

Severe telangiectasia in a neonate Adams-Oliver syndrome

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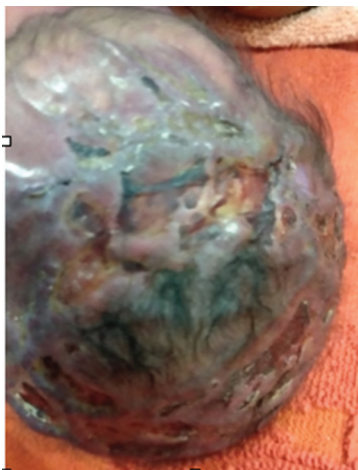
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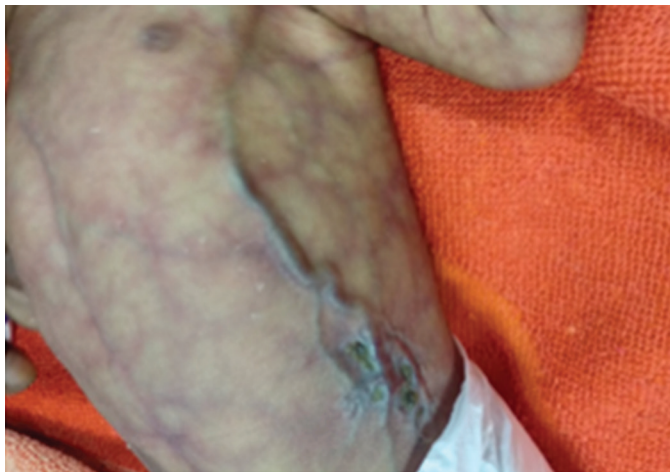
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Baby X was full-term, female, and appropriate for gestational age with a birth weight of 2.5 kg. The baby was born to G3P3L2D1 mother under third-degree consanguineous marriage. The previous male baby (second in birth order) expired at 6 months of age due to sepsis. The antenatal history was non-significant except mild oligohydramnios in the third trimester. The baby was born by a lower segment cesarean section (LSCS) in view of the previous LSCS. The baby cried immediately after birth and accepted breastfeeds within 3 h of life. The baby presented to our outpatient department on day 1 of life with complaints of prominent veins over the scalp extending into the forehead and abdomen.

On examination, aplasia of the skin and crusting lesions was noted over the scalp, and the baby had severe telangiectasia all over the body. The baby had retrognathia, and no other external or internal anomalies were observed. Internal anomalies were excluded by infantogram, skull X-ray, neurosonogram, ultrasound abdomen, echocardiography, and computed tomography brain. The baby's clinical features were similar to Adams-Oliver syndrome (AOS). The clinical criteria for the syndrome include the major and minor criteria. The major criteria features are aplasia cutis, congenital terminal transverse limb defects, and family history of AOS. The minor criteria include cutis marmorata telangiectatica congenita (CMTC), congenital cardiac defects, and vascular anomalies. The presence of two major criteria are sufficient for diagnosis, while the combination of one major and one minor criteria is denoted a high likelihood of AOS.

A wide phenotypic variability is the characteristic of this syndrome. Our case had one major and two minor criteria. The incidence of AOS is about 1 in 225,000 live births. AOS is classified as type 2 aplasia cutis congenital (ACC) under Frieden's classification. In 1945, Adam and Oliver described the first case of AOS which was an autosomal dominant form. The possible mechanisms of pathogenesis are vascular impairment, amniotic bands formation during embryogenesis, and aberrant morphogenesis. More recently, abnormal pericyte recruitment to the blood vessels was postulated as a possible cause. There are multiple forms as autosomal dominant, autosomal recessive, and sporadic form, and the most common is the sporadic form.





The autosomal recessive form usually involves the DOCK6 and EOGT genes. In our case, there was no family history of AOS, so it was most likely a sporadic form. About 20% of infants with AOS may have an associated heart defect.

Diagnosis is based on clinical features, family history, and imaging. It should be differentiated from isolated hereditary CMTC, Bart type epidermolysis bullosa dystrophica, focal dermal hypoplasia, trisomy 13 syndrome, Johanson–Blizzard syndrome, ACC, and amniotic band syndrome. These disorders usually have other physical features that may differentiate them from AOS. Isolated hereditary CMTC does not have scalp defects. The focal dermal hypoplasia is characterized by linear streaks of atrophied skin, telangiectasia with soft, fatty nodules, and digital malformations. The treatment focuses mainly on symptom management such as skin grafting, flap rotation, and cranial surgery, using of prostheses for limb malformations and lifestyle modifications such as wearing of helmets to protect the skull. The long-term prognosis varies depending on the specific signs and symptoms and their severity. Cutaneous telangiectasia usually disappears with age. It should be noted that in the absence of major abnormalities, those with this condition should have a normal lifespan.

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