Heart-Hand Syndrome with Sub-aortic Membrane - A Case Report

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Abstract:
Background: Congenital cardiac and upper-limb malformations frequently occur in association and are classified as heart-hand syndromes. The classical form of Holt-Oram syndrome is an autosomal dominant disorder characterized by congenital cardiac and forelimb anomalies, first described by Holt Oram in a family with atrial septal defect and congenital anomalies of the thumbs in 1960. Case report: A 28-year old male with anomalies in both upper limbs was admitted with history of NYHA Class III dyspnea and fever of 1 week duration. There was no family history of heart disease. On examination, was short-statured and had skeletal anomalies in both his upper limbs. Cardiovascular examination revealed grade 4 pansystolic murmur in the left parasternal area and bilateral basal rales in both lung fields. Skeletal X-rays showed hypoplastic thumb in left hand and club radius and ulna with hypoplastic thumb on the right side. Transthoracic Echocardiogram (TTE) revealed a small perimembranous ventricular septal defect (VSD) with a sub-aortic membrane and mild aortic regurgitation. A vegetation was seen attached to the aortic side of the right coronary cusp. His blood cultures grew Corynebacterium species which was treated with parenteral antimicrobial therapy for 4-weeks after which he was referred for surgical correction of his cardiac defects. Conclusion: The heterogeneity of presentation of Heart-hand syndromes is well known. Our patient, probably a sporadic mutant had ventricular septal defect, sub-aortic membrane, aortic regurgitation and infective endocarditis of the aortic valve as cardiac defects. Sub-aortic membrane has hitherto never been reported in association with Holt-Oram syndrome in literature. Keyword: Heart Hand Syndrome subaortic membrane

Background: Congenital cardiac and upper-limb malformations frequently occur in association and are classified as heart-hand syndromes. The classical form of Holt-Oram syndrome is an autosomal dominant disorder characterized by congenital cardiac and forelimb anomalies. It is caused by
mutations of TBX5 gene a member of the T-box family. Holt &Oram first described this syndrome in a family with atrial septal defect and congenital anomalies of the thumbs in 1960.

Case report A 28-year old male with anomalies in both upper limbs was referred to the Department of Cardiology, with history of NYHA Class III dyspnea and fever of 1 week duration There was no family history of heart disease. On examination was short-statured and had skeletal anomalies in both his upper limbs. Cardiovascular examination revealed grade 4 pansystolic murmur in the left parasternal area and bilateral basal rales in both lung fields. Chest X-ray showed cardiomegaly with interstitial edema. Skeletal X-rays showed hypoplastic thumb in left hand and club radius and ulna with hypoplastic thumb on the right side(Fig 1). Transthoracic Echocardiogram (TTE) revealed a small perimembranous ventricular septal defect (VSD)with a sub-aortic membrane and mild aortic regurgitation (Fig 2). A vegetation was seen attached to the aortic side of the right coronary cusp. His blood cultures grew Corynebacterium species sensitive to vancomycin and linezolid. He was treated with parental anti-microbial therapy for 4-weeks after which he was referred for surgical correction of his cardiac defects.

Discussion:
The most common heart-hand syndrome is the Holt-Oram syndrome (Type I)[1]. Diagnosis is based on presence of cardiac defects and skeletal preaxial radial ray abnormalities. More than 85% of affected individuals have cardiac malformations that typically include atrial and/or ventricular septal defects, atrioventricular nodal disease, truncus arteriosus, mitral valve defects, patent ductus arteriosus, and Tetralogy of Fallot. In addition to the classical cardiac defects described in Holt-Oram syndrome our case also had sub-aortic membrane with mild aortic regurgitation and endocarditis of the aortic valve which have not been previously reported in literature.

Fig 1: X-ray - Hypoplastic Thumb & Club radius & Ulna

Conclusion
The heterogeneity of presentation of Heart-hand syndromes is well known. Our patient, probably a sporadic mutant had Ventricular septal defect, sub-aortic membrane, Aortic regurgitation and infective endocarditis of the aortic valve in addition to his skeletal anomalies. Subaortic membrane has hitherto never been reported in association with Holt-Oram syndrome in literature.

Fig 2: TTE - Perimembranous VSD with Sub-aortic Membrane
References:


