Short Communication

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Holt Oram Revisited

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Introduction:

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 and there was difficulty in apposition of thumb to fingertips. Metacarpals and right radius was underdeveloped. On ECG, left axis deviation, broad notched P wave with biventricular hypertrophy was present. On chest radiography there was cardiomegaly (CT ratio >0.58), with left atrium (LA), left ventricle (LV) and right ventricle (RV) enlargement. Echocardiography showed inlet ventricular septal defect (VSD) size 7mm with left to right shunt with LA/LV enlargement with normal LV and RV functions. Diagnosis of Holt Oram syndrome was made. Patient was advised VSD closure. Holt Oram syndrome was first described in 1960 and is also known as the atriodigital dysplasia syndrome. It is a rare autosomal dominant genetic disorder. Several mutations have been described, but the most frequent is in the TBX5 gene of the T-box

complex, located on chromosome 12q24.1

It is clinically characterized by congenital cardiac defects and morphological abnormalities of the upper limbs. 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. There may be malformations of the metacarpals, hypoplastic or absent radii, ulna or humerus and scapulae may be absent or abnormal. Abnormalities may be unilateral or bilateral and asymmetric. Majority of patients also have cardiac malformations and almost every type of cardiac anomaly has been reported, either singly or as part of a group of multiple defects. The most common are atrial septal defect mostly secundum type, VSD, abnormal isomerism and anomalous pulmonary venous return. Rhythm abnormalities include atrioventricular block, atrial fibrillation and sudden death from heart block.

Fig 1

Fig 2