

A case of right sided cardiosplenic heterotaxy syndrome

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Introduction

Heterotaxy is said to exist when the internal thoraco-abdominal organs demonstrate abnormal arrangement across the left-right axis of the body¹. It is categorized into asplenia and polysplenia syndrome variants¹. Asplenia syndrome is characterized by complex congenital heart defects, asplenia and abdominal heterotaxy¹. Evidence with regard to the incidence of isomerism is sparse¹. However, recent studies show that it is commoner in Asians compared to Western populations². Most cases of right isomerism are recognized in early infancy because of the presence of severe forms of complex cyanotic heart disease¹. Infants with right isomerism invariably have obstruction of the pulmonary outflow tract, as well as common mixing at atria and ventricles, and pulmonary atresia is present in 67% of cases³. Here we report an unusual and relatively late presentation of an infant with right sided cardiosplenic heterotaxy syndrome associated with severe cyanotic congenital heart disease.

Case report

A five month old girl was brought to the paediatric casualty by her non consanguineous parents following a history of sudden onset respiratory distress while breast feeding. She had been well, apart from mild discomfort in the erect position while burping the child. She was born uncomplicated at term, weighing 2.16 kg and was discharged on day 2 with normal newborn examination findings.

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At presentation, child was gasping with severe respiratory distress without central or peripheral cyanosis. Capillary refill time was 4 seconds and oxygen saturation in the probe held in right hand was 60%. Venous blood gas showed severe metabolic acidosis. She was given ventilation breaths with volume resuscitation and later transferred to the paediatric intensive care unit for ventilator care. Despite timely intervention, oxygen saturation did not pick up above 70%. Clinically there was no murmur. Liver was not palpable in the right hypochondrium. Lump measuring 3 cm was palpable below the left costal margin. Electrocardiogram showed right axis deviation with right ventricular hypertrophy and chest roentgenogram showed pulmonary oligoemia and a hiatal hernia. Roentgenogram showed liver in the left hypochondrium with coiling of bowel loops in the right hypochondrium (Figure 1).



Figure 1: X-ray abdomen showing liver in the left hypochondrium & bowel loops in the right hypochondrium

Ultrasound scan of abdomen showed asplenia and left sided liver with midline gallbladder. Sliding hiatal

hernia was seen with poorly visualized stomach. However, nasogastric tube was seen to be passing to right hypochondrial region through the epigastrium. Ultrasound imaging of the brain was normal. Echocardiogram revealed complex heart disease comprising atrioventricular canal defect, partial anomalous pulmonary venous drainage, double outlet right ventricle with pulmonary atresia and duct dependant pulmonary circulation. The left ventricle was rudimentary and was suitable only for a univentricular repair.

Discussion

The prognosis of children with complex cardiac lesions and asplenia is poor. Prolonged functional survival is very uncommon in this variant, with death usually caused by congestive heart failure or severe infection⁴. The one-year mortality is greater than 85% for patients with asplenia⁵. Heterotaxy syndrome with polysplenia has been reported in adults⁶. However, asplenia syndrome invariably presents in early infancy with heart failure, septicaemia and cyanosis and delayed presentations are exceedingly rare⁴.

In this case report, a murmur was not present at birth and child remained well without having any signs of heart failure or chest infections, central or peripheral cyanosis until he developed sudden onset, severe metabolic acidosis with respiratory distress leading to initial suspicion with regard to severe cyanotic congenital heart disease. This highlights the importance of pulse oximetry screening of newborns before discharge from the postnatal ward, irrespective of how healthy or sick they are, so that clinically hidden duct dependant circulations are detected at an early opportunity⁷.

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