calvarial defects. Conservative management avoids complications of surgery but exposes the neonate to the risk of infection and haemorrhage. These complications of conservative management occur due to breach in continuity of dura mater. Ischaemic stroke has not been reported as a consequence of this lesion. We present a unique case report of aplasia cutis leading on to stroke during conservative management inspite of absence of breach in dura or obvious CSF leak.
Case Report

A 3 day old term singleton male infant born of non-consanguineous marriage was referred to our hospital for extensive scalp defect. The child was delivered by emergency LSCS for failure of secondary forces. There was smooth perinatal transition and baby could feed from breast soon after birth. There is no history of exposure to teratogenic drugs or radiation. There was no family history of congenital anomalies or genetic disorders. The infant on examination had scalp defect of 10×10 cms over the vertex in the midline. There was an erythematous membrane in place of skin and underlying bone was absent. Cerebral convolutions could just be made out beneath the membrane. No other congenital anomalies were detected. There was no neurological deficit. Other system were normal. Silver sulfadiazine dressing was applied to the defect and antibiotic therapy was instituted. Initial septic work up in child showed qualitative CRP to be positive. Renal function tests and blood counts were normal. Blood culture showed no growth. Imaging studies were done. The CT Brain showed a hypodense lesion in left parietal region of brain suggestive of cerebral infarct. The bone windows for skull with 3D reconstruction showed extensive parietal bone defect and also multiple osteopenic defects all over calvaria and mandible. An MRI with contrast, MR angiogram and MR venogram was done on day 7 of life. A left parietal infarct was present. Foci of hyperintensities, effacement of sulci, periventricular white matter hyperintensity, restricted diffusion, and meningeal enhancement on contrast were also present suggestive of meningoencephalitis. The internal carotids and vertebral arteries were normal on MR angiogram. The dural venous sinuses were also found to be normal on MR venogram. The baby’s CSF exam was normal except for hypoglycorrachia and CSF culture showed no growth. Echocardiography and abdominal ultrasonogram were found to be normal. The baby was given parenteral antibiotics for 21 days. During hospital stay there was no neurological dysfunction in the baby and he was gaining weight on direct breast feeds. Plastic surgery opinion was obtained. A split skin graft followed by a skin flap cover was planned for a later date. At the time of discharge the lesion was showing signs of healing on conservative management of silver sulfadiazine dressings. The baby is currently on follow up is 4 months old and clinically normal on conservative management. Partial skin cover around the edges of the defect has been established.
Discussion:
Aplasia Cutis is congenital defect in the development of the skin which involves defects of some or all layers of skin, underlying subcutaneous tissue and less commonly perosteum and bone. 80% of cases occur on the scalp region, 20% of scalp lesions also show calvarial defects. The lesions may be ulcerated with red glistening base, eczematous, bullous or atrophic at birth. Histopathologically the membrane covering the brain consists of a very thin, flattened layer of cuboidal, cells set in a single layer. The skin bordering these lesions revealed a gradual transition to normal skin. In the intermediate zone, the epidermis thins out to one or two rows of flattened epithelial cells Gross et al. The etiology of aplasia cutis congenital remains uncertain, familial occurrence account for 20-25% cases. The postulated etiologies are genetic, Intrauterine infections, teratogenic drugs and vascular accidents. Syndromes associated with Aplasia cutis include Patau syndrome, Adams-Oliver syndrome and Johanson Bizard syndrome.
Intrauterine herpes simplex and varicella infections have also been correlated with the occurrence of ACC. An arrest in the skin development in embryonic life due to intrauterine vascular abnormality (including placental infarcts and solitary umbilical arteries), intrauterine trauma, or amniotic bands are other proposed etiological factors. Other associated abnormalities include cleft lip and/or palate, abnormal ears, hydrocephalus, brain malformations, meningocele, congenital heart disease, tracheoesophageal fistula, anorectal malformation, absent or polycystic kidney, epidermolysis bullosa like lesions, Golt's syndrome (focal dermal hypoplasia). Frieden proposed classification of ACC into nine groups. This classification was based on the location, pattern of ACC, associated abnormalities, and mode of inheritance. Small defects can be managed conservatively. There is no definite consensus on the management of large defects in newborn period. Some advocate surgery for large lesions greater than 2cms with bone defects. Surgery involves staged procedure like split skin grafting followed by rotation flaps. The risks include graft failure, flap necrosis, haemorrhage, and infection. Therefore, conservative approach is also practiced. Conservative management involves non-adherent dressings, antibiotic creams like Bacitracin or silver sulfadiazine cream and saline application to prevent dessication. The usual reported complications of conservative management have been haemorrhage and infections. Meningitis may occur due to breach in dura mater and CSF leak. Aplasia cutis leading on to meningoencephalitis and stroke is quiet rare. Spread of infection through local vasculature may have served as a portal for meningeal infection observed in this case as there was no breach in dura. Conservative management may still serve as an alternative to early skin grafting in neonatal period if strict asepsis is maintained. Though bone regrowth occurs in, Aplasia cutis scalp is a rare anomaly with varied etiology. Though majority of scalp lesions are small and can be managed conservatively treatment of large lesions with calvarial defects in newborn period is controversial. Both surgery and a conservative approach in neonatal period have their own benefits and attending complications. During a conservative approach, strict wound asepsis is required to avoid CNS infection as local vasculature may serve as portal for infection. Arterial ischaemic Stroke demonstrated in this baby is a very unusual complication of conservative management.

**Conclusion:**

Aplasia cutis scalp is a rare anomaly with varied etiology. Though majority of scalp lesions are small and can be managed conservatively, treatment of large lesions with calvarial defects in newborn period is controversial. Both surgery and a conservative approach in neonatal period have their own benefits and attending complications. During a conservative approach, strict wound asepsis is required to avoid CNS infection as local vasculature may serve as portal for infection. Arterial ischaemic Stroke demonstrated in this baby is a very unusual complication of conservative management.

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