APLASIO CUTIS CONGENITA: CASE REPORT

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Abstract: Background: Aplasio cutis congenita is a heterogeneous group of disorders characterized by absence of skin layer in a localized or wide spread area at birth. Aplasio cutis congenita is rare anomaly within the newborn population. The incidence is approximately 3 cases in 10000 births. Etiological factors include influence of genetic factors, teratogens, compromised vasculature to the skin, infection, neural tube defects and trauma. Case report: In August 2022 a male infant was born with membranous type of aplasio cutis congenita in the parietal skin area above the large fontanel, round, diameter 10 cm without associated malformations on the rest of the body. The lesion of the head was treated with medical oil, three times a day. After one month the lesion of the scalp was better; the superficial veins partially were obliterated and during this period only one episode of bleeding was recorded. The bleeding was stopped with local compression. After three months the lesion was completely resolved and only very thin blood vessels were noticed. During this period the baby was in good condition and proper weight gain. After discharge of the hospital, heart and brain ultrasounds were performed and they were normal. Follow up consultations with a dermatologist and a geneticist recommended continuous observation of the skin lesion and the development of the baby in general. Conclusion: The treatment of our case demonstrated that beside the very wide spread lesion on the scalp, we prevented: further destruction of the epiderm, bleeding from the very thin and fragile superficial veins, infection and achieved obliteration of superficial veins. The medical oil that was used in this particular case (lesion with diameter more than 3 cm) comprised of: butyrum bovis, skin lipids, pyrus malus fruit extract, pentylene glycol, stellaroides longibracteata leaf extract, tocopheryl acetate, tocopherol. These components have preventing effect of skin inflammation, healing of damaged skin and preservation of skin integrity.
Keywords: newborn, aplasio cutis congenita, medical oil treatment.

1. INTRODUCTION

Aplasio cutis congenita is a heterogeneous group of conditions manifested with nonexistent skin layer in a localized or extensive area at birth. Aplasio cutis congenita is rare anomaly within the newborn population. Over 500 cases have been reported since it was described by Cordon in 1767. The incidence is approximately 3 cases in 10000 births. An influence of genetic factors, teratogens, compromised vasculature to the skin, infection, neural tube defects and trauma are considered as possible reasons for the occurrence of aplasio cutis congenita. The lesions most frequently occur on the scalp vertex with sporadic cases where malformations are observed on the face, body or limbs and they may be presented as circular, oval, linear or stellate patterns. Aplasio cutis congenita may impact only the epidermis and upper dermis or it may protrude further into the subcutaneous tissue, periosteum, skull and dura. Skin anomalies formed within the gestation period may heal before delivery and appear as an atrophic, membranous, bullous or parchment like scar post-delivery. The membranous type of aplasio cutis congenita is most frequently observed. The characteristics of the cutaneous defect (size, location, depth) predetermine whether the assumed treatment will be medical, surgical or combination of the mentioned. The goal of the treatment is to restore physiological and immunological integrity of the skin and to prevent / limit fluid leakage or organ rupture. The prognosis of aplasio cutis congenita is commonly with excellent prospect of recovery. If the defect is small, recovery is with gradual epithelization and formation of a hairless atrophic scar within several weeks. Small bone defects usually close spontaneously during the first year of life. Body and limb defects usually epithelialize and form atrophic scars. Whereas an association of aplasio cutis congenita with further abnormalities is determined, the outcome is dependent on the extent of the manifested anomalies.

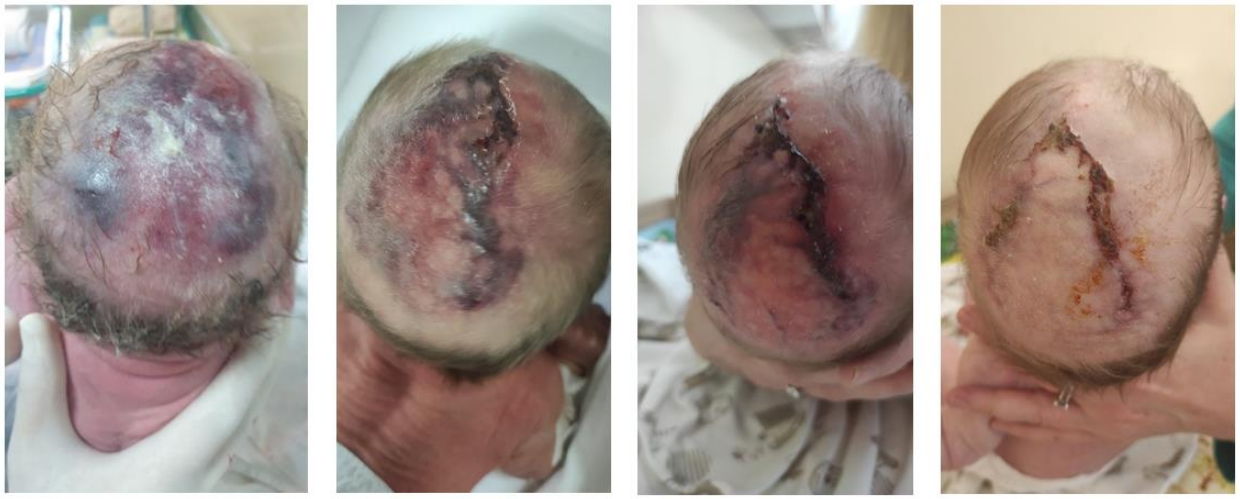
Defects on the scalp and other areas usually heal well with conservative skin care using topical antibiotic ointment. Some defects are prone to complications of hemorrhage and infection. Extensive aplasio cutis congenita is considered to be associated with increased risk of sagital sinus thrombosis.

2. CASE REPORT

The patient was male infant born in 38 gestational age delivered by caesarean section with birth weight 2950 grams and birth length of 48 cm; APGAR score 8/9. He was born from fourth regular pregnancy of a 40 years old mother. A cervical cerclage had been performed in early pregnancy. The pregnancy was gemellar, but one of the gemellus deceased in the late first trimester of pregnancy. A skin defect with approximate size of 10 cm was noticed at birth in the parietal skin area above the large fontanel without associated malformations on the rest of the body. The

defect was with intact epidermis, missing dermis, absence of hair growth and marked superficial veins bulging during crying. Both parietal bones were intact.

Figure 1 - Aplasio Cutis Congenita manifesation at birth; third day after delivery; 2 weeks after delivery and 1 month after delivery



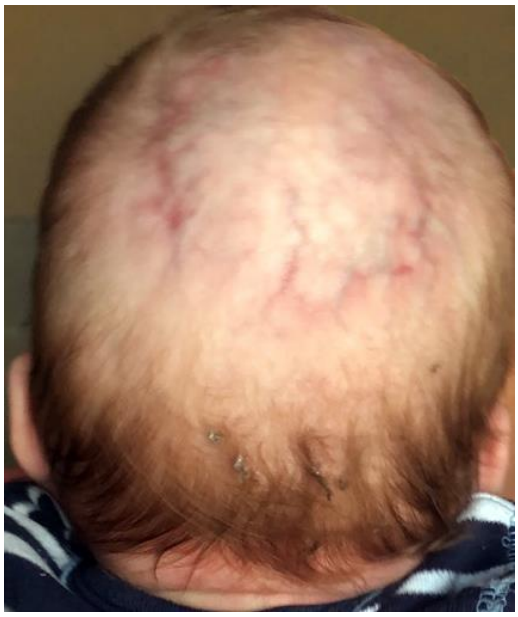
Source – General Hospital Kumanovo, Department of Neonatology; 2022

We performed biochemical analysis of the blood whilst hospitalized in our maternity hospital. The analysis reflected elevated CRP, anemia and hypoglycemia. The baby was treated with first line of AB (sulbactam and clavulonic acid) and aminoglycosides. Early feeding was introduced and the hypoglycemic episodes never repeated. After three days, the laboratory findings were normal with exception of an anemia that was treated with supplementary therapy. The lesion of the head was treated with medical oil three times daily. The medical oil that was used in this particular case (lesion with diameter more than 3 cm) comprised: butyrum bovis, skin lipids, pyrus malus fruit extract, pentylene glycol, stellaroides longibracteata leaf extract, tocopheryl acetate, tocopherol. An ultrasound of abdomen and hearing screening were normal.

After discharge of the hospital, heart and brain ultrasounds were performed and they were normal. Follow up consultations with a dermatologist and a geneticist recommended continuous observation of the skin lesion and the development of the baby in general.

After one month the lesion of the scalp was better; the superficial veins partially were obliterated and during this period only one episode of bleeding was recorded. The bleeding was stopped with local compression. After three months the lesion was completely resolved and only very thin blood vessels were noticed. During this period the baby was in good condition and with proper weight gain.

Figure 2 - Aplasio Cutis Congenita at 3rd month



Source – General Hospital Kumanovo, Department of Neonatology; 2022

3. DISCUSSION

Aplasio cutis congenita is a rare skin malformation characterized by localized absence of the skin predominantly on the scalp, but may also appear on other parts of the body. Aplasio cutis congenita is principally detected by medical stuff within the first day of life, as a single or multiple dispersed lesions on the scalp or other locations: face

(palpebrae superior of the eye), abdomen and limbs. The typical lesions are small 0.5-10 cm; well demarcated with various forms: circular, oval, linear or stellate, membranous or non-membranous.

Cases where the lesion is localized at the vertex (ulcerations on dermis, subcutis and periosteum, skull and dura) are predisposed for possible complications such as: sagittal sinus hemorrhage or thrombosis, focal infection or systemic infection including meningitis.

Etiological factors associated with aplasio cutis congenita are: chromosomal abnormalities, traumatic factors, (the hair collar sign is significant indicator for underlying tensile force during rapid brain growth in that region), amniotic defects, intrauterine problems, thrombotic events and teratogens used in pregnancy (cocaine, methotrexate, angiotensin inhibitor, methimasole, misoprostol, benzodiazepines, valproic acid).

A classification system (Frieden, 1986) for aplasio cutis congenita benefiting treatment alternatives was introduced consisting of 9 groups based on location and presentation of other malformations.

Group 1 - Scalp aplasia cutis congenital without multiple anomalies. Almost 86% of all solitary lesions are localized on the scalp.

Group 2 - Scalp aplasia cutis congenital with limb anomalies. Adams-Oliver Syndrom is a distinct disorder in which distal limb reduction abnormalities are determined to be in association with solitary midline scalp defects; Hypoplastic or absent distal phalanges; Haemangiomas, Cranial AV malformation and supernumerary nipple.

Group 3 - Scalp aplasio cutis congenita with epidermal and sebaceous nevi encompassing the scalp adjacent to the aplasio cutis.

Group 4 - Aplasio cutis congenita overlying deeper embryologic malformations, such as meningomyelocela, porencephaly, cranial stenosis, spinal dysraphisam

Group 5 - Aplasio cutis congenita associated with fetus papyraceous or placental infarct.

Group 6 - Aplasio cutis congenita associated with epidermolysis bullosa.

Group 7 - Aplasio cutis congenita localized to the extremities without EB.

Group 8 - Aplasio cutis congenita due to teratogens. Several cases of aplasio cutis congenita have been related to intrauterine infection with HSV, Varicella zoster virus or exposure to methimasole.

Group 9 - Aplasio cutis congenita associated with malformation syndromes. Aplasio cutis congenita has been observed in various syndromes, including Patau syndrome, Setlies syndrome, Jochanson Blizzard syndrome, Goltz syndrome etc.

Treatment alternatives may include conservative treatment or plastic surgery interventions. Medical oil application as conservative treatment option is viable in cases where the manifestation is present at skin level with coverage of less than 3 cm and without protrusion to the bones. Plastic surgery interventions are recommended in cases where the malformation is significant and with immersion of critical organs.

4. CONCLUSION

The treatment of our case of aplasio cutis congenita demonstrated that beside the very wide spread lesion on the scalp, we prevented: further destruction of the epiderm, bleeding from the very thin and fragile superficial veins, infection and achieved obliteration of superficial veins. The ingredients of the used medical oil have inhibiting effect of skin inflammation, healing of damaged skin and preservation of skin integrity.

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