

Case Reports

Scimitar syndrome in a child with Turner syndrome

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Introduction

Turner syndrome is known to be associated with significant cardiovascular abnormalities in 23-45%¹. Scimitar syndrome is a rare congenital anomaly characterized by anomalous connection of the pulmonary veins to the inferior vena cava. It represents 3-5% of all partial anomalous pulmonary venous return (PAPVR)². We report a rare case of Scimitar syndrome in a child with Turner Syndrome.

Case Report

A term female newborn presented with swelling of all four limbs. The birth history and family history were unremarkable. On examination, baby was active with normal vital functions. The baby had pitting oedema of hands and feet, webbed short neck, low posterior hairline, broad chest and loose skin folds over the nape of neck. Her cardiac impulse was shifted to the right with a systolic murmur. Ultrasound abdomen suggested horse-shoe kidney, absent ovaries and small uterus. Karyotyping revealed 45XO genotype. Chest x-ray suggested dextroposition of the heart with a 'Scimitar sign'. Echocardiography showed cardiac dextroposition with PAPVR, suggestive of Scimitar syndrome. Thoracic angiography revealed PAPVR with right, lower pulmonary vein draining into the inferior vena cava [IVC] (Figure 1).

She was started on Frusemide 2 mg/kg/day as the pulmonary pressures were high. On follow-up, the limb oedema disappeared. There was no evidence of pulmonary hypertension on echocardiography. Frusemide was stopped after 1 year as the child was asymptomatic. The child is 7 years old at present (Figure 2) and maintains a regular follow-up at the hospital.

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Figure 1: Angiography showing PAPVR with right, lower pulmonary vein draining into IVC



Figure 2: Phenotypic features of Turner Syndrome

Discussion

The frequency of cardiovascular anomalies in Turner syndrome is about 23-45%¹. PAPVR is seen in 13% of Turner syndrome². Scimitar syndrome is a rare congenital cardiopulmonary anomaly seen in 3-6% of patients with PAPVR². This is commonly associated with hypoplasia of the right lung, pulmonary sequestration, persisting left superior vena cava and dextroposition of the heart. The hallmark is drainage of pulmonary veins into the upper part of the IVC, either above or below the diaphragm, and usually of those draining the lower and at times the middle lobe of right lung (79%) or the whole right lung (21%)^{3,4}.

Dupuis et al⁵ divided this syndrome into 3 forms, infantile, adult and a form with associated congenital cardiac anomalies. Infantile form presents within the first 2 months of life with tachypnoea, cyanosis, recurrent pneumonia, failure to thrive, and is often complicated by severe pulmonary hypertension and cardiac failure. Adult form is usually found as an incidental finding on a routine chest x-ray⁶. Our patient was diagnosed while routinely screening the heart for Turner syndrome. Classic findings on physical examination include a shift in heart sounds, cardiac impulse to the right and a systolic murmur, which were found in our case. Scimitar syndrome is almost exclusively a right-sided anomaly, as in our case but rarely may involve the left lung.

The diagnosis is based on the characteristic 'scimitar sign' on chest x-ray [Scimitar means 'Turkish sword']. The diagnosis is confirmed by computed tomography (CT) or magnetic resonance (MR) angiography, though it can be seen on transthoracic or trans-oesophageal echocardiography⁷. Chest radiography did show dextroposition of heart with a scimitar sign in our case and echocardiography with CT angiography confirmed the diagnosis.

Asymptomatic patients in absence of associated abnormalities can be followed-up conservatively. Surgical correction is recommended for patients with congestive heart failure, repeated pneumonia or pulmonary-to-systemic blood flow ratio greater than 1.5 suggestive of pulmonary hypertension^{3,5}. Our patient was conservatively managed and child is now asymptomatic without medication though we supported her with Frusemide in the first year of life.

A high index of suspicion for Scimitar syndrome is important in cases of Turner syndrome with PAPVR. If present, they should be monitored for development of pulmonary artery hypertension and right heart

failure to ensure timely, definitive surgical correction.

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