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Case Report

Paediatrics Section

Aplasia Cutis Congenita and Cutis Marmorata in a Neonate with Down Syndrome: A Rare Case Report

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ABSTRACT

The rare congenital skin malformation known as Aplasia Cutis Congenita (ACC) can be identified by a localised lack of skin. Cutis marmorata, characterised by a transient, net-like, violaceous mottling of the skin, is often seen in preterm or low birth weight infants due to immature vascular responses to cold or stress. Down Syndrome (DS) is linked to uncommon dermatological conditions and has a higher incidence of some common dermatoses. This case presents a rare combination of cutis marmorata and ACC in a neonate diagnosed with DS. The management primarily focused on supportive care, including maintaining a thermoneutral environment, ensuring the baby remained warm, and close monitoring of vital signs. Stabilisation of respiration was achieved through appropriate oxygen support based on the degree of respiratory distress and oxygen requirement, along with prevention and management of potential infections. By doing so, the neonate can be prevented from going into shock, and the need for prolonged ventilation can be avoided. The presence of both cutis marmorata and ACC in this infant may not be coincidental. These skin findings could reflect overlapping effects of an underlying chromosomal abnormality. The favourable outcome reinforces the role of timely diagnosis, tailored supportive therapy, and multidisciplinary coordination, particularly when dealing with multiple congenital anomalies.

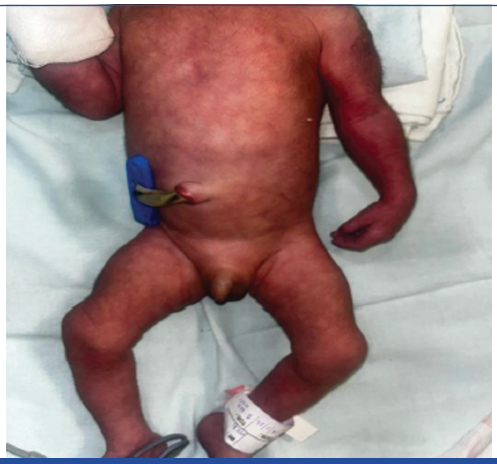
Keywords: Chromosomal anomaly, Congenital scalp defect, Neonatal skin disorder, Trisomy 21 dermatology, Vascular mottling

CASE REPORT

A male child weighing 1.7 kg, was born to a 31-year-old, 3rd gravida mother with one living 10-year-old male child and a history of one abortion nine years back, via lower section caesarean section i/v/o preterm premature rupture of membrane at 35+1 weeks of gestational age. The baby cried immediately after birth and was transferred to Neonatal Intensive Care Unit (NICU) in view of low birth weight and a reddish lesion over the scalp. The mother had no history of diabetes, hypertension, hypothyroidism/hyperthyroidism, blood transfusion, or previous medical or surgical illness. She has a 10-year-old male child with no significant medical or surgical illness. Family history of the baby was not significant.

However, the baby developed respiratory distress with visible subcostal retractions after two hours of admission to NICU, with a heart rate in the range of 130-140 beats per minute, a respiratory rate in the range of 50-60/min, and SpO_2 -96%. His Silverman-Anderson score was 4, and he was transferred to the NICU for further management. He was started on Continuous Positive Airway Pressure (CPAP) with settings of Positive End-Expiratory Pressure 5, Fraction of inspired Oxygen (FiO_2) 30%, and flow of 6 L/min. During initial examination, a 5×2 cm area of hairless, red skin was noted over the scalp, suggestive of aplasia cutis. In addition, his skin showed a mottled, lace-like appearance consistent with cutis marmorata. [Table/Fig-1] shows the lesion on the scalp. Physical examination showed solitary, well-demarcated lesion of 5×2 cm was noted over the scalp on the vertex area, extending backward, blackish red in colour, with a surrounding bald area and no signs of inflammation, and no active bleed, or foul-smelling discharge. The lesion showed central depression with a visible skull defect beneath. Cutis marmorata – a transient, mottled, bluish-red reticular discolouration of the skin that gives a marbled appearance, typically seen over the limbs and trunk, caused by physiological vasomotor instability. The discolouration became more prominent on exposure to cold and fades on warming [Table/Fig-2]. The skin was otherwise normal in texture and temperature with no ulceration or oedema.

[Table/Fig-1]: Well-demarcated, erythematous, hairless patch measuring approximately 5×2 cm located on the vertex of the scalp, consistent with aplasia cutis congenita.



[Table/Fig-2]: Mottled, lace-like appearance of skin as cutis marmorata.

Baseline investigations were sent. His haemoglobin was 18.2 g/dL, total leukocyte count was 15,900/mm^3, platelet count was 1.94×10^5/mm^3, and C-Reactive Protein (CRP) was 0.4 mg/dL. A chest X-ray was performed and bilateral reticulogranular appearance was noted. He was started empirically on intravenous antibiotics-ampicillin 45 mg every 12 hours and gentamycin 8 mg every 36 hours, both administered as slow pushes.

Antenatal records revealed that a quadruple screening test performed earlier in pregnancy was positive for DS. The differential diagnosis for such lesions is broad and includes traumatic causes, such as birth trauma or intrauterine injury, as well as infectious aetiologies like varicella, Herpes Simplex Virus (HSV), and congenital syphilis. Genetic or syndromic associations may also be responsible, including conditions such as Adams-Oliver syndrome, Trisomy 13, and Bart syndrome. Vascular or ischemic causes, particularly amniotic band sequence and complications related to twin-to-twin transfusion, should also be considered. Certain maternal medications, including methimazole and valproate, are known teratogens and may contribute to similar presentations. Finally, structural or neoplastic lesions such as encephalocele or dermoid cysts can mimic this appearance and must be differentiated clinically and radiologically, based on clinical features and the screening result, the diagnosis was consistent with trisomy 21. The final diagnosis was a late preterm male infant with low birth weight, born by caesarean section, with cutis marmorata, ACC, congenital pneumonia, and features of DS.

The baby was given oxygen support with CPAP, despite which the baby continued to have respiratory distress and carbon dioxide retention, with slightly delayed capillary refill time of around four to five seconds, which should be normally less than three seconds. The baby was intubated and was put on mechanical ventilation with settings of FiO_2 of 30%, PEEP of 5, peak pressure of 9, and a respiratory rate of 40. The baby was also given inotropic support for 24 hours. The blood reports were within the normal limits, and culture sensitivity was suggestive of growth of no organism. After approximately 48 hours, an extubation trial was given, and the baby was maintaining saturation and was weaned off from higher to lower oxygen support.

Feeding was introduced via an orogastric tube, and after removing all the oxygen support, two days later, Katori spoon feeding was initiated and was well tolerated by the baby. Neurosonography was normal, and blood cultures remained negative. Antibiotics were continued for a total of seven days. Over the next several days, he improved steadily. He was active, feeding well, and passing urine and stools regularly. Anthropometric measurements at discharge were: weight 1.6 kg, length 44 cm, and head circumference 29 cm. As there were no new concerns, he was shifted to his mother's side in stable condition. Daily cleaning with sterile normal saline and non-adherent dressing using paraffin gauze, hydrocolloid, or hydrogel gauze once daily was advised. A plastic surgery consultation was recommended in view of debridement (removal of dead tissue) and split-/full-thickness skin grafting, and the patient was advised to follow up after six weeks.

As no genetic testing report of the baby was available, and the diagnosis was made based on Quadruple marker testing of the mother, the parents were advised to get a genomic sequencing of the baby. A referral to the plastic surgery team was also made for further evaluation of the scalp lesion. Surgical debridement was planned following the formation of a well-defined line of demarcation. Before discharge, a series of screening evaluations were performed. A neurosonogram revealed no abnormalities, and an ultrasonogram of the abdomen and pelvis was also unremarkable. A 2D echocardiogram demonstrated a 3 mm perimembranous ventricular septal defect. The parents were advised to obtain a thyroid profile and screening for metabolic bone disease after four weeks during follow-up. Clinical photographs from the time of discharge were not available. However, the patient did not return for the scheduled follow-up, and further management could not be pursued.

DISCUSSION

The DS is linked to uncommon dermatological conditions and has a higher incidence of some common dermatoses. Infections and autoimmune disorders are common in DS subjects [1]. The rare congenital skin malformation known as ACC can be identified by a localised lack of skin. The dermis, epidermis, and/or subcutaneous fat may be affected [2]. The co-occurrence of cutis marmorata and ACC in a neonate with DS is an uncommon finding and has been least documented in the literature. This case reveals the need for careful dermatological and systemic examination in neonates with similar features as such dermatological presentations may indicate underlying chromosomal anomalies.

Cutis marmorata, characterised by a transient, net-like, violaceous mottling of the skin, is often seen in preterm or low birth weight infants due to immature vascular responses to cold or stress. While typically benign and self-resolving, its persistence or prominence can sometimes be associated with genetic syndromes, including DS [3,4]. In this case, the appearance of cutis marmorata was seen in the early neonatal period and slowly improved with regulation of temperature and circulatory status.

The ACC is most often seen on the scalp and usually appears as a patch of missing skin present at birth. It can show up on its own or as part of a broader genetic syndrome. Although ACC has been more commonly linked to chromosomal conditions like trisomy 13 and 4p 4p-syndrome, its presence in infants with DS is considered uncommon [5]. In this case, the baby had a well-defined, 3 x 2 cm area on the scalp that was hairless and reddish in appearance, which matched the typical presentation of Type I ACC, where there is no involvement of the underlying bone or blood vessels.

The presence of both cutis marmorata and ACC in this infant may not be coincidental. These skin findings could reflect overlapping effects of an underlying chromosomal abnormality. While the exact cause isn't fully understood, some researchers suggest that issues with blood vessel development and connective tissue structure might be involved [6-8]. In our case, the antenatal quadruple screening had already indicated a high risk for DS, lending further support to a chromosomal basis for these clinical features. In a study by Chen Z et al., size of the scalp defect was 2.5 cm × 3 cm, which was like a cat's paw. The authors concluded that even though the child had a large scalp defect that measured more than 2 cm, conservative treatment still had a significant impact on him. This suggests that for ACC neonates without skull defects, conservative treatment can be the first option, and surgical treatment can be considered when necessary [9].

Previous literature describes ACC most commonly as a solitary scalp defect measuring 0.5-3 cm, similar to the 3×2 cm lesion seen in our patient. Mesrati H et al., (2015) reported that most ACC cases involve the scalp without underlying bone defects, and conservative management is usually effective- consistent with the course in our neonate. Cutis marmorata has also been documented in neonates with chromosomal abnormalities [2]. Bui TNPT et al., (2019) highlighted that persistent or pronounced cutis marmorata may be associated with syndromes such as trisomy 21, matching our patient's presentation [4]. Cases combining both ACC and cutis marmorata are rare, but Devillers ACA et al., (1999) described vascular instability as a possible link between these conditions [6]. More recently, Boia M et al., (2025) reported CMTC cases with overlapping vascular and connective tissue abnormalities, supporting the possibility of shared pathogenic mechanisms. Compared with these reports, our case aligns with the phenotype described in infants with underlying chromosomal anomalies, particularly DS, where vascular dysregulation and structural skin defects may co-exist [10].

The recent case series by Boia M et al., (2025) emphasised that Cutis Marmorata Telangiectatica Congenita (CMTC) and related vascular anomalies may present alongside other congenital skin defects, including aplasia cutis. Their review highlighted variable severity, asymmetry of lesions, and associations with limb anomalies or chromosomal conditions. Although our patient did not exhibit the classic asymmetric telangiectatic pattern of CMTC, the presence of persistent mottling and ACC together resembles the spectrum of vascular dysregulation described in their series. Boia M et al., also stressed the importance of thorough systemic evaluation in neonates with combined vascular and structural skin defects, which aligns with the comprehensive screening our patient underwent [10].

CONCLUSION(S)

This case presents a rare combination of cutis marmorata and ACC in a neonate diagnosed with DS. Each of these conditions can occur independently; their presence together, especially in the context of a chromosomal disorder, signifies the importance of detailed neonatal examination, including dermatological findings. Such features may serve as early signs of underlying syndromic diagnoses. The management focused mainly on supportive care, including stabilising the respiration and addressing the risk of infection. The skin lesions were handled conservatively with gentle monitoring and careful hygiene, without the need for invasive treatment. Early recognition can facilitate timely investigations and multidisciplinary care, improving neonatal outcomes.

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