



Holt Oram Revisited

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Clinical Image

24-year male presented with history of repeated chest infections since childhood. On examination there was right upper limb abnormality in the form of foreshortened arm, underdeveloped small sized thumb, Metacarpals and right radius. Echocardiography showed inlet ventricular septal defect (VSD) size 7 mm with left to right shunt. Diagnosis of Holt Oram syndrome (HOS) was made which is clinically characterized by congenital cardiac defects and morphological abnormalities of the upper limbs [1]. Patient was advised VSD closure. Holt oram syndrome also known as the atriodigital dysplasia syndrome is a rare autosomal dominant genetic disorder with most frequent mutation in TBX5 gene located on chromosome 12q24.1 [2]. Defective development of the embryonic radial axis (e.g. hypoplasia, aplasia, fusion, or other anomalous development) results in a wide spectrum of phenotypes, including thumb may be absent, underdeveloped or triphalangeal [3]. There may be malformations of the metacarpals, hypoplastic or absent radii, ulna or humerus (Figure 1a and 1b). Abnormalities may be unilateral or bilateral and asymmetric. Majority of patients also have cardiac malformations which include atrial septal defect mostly secundum type, VSD, abnormal isomerism and anomalous pulmonary venous return. Rare but other cardiac associations include pulmonary stenosis and mitral valve prolapse [4].

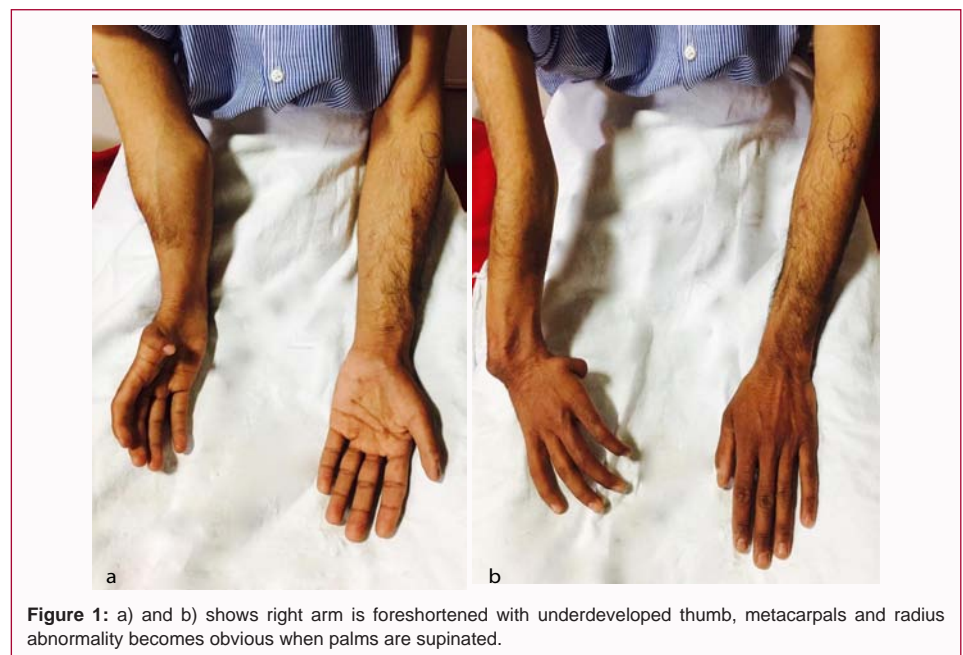


Figure 1: a) and b) shows right arm is foreshortened with underdeveloped thumb, metacarpals and radius abnormality becomes obvious when palms are supinated.

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