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Case Report

Adams-Oliver syndrome: A case report

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Abstract Adams-Oliver syndrome is a rare disease, which presents with cutaneous, cardiac and skeletal defects. We, herein, describe a case of 1-month-old girl with aplasia cutis, cutis marmorata telangiectatica and terminal transverse limb reduction defects with a positive HCV serology. The patient is put on regular follow-up.

Key words
Adams-Oliver syndrome, aplasia cutis congenita, cutis marmorata telangiectatica, limb reduction defects.

Introduction

Adams-Oliver syndrome (AOS) was first described by Forrest H. Adams and CP Oliver in 1945.[1-3] It is a clinical syndrome associated with aplasia cutis congenita (ACC), which is classified into nine groups according to location, hereditary mode and associated anomalies. AOS belongs to ACC group II and presents with variable clinical manifestations including limbs reduction, aplasia cutis congenita, cutis marmorata and cardiac anomalies. The incidence is 0.44 per 100000 live births with no difference in gender distribution according to a Spanish study.[4] Multiple hereditary patterns are reported including autosomal dominant, recessive and some sporadic cases. MSX1, CART1, P63 (P73L), RUNX2, and HOXD13 genes were sequenced in 9 previously reported families, but no disease-causing mutations were found.[5]

Case Report

A female child of 1-month age, born to consanguineous parents presented to our department with history of a diamond-shaped scalp defect on the vertex since birth (**Figure 1**). There was shortening of digits of both hands and feet (**Figure 2**). She also had cutis marmorata telangiectatica (**Figure 3**). Our patient was the youngest of the three siblings. One sister was megalocephalic but none had any of the above mentioned defects. Mother was HCV positive with history of taking hakeem medication for its treatment for three days during pregnancy in third trimester. The baby was delivered at home without any complications. She was unvaccinated. Her family history revealed that her maternal great grandmother had similar digital defects but no other feature of the disease. The baseline investigations of the patient were normal except that she was also HCV positive, confirmed by ELISA. Her chest X-ray and abdominal ultrasound were normal. Her echocardiography did not reveal any abnormality. X-rays of hands and feet showed absence of distal phalanges (**Figure 5** and **6**). The patient was given topical antibiotic to cover the cutaneous infection over the scalp. She was referred to plastic surgery and neurosurgery departments. A surgery is planned by the plastic surgery department to cover aplasia cutis. As there was no CNS involvement, no surgical intervention was planned.

Our patient was HCV positive with normal liver function tests, she would be regularly

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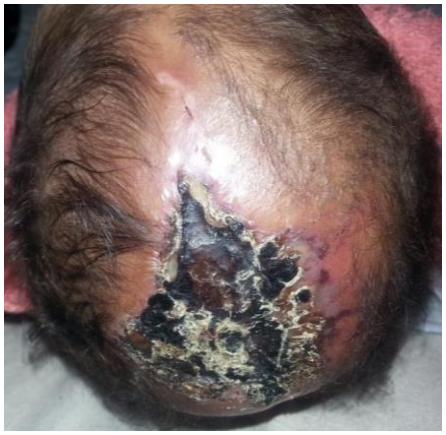

Figure 1 Atrophic plaque on vertex since birth.

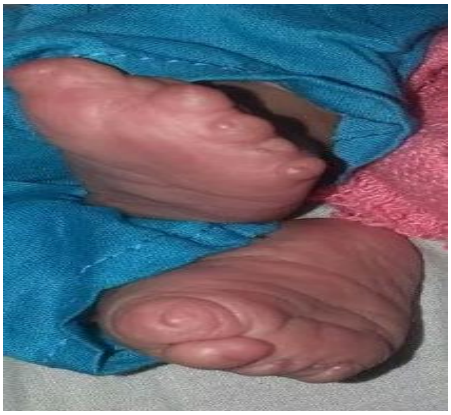

Figure 2 Terminal transverse limb defects including hypoplasia of fingers of right hand and both feet.

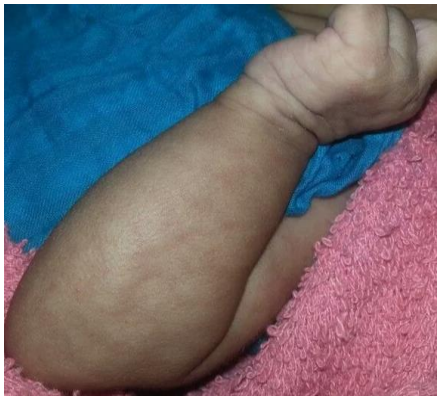

Figure 3 Reticulate erythema on trunk and limbs.

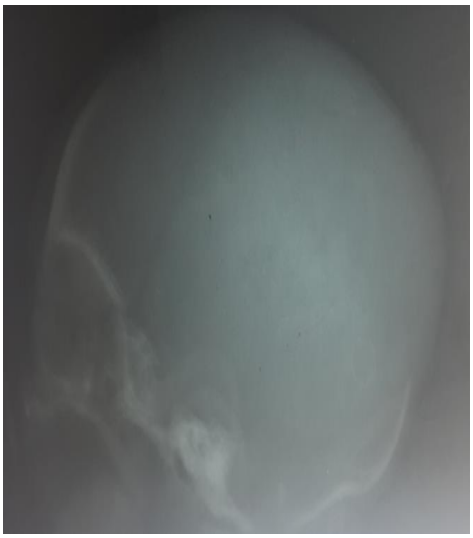

Figure 4 X-ray skull showing absent parietal bone.

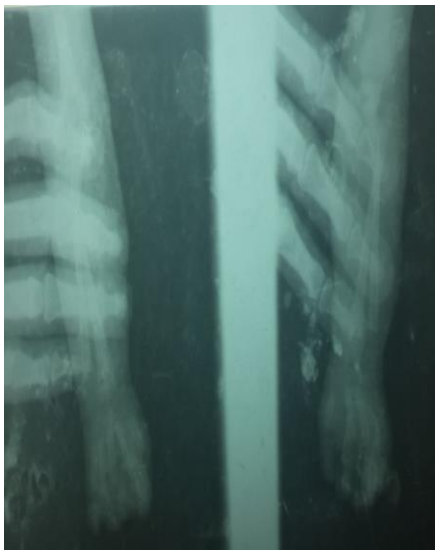

Figure 5 X-ray upper limbs showing absent distal phalanges.

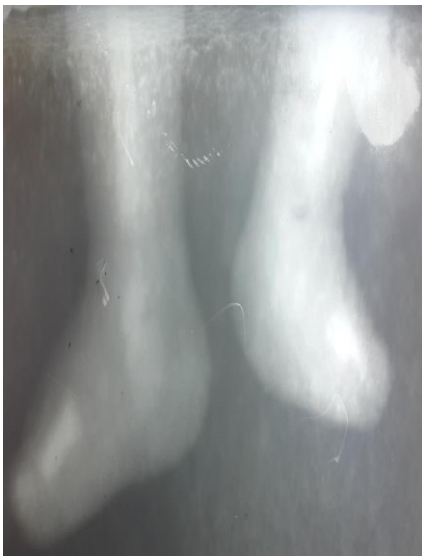

Figure 6 Lower limb radiograph showing absent distal phalanges.

followed up. Her digital defects did not cause functional impairment so were not be treated.

Discussion

Dr. Adams and Dr. Oliver for the first time described 8 cases in a family with aplasia cutis congenita and distal limb reduction defects in 1945.[1-3] Different cases with various physical anomalies have been reported since then nationally and internationally.[4-6]

Different theories have been proposed to explain the pathogenesis of the disease. Exact etiology of the disease is unknown. A possible explanation could be disruption of embryonic blood vessels which become fragile due to mutation in a gene involved in vasculogenesis.[7,8] Other hypotheses include rupture of amniotic bands,[9] teratogenic factors, intrauterine infections like chickenpox, zoster or herpes simplex, fetal exposure to cocaine, heroin, alcohol or antithyroid drugs, oligohydramnios and external compressions.[10] In our patient a possible family history of similar disease in great grandmother is present which suggests autosomal recessive mode of inheritance. Other factors are hakeem medications taken by the mother that could have been teratogenic or the transplacental transfer of hepatitis C virus infection.

Various clinical presentations of AOS include oligohydramnios, cutis marmorata, upper limb micromelia and brachypodia, acrania, microcephaly, palatine or auricular malformations, intracranial calcifications, hydrocephaly, arhinencephaly, spina bifida, epilepsy, mental retardation, anatomic bronchial anomalies, renal abnormalities, and cardiovascular anomalies such as bicuspid aortic valve, atrial septal defect, Shone's complex, aortic valve stenosis, hypoplastic left heart syndrome, tetralogy of Fallot, double outlet right ventricle, portal hypertension and pulmonary hypertension.[11,12] Our patient had cutis marmorata telangiectatica, aplasia cutis congenita, and terminal transverse limb reduction defects. Her abdominal ultrasound, echocardiography and CT-scan of brain were normal and did not show any systemic abnormality. X-rays of limbs were consistent with the limb defects showing absence of digits. She had normal blood tests except that she was HCV positive which has not been reported anywhere in various articles on Adams-Oliver syndrome.

Conclusion

We are presenting this case as it is a very rare disease and not many cases have been reported and our case is the first one to be reported with positive serology for hepatitis C virus. AOS requires a multidisciplinary approach to achieve better management and control and our patient will be investigated accordingly in future.

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