WCC 2016-109 : Familial Holt-Oram Syndrome- CH Samba siva rao, K.Suneetha, O.Adikesava naidu, Y.V.Sbbareddy

Introduction : Holt-Oram syndrome is the prototype of hand heart syndromes. It is characterized by abnormalities of upper limb and congenital cardiac defects. It is a rare genetic disorder seen in 1 in 100,000 children born, with autosomal dominant inheritance. Despite genetic heterogeneity, it is most frequently caused by mutation in TBX5 gene located on chromosome 12.

CASE REPORT:A 18 years female came with complaints of shortness of breath on exertion since 1 year. On physical examination she had pectus excavatum and bilateral hand abnormality and clinodactyly.. Cardiovascular examination showed grade 2 left parasternal heave, wide fixed split second heart sound and loud pulmonic sound. Her ECG showed right axis deviation, RBBB. 2D Echo of showed RA and RV enlargement. TEE showed 1.8 cm ostium secundum atrial septal defect with left to right shunt. On history evaluation she is born out of non-consanguinous marriage with five siblings. Her elder sister died at the age of 16 during the surgery for ASD correction. Other four siblings were screened. Among them one male of age 34 was diagnosed to have upper limb deformity and ostium secundum ASD, with cyanosis and clubbing. X ray of her sibling upper limbs showed triphalangeal thumb and carpal bone deformity 2D-echo of him showed ostium secundum atrial septal defect of size 4.4 cm with bidirectional shunt. RA and RV dilated.This was confirmed by TEE. Patient referred for surgery as not suitable for device closure.

CONCLUSION: Holt Oram Syndrome to be suspected in siblings of patient with typical findings, even in absence of trpihalangeal thumb. Mere presence of carpal bone abnormalities is considered as phenotype of Holt Oram Syndrome .