

Heart Failure Secondary to Holt Oram Syndrome, A Rare Congenital Heart Disease

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ABSTRACT

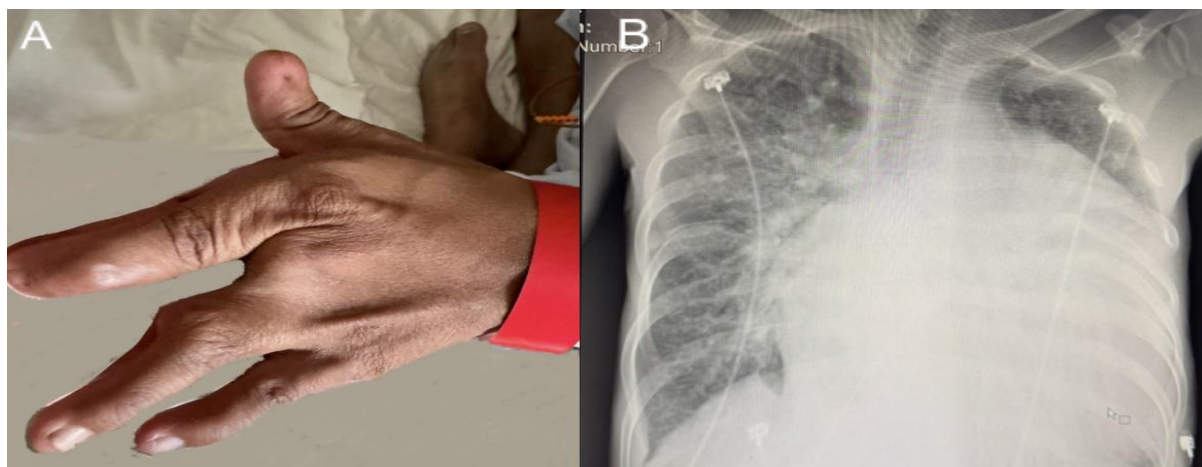
This is a 30-year-old man with a history of cardiomegaly and fingerprint malformations since childhood, originally from an indigenous community, who presents with functional class deterioration with dyspnea and edema in the lower extremities, is found with oxygen saturation of 82%, in electrocardiogram with right bundle branch block and biatrial dilation with atrial flutter. In chest x-ray with grade IV cardiomegaly, echocardiogram with LVEF of 48%, severe mitral regurgitation, with dilation of the 4 cavities, computed tomography reported patent ductus arteriosus type B of Krichenko with pulmonary hyperflow and data of pulmonary hypertension, anomalous connection of pulmonary veins. Treatment with intravenous diuretic was started with clinical improvement, the genetics service was consulted with a final diagnosis of Holt Oram syndrome; Holt-Oram syndrome, also known as heart-hand syndrome, is an autosomal dominant disorder characterized by upper limb abnormalities associated with congenital heart lesions. The incidence is estimated at 1 in 100,000 people. Overall, congenital heart defects are present in approximately 78.7%. Management involves a multidisciplinary team of specialists in medical genetics, cardiology, orthopedics, and hand surgery. Because Holt-Oram syndrome is a rare condition, information on long-term survival may be limited and should be interpreted with caution.

KEYWORDS: Heart-hand syndrome, Holt-Oram Syndrome, *TBX5* variant.

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This is a 30-year-old man with a history of cardiomegaly and finger malformations since childhood, originally from an

indigenous community, who comes presenting functional class deterioration with dyspnea and edema in the lower

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extremities, malformation of the hands and feet with brachydactyly, syndactyly in the bilateral middle finger (Fig.1A). Upon receipt, he is found to have 82% oxygen saturation, an electrocardiogram with right bundle branch block and biatrial dilation with atrial flutter. Chest x-ray (Fig.1B) with grade IV cardiomegaly and data of venocapillary congestion. Echocardiogram with LVEF of 48%, severe mitral regurgitation, with dilation of the 4 cavities, VRT 3.4. Computed tomography reported Krichenko type B patent ductus arteriosus with pulmonary hyperflow and data of pulmonary hypertension, anomalous connection of pulmonary veins supracardiac variety, global cardiomegaly. Treatment with intravenous diuretics was started with clinical improvement. The genetics department was consulted with a final diagnosis of Holt-Oram syndrome. The patient was admitted to the congenital heart disease department.

Holt-Oram syndrome, also known as heart-hand syndrome, is an autosomal dominant disorder due to a mutation in the TBX5 gene located on chromosome 12, which is characterized by abnormalities in the upper limbs associated with congenital heart lesions. (1) First described in 1960 by Holt and Oram, the syndrome was identified when it was associated with thumb abnormalities and atrial septal defects (ASD). (2) The incidence is estimated at 1 in 100,000 people, with no predilection for male or female sex. Cardiac problems are present in approximately 75% of patients with Holt-Oram syndrome and can be fatal. The most common problem is a septal defect.(3) Overall, congenital heart defects, which are present in approximately 78.7% of patients, are a critical factor influencing prognosis.(4) Treatment of the manifestations involves a multidisciplinary team of specialists in medical genetics, cardiology, orthopedics, and hand surgery. Treatment of the arrhythmias may require medication, surgery, and/or pacemaker implantation, in addition to drug therapy for individuals with pulmonary hypertension, and cardiac surgery for congenital heart defects is standard.(4,5) Because Holt-Oram syndrome is a rare condition, information on long-term survival may be limited and should be interpreted with caution.(5) Individualized medical care and ongoing follow-up are needed to optimize outcomes in these patients.

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