

## **Case Report**

### **Holt–Oram Syndrome: A Case Report**

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**Abstract:** Holt-Oram syndrome or heart–hand syndrome is a multisystem congenital disorder, characterized by distinctive malformations of bones of the thumb and forearms and associated abnormalities of the heart. Here we are reporting two cases of this syndrome in a family.

**Keywords:** Holt-Oram syndrome, Dyspnea, Palpitation, Family History

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#### **INTRODUCTION**

Holt-Oram syndrome or heart–hand syndrome is a multisystem congenital disorder[1]. The name was given by Dr. Mary Holt and Dr. Samuel Oram in 1960 [2]. Since then, about a few hundred cases have been reported worldwide. It is characterized by distinctive malformations of bones of the thumb and forearms and associated abnormalities of the heart. We report 2 such cases of the syndrome in a family.

#### **CASE REPORT**

A 30 year old Hindu female presented in the outdoor with complaints of excessive breathlessness and palpitation since 4 days. The patient gave history of dyspnea and palpitation since past 8 years. She had been 1<sup>st</sup> hospitalised 8 years ago, when she was diagnosed as congestive cardiomyopathy with ASD. Since then she had been admitted again when she had atrial fibrillation and atrial block. She had been taking digitalis and a diuretic since then. She had stopped her medication since the past 10 days when she reported to us.

On examination: The patient was averagely built and poorly nourished. Her vital parameters were-pulse - 104/mt, BP-110/70 mmHg. Resp. rate-30/mt. She had polydactly (5 fingers) in her left hand. Both thumbs were triphalangeal. The forearms were shortened. There was restriction of supination and pronation in both forearms. Both lower limbs were normal. There was no significant lymphadenopathy, splenomegaly, cyanosis or edema feet. Liver was palpable, 3 cms below the right costal margin.firm,tender with smooth margins. CVS examination showed tachycardia and engorged neck veins. Precordial pulsations and thrill was present. 1<sup>st</sup> heart sound was loud, 2<sup>nd</sup> heart sound was split.

There was an ejection systolic murmur grade 4/6,heard best in the 3<sup>rd</sup> and 4<sup>th</sup> intercostal space along the left sternal border. The patient was orthopneic but there were no basal crepts. Bilateral air entry was equal. She was hospitalised and put on digitalis, diuretic and supportive treatment.

Her investigations revealed a normal hemogram. X-ray chest showed enlarged right ventricle and atria. Pulmonary vascularity was increased. ECG showed volume overload of right ventricle and axis deviation.

Echocardiography showed evidence of congestive heart disease, large ASD, prolapse of anterior mitral leaflet, mild MR,PAH and mild TR.

The patient was advised corrective surgery for the cardiac lesion but she refused.

#### **Family History**

Further enquiries revealed that the patient's mother had similar thumb deformity with polydactly, had been often ill, with breathlessness and recurrent respiratory tract infections and had died at the age of 35 years. None of her 2 siblings had similar limb deformity. The patient has 2 children who were examined. The male child aged 10 years was normal. The female child was 5 years old, had sloping shoulders, absence of thumb in both hands, with loss of supination and pronation of the forearm. The child was asymptomatic. There was no history of recurrent fever, cough or other illness. Her height was 105 cms (normal-107+/-13 cms), weight was 16 kgms (normal-18+/-3.6 kgms).CVS examination revealed loud 1<sup>st</sup> heart sound and an ejection systolic murmur which was best heard in the middle and lower border of left side of sternum which on

echocardiography was diagnosed as ASD. The parents were informed about her lesion and were counselled to bring her for regular checkups.

Based on the typical limb deformities and associated cardiac lesions, a diagnosis of Holt-Oram Syndrome was made.



Fig. 1: Picture of Child and Mother with Holt-Oram Syndrome

## DISCUSSION

Holt-Oram Syndrome, also called as the Heart-Hand Syndrome or Atrio-Digital Dysplasia is a rare autosomal dominant disorder [3]. The symptoms and physical findings associated with Holt-Oram Syndrome may vary greatly, the severity being greater in succeeding generation.

There are malformations in the upper limb with associated cardiac lesions. The thumb may be absent, underdeveloped or triphalangeal [4-6]. There may be malformations of the metacarpals, hypoplastic or absent radii, ulna or humerus [13]. The scapulae may be absent or abnormal. Shoulders may be narrow or sloping.

The cardiac disorder may be atrial or ventricular septal defects, PDA, endocardial cushion defects, hypoplasia of left ventricle or conduction disturbances (1<sup>st</sup> degree heart blocks) [7-9]. There may be associated renal anomalies [10]. Prognosis depends on the severity of cardiac and orthopaedic lesions.

Differential diagnosis must be made from Fanconi Anaemia, Vactrel associations and radial ray choanal atresia [11].

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