

Picture stories

Holt Oram syndrome

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A 22 days old baby, a product of non consanguineous marriage, fourth in birth order presented with fever and respiratory distress of 3 days duration. The left upper limb had absent thumb and ulnar side deviated forearm while the right upper limb had a partially attached thumb (Figure 1).



Figure 1: Clinical photograph of baby

On examination, baby had tachypnoea and a pansystolic murmur in parasternal area. On radiographic examination left upper limb had absent radius, first metacarpal and phalangeal bones of thumb. Right upper limb also had absent first metacarpal and no phalangeal bone in partially attached thumb (Figure 2). There was no abnormal rhythm or conduction defect on electrocardiography. Echocardiography showed ASD of 5 mm size, VSD of 5 mm size with mild pulmonary hypertension. Because of characteristic upper limb and cardiac anomalies diagnosis of Holt Oram syndrome was considered and baby managed conservatively.

Holt Oram syndrome, also known as hand heart syndrome or atridigital dysplasia is an autosomal

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Figure 2: Radiograph of upper limbs of baby

dominant disorder, characterized by upper limb and cardiac abnormalities¹. It is caused by mutation in TBX5 gene located on 12q24.1 chromosome¹. The prevalence is estimated to be 0.95 per 100,000 total births with 85% of cases due to new mutations¹. The clinical manifestations vary from sub-clinical radiographic findings to overt life threatening disease. Upper limb defects may be unilateral or bilateral involving structures of the embryonic radial ray causing aplasia, hypoplasia, fusion and anomalous development of the radial, carpal and thenar bones¹. Defects include triphalangeal or absent thumbs, foreshortened arms and phocomelia¹. Cardiac abnormalities, present in more than 85% of cases are either structural e.g. ASD, VSD, or functional e.g. bradycardia, AV block. Other defects in Holt Oram syndrome include vertebral, anal, tracheo-oesophageal, and renal defects¹.

References

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