International Journal of Science and Research (IJSR)
ISSN: 2319-7064
SJIF (2022): 7.942

Aplasia Cutis Congenita: A Case Report

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Abstract: ***Aplasia Cuties Congenita (ACC) is a rare, heterogeneous group of congenital disorders, characterized by localized or widespread, complete or partial, absence of skin at birth mostly involving scalp. Incidence is about 3 in 10, 000 births and 500 cases have been reported. Causes include Intra-uterine infections, teratogenic drugs (e.g. methimazole), intra-uterine trauma, vascular compromise, genetic mutations. ACC is clinical diagnosis. USG (skull), MRI (Brain) and bone survey helps to rule out associated malformations. Case: We report a case of full-term newborn female baby, born by non-consanguineous marriage, delivered by Lower-segment Caesarean section (done in view of non-progression of labor and severe pre-eclampsia) with skin defect of 4×5 cms in size on occipital region (Vertex) since birth. No history of intra-uterine or birth trauma, skin infection/ fever/ teratogenic drugs during ANC period. No history of scalp defects/ ACC in family. On head-to-toe examination, 4×5 cms sized, Single, Oval shaped skin defect was present only over vertex of scalp, involving superficial skin layers, with no signs of local infection/ external hemorrhage atsite of defect. USG (Skull) and bone survey were normal. Diagnosis of ACC-I (ACC without associated anomalies) was made. Baby was managed Conservatively with topical antibiotics, skin care and follow up was taken. Conclusion: Aplasia Cutes Congenita: Type I (ACC-I) is a rare condition involving scalp without multiple anomalies. It is Clinical Diagnosis. Prognosis and Management of ACC depends on size, location and severity of defect, underlying cause and associated anomalies. Superficial lesions are treated conservatively, while large defects or deeper lesions involving large bone area require surgical intervention.***

Keywords: Aplasia Cuties Congenita, Congenital disorder, Teratogenic drugs, Intra-uterine infections, Intra-uterine trauma

1. Introduction

Aplasia Cuties Congenita (ACC) is a rare, heterogeneous group of congenital disorders, characterized by localized or widespread, complete or partial, absence of skin at birth mostly involving scalp (in 70% cases), but can involve other body parts too [1]. Incidence of ACC is about 3 in 10, 000 Birth with slightly higher female predominance and nearly 500 cases have been reported in literature till now [2].

Aplasia Cuties Congenita is classified into 9 subtypes based on location and pattern of lesions, associated malformations and mode of inheritance. Causes of ACC includes intrauterine infection (Herpes simplex, Varicella zoster), Teratogenic drugs (Methimazole, carbimazole, Valproate, cocaine), Vascular compromise, Intrauterine trauma, Genetic mutations and Chromosomal abnormalities (e.g. Mutation in ribosomal GTPase BMS-1, UBA2 gene, SUMOylation pathway), etc [3]. Diagnosis of ACC is based on Clinical examination, while Ultrasonography (Skull), MRI (Brain), Bone survey helps to rule out associated congenital anomalies. Complications of ACC Includes local infections, hemorrhage, sagittal sinus thrombosis and intra-cranial infections (e.g. Meningitis) in case of deeper defects.

Management and Prognosis of ACC depend on size and depth of defect, presence of associated abnormalities and risk of complication. Small localized lesions are treated Conservatively, while Larger and deeper defects may require Surgical intervention [4].

2. Case Report

We report a case of full-term newborn female baby, born by non-consanguineous marriage, first by birth order, delivered by Lower-segment Caesarean section (done in view of non-progression of labor and severe pre-eclampsia) with skin defect of 4×5 cms in size on occipital region (Vertex) since birth. There was no history of birth trauma due to forceps or use of vacuum pump. No history of intrauterine trauma. No history of Fever/Skin rash to mother in Antenatal period. There was no history of consumption of any teratogenic drugs (e.g. Methimazole, carbimazole, Valproate, methotrexate, Cocaine, misoprostol, ACE inhibitors) by mother during Antenatal period.

There was no history of scalp skin defects or similar complaints in the family. Family belongs to upper-middle socio-economic status.

Mother was booked case of ANC and have taken folic acid supplementation and 2 doses of tetanus toxoid in first trimester. Five scans were done during ANC period and all were normal. Mother had history of severe pre-eclampsiain in 3rd trimester.

Full term female baby was born by Lower-segment Caesarean section done in view of non-progression of labor and severe pre-eclampsia. Baby cried soon after birth. APGAR score was 8 and 9 at one and five minutes respectively. Birth weight was 2600 grams, Length 55 cms, Head circumference 35 cms. Baby passed meconium and urine soon after birth and started on breastfeeding within 1 hour of birth.

Head to toe examination revealed 4×5 cms sized, Single, Oval shaped, well defined skin defect over vertex of scalp, involving superficial skin layers. There was no signs of local infection or external hemorrhage at the site of defect. Bone involvement was absent. No similar lesions were present at sites other than scalp. Face, ears, eyes, oral cavity, neck, trunk, both upper limbs and lower limbs were normal.

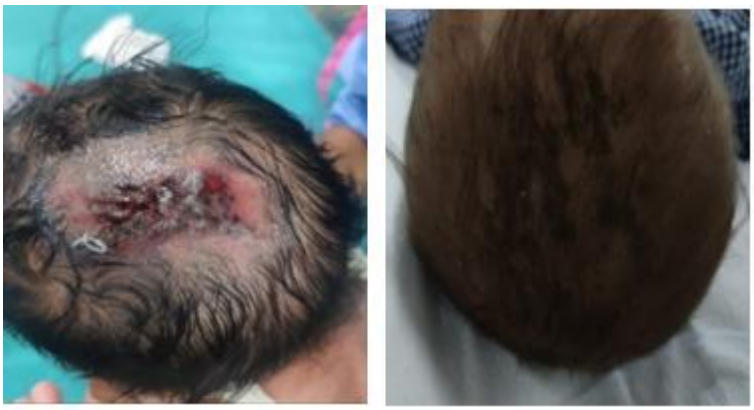

Image - Healed lesion after 4 weeks

The vital signs (Temperature-36.8 °C, Heart rate-146 bpm, BP-64/30 mmHg, Respiratory rate-38/min, SpO2-98% on room air, CRT < 3 sec) were normal. Peripheral pulsations-well felt. Anterior fontanel - at level. On systemic examination, Cardiovascular System-S1S2 heard, No murmur. Respiratory system-Air entry bilaterally equal, no adventitious sounds heard. Per abdomen-Soft, non-tender. CNS-Child was Alert, Active and cried immediately after birth. BCG vaccine was administered within 24 hours of birth.

Investigations:
Ultrasounds of brain and abdomen were normal. Skeletal survey was normal. Inflammatory markers and blood counts were normal with no evidence of infection. Histology of skin defect was not done as parents did not give consent for procedure.

Diagnosis of 'Aplasia Cuties Congenita: Type 1' was made based on clinical examination, bone survey and ultrasonography findings.

Decision of Conservative management was taken as skin defect was small in size and superficial in nature. Baby was commenced on Topical antibacterial cream (Ointment T-bact, local application, twice a day) after Condy's compression (1: 10, 000 diluted Potassium permanganate) and proper skin care. Mother was assured regarding isolated nature of skin defect and absence of associated abnormalities. Follow-up at 4 weeks of age showed evidence of complete recovery of skin defect.

3. Discussion

Aplasia cutes Congenita (ACC) is a heterogeneous group of rare congenital disorders characterized by a local or widespread, complete or partial absence of different layers of skin at birth, occasionally extending to bone, rarely involving Duramater and meninges [5]. Lesions occurs mostly on vertex of scalp, just lateral to midline, but may also involve face, trunk and extremities. Lesions may be Circular, Oval, Linear or stellate shaped in configuration. They are non-inflammatory, well demarcated in character or may be healed with scarring if intrauterine trauma. Main pathophysiological hypothesis regarding ACC is tension induced disruption of overlying scalp skin during 10-15 weeks of gestation, when rapid brain growth occurs, along with hair direction and patterning.

Causes of ACC includes [6]:

a) Intra-uterine infections (Herpes simplex, Varicella zoster)
b) Teratogenic drugs (e.g. Methimazole, carbimazole, misoprostol, valproate, methotrexate, cocaine, ACE inhibitors, etc)
c) Vascular compromise (specially in a case of stellate or angular lesions)
d) Genetic mutations and chromosome abnormalities (e.g. Mutation in ribosomal GTPase BMS-1 gene, UBA-2 gene, SUMOylation pathway)
e) Intra-uterine trauma

Inheritance of ACC is sporadic or Autosomal dominant.

ACC is classified into 9 types by Frieden based on site of skin defect, associated anomalies, underlying causes and associated syndromes as follows [7] [8]:

a) Type I: Aplasia Cuties Congenita (ACC) of scalp without anomalies.
b) Type II: ACC of scalp associated with Limb anomalies (commonest being hypoplastic or absent distal phalanges of lower limbs)
c) Type III: ACC of scalp associated with Epidermal nevi (Neurological and Ophthalmological abnormalities often present)
d) Type IV: ACC overlying embryological malformations (e.g. Meningomyelocele, Porencephaly, Omphalocele, Spinal dysraphism, Gastroschisis)
e) Type V: ACC associated with fetus papyraceus or placental infarct.
f) Type VI: ACC associated with 'Epidermolysis bullosa'.
g) Type VII: ACC of extremities without blistering (Mostly pretibial lower limb and dorsal aspect of hands or feet)
h) Type VIII: ECC caused by specific teratogens (e.g. Methimazole, carbimazole, Herpes simplex, Varicella zoster)
i) Type IX: ACC associated with malformation syndromes [e.g. Patau syndrome/ Trisomy 18, Goltz syndrome (Focal dermal hypoplasia), Wolf Hirschhorn syndrome (Midline scalp defect), Johnson Blizzard syndrome (Stellate scalp defect), Setlei's syndrome (Bitemporal ACC with abnormal eyelashes), Delleman syndrome/ Occulo-cerebro-cutaneous syndrome, Finaly-Mark Syndrome/ Scalp-ear-nipple syndrome)]

Diagnosis of Aplasia Cuties Congenita (ACC) is based on Clinical examination. USG (Skull), MRI (Brain) And bone survey can be done to rule out associated malformations. Alpha fetoprotein (AFP) and Acetyl-choline esterase are found in amniotic fluid in ACC. But, these tests are neither specific nor sensitive [9].

Histological findings varies according to depth of defect and duration. At birth, ulcerated lesion may show complete absence of skin. After healing, epidermis may appear flattened with proliferation of fibroblasts within connective tissue stroma and absence of adnexal structures [10].

Management of ACC depends upon size and depth of defect, presence of associated abnormalities and risk of complications. For small and localized skin defects (<4 cms in size) without bone involvement, Conservatory line of management with daily cleaning of lesion, proper skin care and application of topical antibiotic ointment until healing is complete is recommended. Systemic antibiotics are given if signs of infection are present. Such small lesions have uneventful recovery with gradual epithelialization and formation of hairless, atrophic scar over 4 weeks [11] [12]. Even small underlying Bony defects of skull (<1 cms), closes spontaneously within 1 year of life [13].

For large skin defects (> 4 cms in size) involving underlying bone/ duramater/ meninges, Surgical intervention may require, which includes Primary wound closure, Local scalp flaps, full or split thickness skin transplantation, Cranial vault reconstruction using bone grafts, etc [14] [15]. Full thickness defects of scalp/ skull/ Duramater have mortality rate of >50% [16].

Complications of ACC includes local infections, hemorrhage, sagittal sinus thrombosis and intracranial infections (e.g. Meningitis) in case of deeper defects [17].

Differential Diagnosis of ACC includes:
Birth trauma by forceps or vacuum pump
Epidermolysis bullosa
Nevus sebaceous
Focal dermal hypoplasia syndrome

4. Conclusion

Aplasia Cutes Congenita: Type I (ACC-I) is a rare condition involving scalp without multiple anomalies. We report a newborn baby with small skin defect (4×5 cms) involving superficial skin layers. Diagnosis of ACC-I was made based on Clinical examination after ruling out intra-cranial malformations by USG (Skull) and bone survey. Baby was managed conservatively with proper skin care and topical antibiotics. Complete recovery was observed after 4 weeks.

Management of ACC depends on size, location and severity of defect, underlying cause and associated anomalies. Superficial lesions are treated Conservatively, while large defects or deeper lesions involving large bone area requires surgical intervention.

5. Future Scope

Outcomes of Aplasia Cutes Congenita can be well studied in case series where a greater number of cases and their outcomes can be compared.

Conflicts of Interest:
The Authors have no conflicts of interest.

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