INTRODUCTION

Aplasia cutis congenita implies failure of skin development at birth. Adams-Oliver syndrome is characterized by congenital midline scalp defects (aplasia cutis) in association with asymmetrical terminal transverse limb reduction anomalies of variable severity. Cutis marmorata telangiectatica congenita has been associated with 12% of cases. It was described initially in 1945 by Adams and Oliver. It is an autosomal dominant trait with variable penetrance. Limited forms of the syndromes with only persistent cutis marmorata and aplasia cutis in the absence of limb reduction defects have also been described. Since its original description, many associations have been highlighted involving the central nervous system, cardiovascular system, genitourinary system and lungs. These internal anomalies may be severe and determine the prognosis of individual patients. The exact pathogenesis remains unclear.

CASE REPORT

A male child of Pakistani origin was the third of three children born to consanguineous healthy parents at 40 weeks of gestation. He had a birth weight of 2600 gm. At birth, he was noted to have a small patch about 2 cm in diameter of aplasia cutis (Figure 1). He had symmetric shortening of the distal phalanges of both thumbs and all the fingers of both hands and both big toes (Figure 2). The nails were hypoplastic except for the thumbs and big toes where the nails were entirely absent (Figure 3). He had a short prepuce; both testes had descended. He did not have cutis marmorata telangiectatica congenita. His chest X-ray and ultrasound of abdomen were normal. X-ray of the feet showed absence of the distal and middle phalanges of the big toe and absence of distal phalanges of the rest of the digits. X-ray of the hands showed absence of the distal phalanx in all the digits. Echocardiography and MRI of brain were normal. His parents and siblings were normal. His family history revealed a paternal first cousin who had been born to consanguineous parents, with similar problems of short digits and a scalp defect. This child had later died at the age of 3 months of pneumonia.

The newborn was placed on regular follow-up to detect any associated vascular, cardiac or central nervous systems anomaly that may develop or present later in life. Since the patient had no systemic involvement, a normal life expectancy is expected. The patch of aplasia cutis will be covered once the child scalp hair grow in length. The short digits will remain a cosmetic disability but the child is not expected to be functionally compromised.

DISCUSSION

In 1945, Dr. Adams and Dr. Olivers described 8 members of a family with Aplasia Cutis Congenita (ACC) and distal transverse limb reduction defects. Since then, Adams-Oliver syndrome (AOS) has been described with physical anomalies ranging from minor cases of aplasia cutis congenita to extensive areas of denuded skin at birth in association with skull defects, reduction limb defects and cutis marmorata telangiectatica congenita. An autosomal dominant inheritance has been proposed with highly variable penetrance and expression. Other mechanisms of inheritance, autosomal recessive and new mutations have been described. Since the parents of this patient were normal and consanguineous and there was a history of similar disease in a first cousin the inheritance is most likely to be autosomal recessive in the patient.
Adams-Oliver syndrome

The most frequent limb malformations are syndactyly (bony/cutaneous), brachydactyly, poly and oligo-dactyly and hypoplastic finger and toe nails.4 This patient had symmetrical shortening of the distal digits of both hands and feet with hypoplastic nails. X-rays showed absence of the distal and middle phalanges of the big toe and absence of the distal phalanges of all the other toes. Similarly, the distal phalanges of all digits of the hand were absent on X-ray.

More recently, the association of vascular and cardiac malformations has been recognized. Congenital heart defects, ventricular and atrial septal defects, Tetrology of Fallot, coarctation of aorta and bicuspid aortic valve have been described in 15 of 112 cases of AOS.5 Pulmonary vascular abnormalities that have been reported include pulmonary vein stenosis and pulmonary hypertension or severe infantile unexplained pulmonary hypertension. Abnormal pulmonary vasculature with persistence of primitive systemic and pulmonary artery vascular connection and supra systemic pulmonary artery pressures have been reported in a patient with limb reduction defects in AOS in the absence of aplasia cutis.6 Another report describes a pulmonary arterio-venous malformation in two members of a family suffering from AOS.6 This patient's X-ray chest and echocardiography was normal. Other invasive investigations were not carried out.

CNS malformations have also been described although our patient had no evidence of any problem on MRI of brain. A child has been reported with hypoplastic corpus callosum and focial findings in the periventricular white matter.2 Another report describes the presence of microcephaly,2 epilepsy,7 mental retardation,2 arthrogenic,2 hydrocephaly,2 encephalocele,2 acrania,2 ventriculomegaly,2 cerebral atrophy,2 thalamic and periventricular calcifications and polymicrogyria.2 Another report described association of hepatoportal sclerosis in three unrelated children with AOS.4 Cryptorchidism and renal anomalies have been described.7 Bilateral congenital cataract has also been reported.8

To identify the disease-causing gene, the MSX1, CART1, P63 (P73L), RUNX2, and HOXD13 genes were sequenced in 9 previously reported families, but no disease-causing mutations were found.7

It remains somewhat unclear as to whether the prognosis of AOS without lethal anomalies alters the lifespan. However, it is very likely that limited forms of the disease with only skin and skeletal abnormalities are likely to be associated with a better prognosis and no reduction in lifespan as compared to those patients with systemic involvement.10

REFERENCES