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Holt Oram Syndrome with PDA

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***Abstract**: Holt –oram syndrome is clinically characterised by morphological abnormalities of upper limbs and congenital heart defects.Although the disease is congenital,the diagnosis may only be made later in life.We describe the case of one year female child with hypoplasia of upper limbs since birth and congenital cardiac defect,patent ductus arteriosus which is not common in this syndrome.*
***Key Words**: Congenital heart defects ,hypoplasia of upper limbs.*

I. Introduction

This syndrome was defined for the first time in 1960.1 It includes a set of cardiac disorders and thumb aplasia and/or hypoplasia which may arise in variety forms and with relative frequency of 1:100,000.3, 4, 12–14. HOS syndrome is an inherited autosomal dominant trait with high rate of interference and different variants 1– 5 The position of gene is on chromosome 12 (12q24.1) and it is caused by a mutation in the *TBX5* gene located on chromosome 12, but sporadic cases have also been reported. Although the disease is congenital, the diagnosis may only be made later in life. Holt-Oram syndrome is clinically characterized by morphological abnormalities of the upper limbs and congenital cardiac defects. Upper limb involvement as aplasia, hypoplasia, fusion or anomalous development of carpal, radial and thenar bones. Cardiac anomalies are present in approximately 75% of patients mostly atrial or ventricular septal defect and conduction defect. We present a classical case of Holt oram syndrome with Patent Ductus Arteriosus which is a rare finding.

II. Case report

A 1y/f child came with complaints of loose stool and fever for 3 days and cough and cold for 1 day.On examination the child had a weight of 4.65 kg against 10 kg (percentile in WHO growth chart).The child has normal developmental milestones. The vital were normal. On head to toe examination there was bilateral absence of thumb with a hanging skin tag on the left hand and complete absence of thumb on the right hand. Both the forearms were curved inwards (radial bowing) with hypoplasia of both radii. She was unable to pronate or supinate her forearms. On systemic examination there was a continuous murmur over the pulmonary area. Other systems were normal.

The investigations confirmed the clinical findings. The complete blood count was normal with normal platelet count of 2.12lakh/ul. The echocardiography finding showed acynotic congenital heart defect with a small PDA of 3mm, with no pulmonary stenosis and Qp:Qs= 2:1 The x-ray of forearms confirmed bilateral hypoplasia of radii with absent thumb.

There was no history of similar findings in the family, thus it is a sporadic mutation. Many cases with Atrial and Ventricular septal defect have been reported but our case has a rare association of Patent ductus arteriosus.

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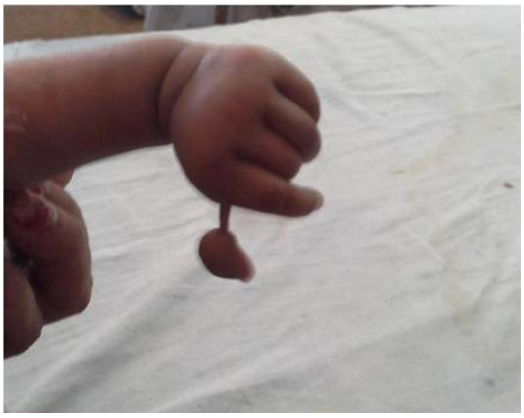

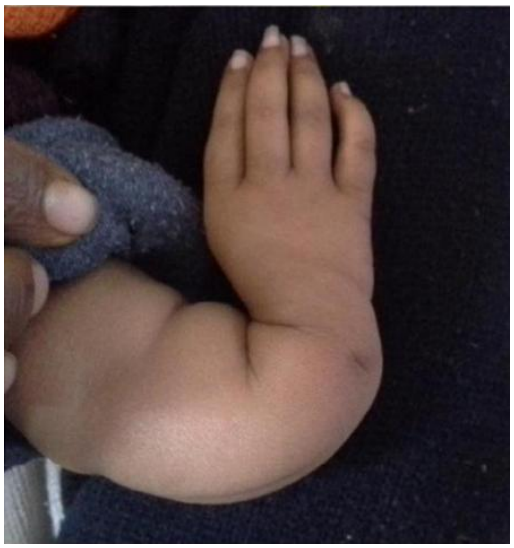

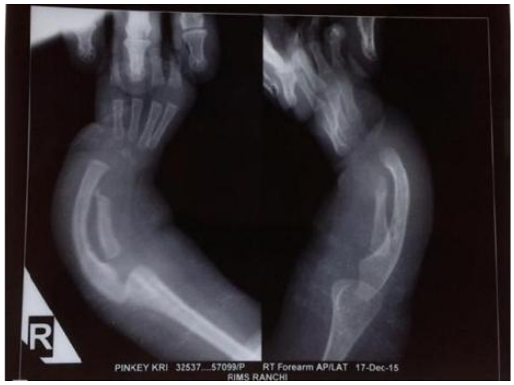
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