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Case Report

Ivemark syndrome with obstructed supracardiac TAPVC- case report

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ABSTRACT

Heterotaxy disorder is a disturbance in the usual left and right distribution of the thoracic and abdominal organs. Ivemark syndrome is one such heterotaxy disorder. It is a rare disorder which affects males more than females with majority of cases presenting in the neonatal period mainly due to complex congenital cardiac disease. Here is a case report of rare disorder of the neonate with Ivemark syndrome with obstructed supracardiac total anomalous pulmonary venous connection (TAPVC). Management of obstructed supracardiac TAPVC is complicated as PGE1 infusion is contraindicated and immediate surgical correction is advised.

Keywords: Heterotaxy, Obstructed supracardiac TAPVC, PGE1

INTRODUCTION

Ivemark syndrome is a rare disorder of right heterotaxy which affects multiple organ systems of the body as a result of lateralization defects. It is characterized by asplenia, complex congenital cardiac diseases along with abnormal arrangement of the internal viscera of the chest & abdomen. It is a rare disorder with high mortality due to cyanotic cardiac anomalies along with repeated infections due to the absence of the spleen.[1] Asplenia syndrome occurs in 1% of newborn with symptomatic CHDs, with male predominance.

CASE REPORT

A Term male neonate of weight 3390 grams, born to a non-consanguineous couple to G2P1D1 mother by emergency LSCS. The first pregnancy resulted in the stillbirth of a male child at 37 weeks of gestation due to unknown reasons. In present pregnancy, antenatal history revealed no exposure to radiation or any drug intake. The baby cried immediately after birth and developed respiratory distress with central cyanosis, within few minutes of life.

On physical examination HR-160/m, RR-75/m, SPO2 – 79%, at room air which improved to 85-90% with oxygen, blood pressure was 68/40 (48). A cardiac examination revealed a grade-3 Systolic murmur and liver was easily palpable in the epigastrium extending on both sides.

Chest radiograph revealed cardiomegaly with CT ratio of 62% with pulmonary plethora with right-sided gastric shadow and midline liver.

Echocardiography showed situs ambiguus, levocardia, obstructed supracardiac TAPVC, double outlet right ventricle, dilated right ventricle and hypoplastic left ventricle, ventricular septal defect (VSD), ostium primum Atrial septal defect, and right ventricle outlet tract obstruction of the pulmonary artery and with common AV valve with normal coronary arteries.

Ultrasound abdomen showed midline liver, right side stomach, absence of spleen. A diagnosis of Ivemark

syndrome was made. Due the presence of obstructed TAPVC, PGE1 infusion was not started and immediate corrective cardiac surgery and risks of surgery were explained to the parents and they refused further intervention. Despite counselling, parents took their child home against medical advice after giving written consent.

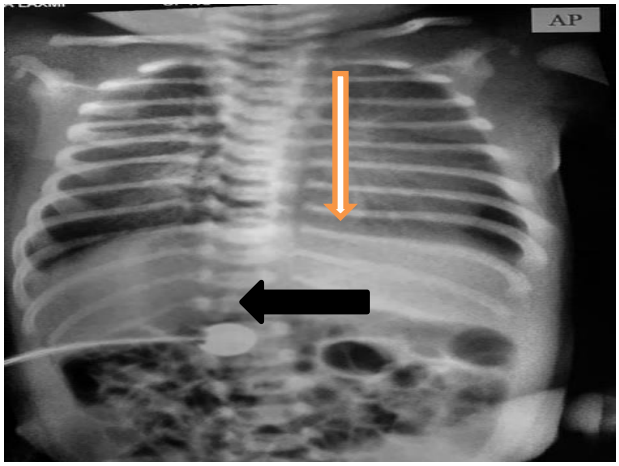

Figure 1: Chest and abdomen radiograph showing levocardia with midline liver extending till left (white arrow) and stomach shadow with orogastric tube towards the right (black arrow).

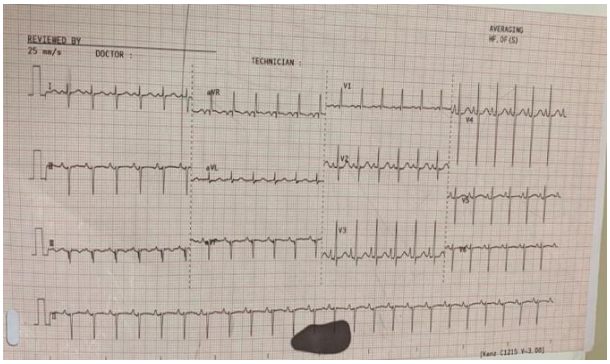

Figure 2: Electrocardiography showed normal p waves and right ventricular hypertrophy.

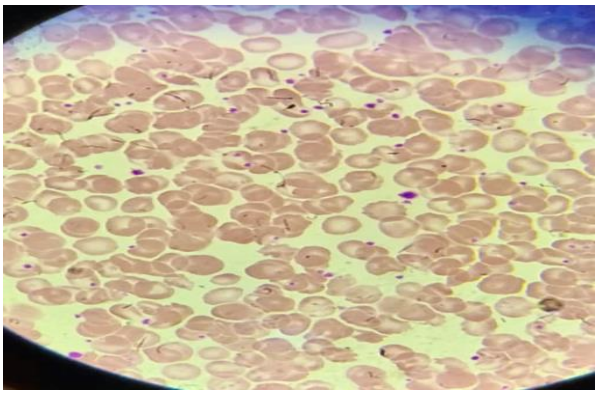

Figure 3: Microscopic picture of peripheral smear showing howell jolly bodies (black arrow).

Baby came for follow up on 12 day of life, and was accepting direct breastfeeds, without respiratory distress with all four limb saturations around 85-87%.

DISCUSSION

Asplenia with cardiovascular anomalies, usually a right atrial isomerism, is an example of a heterotaxy syndrome. It is known as Ivemark syndrome.

Incidence is approximately 1 in 10,000-40,000 births with autosomal recessive inheritance, though development of the Ivemark syndrome may occur either sporadically or in many members of the same family.[2] Although some of the cases are sporadic; autosomal dominant, autosomal recessive and X-linked inheritance pattern have also been described. This X-linked inheritance explains the male predominance.[2,3]

In Heterotaxia syndromes genes involved are NODAL (Asymmetric Gene) involved in nodal signalling and also TGF-B and LEFTYA, LEFTYB and Pitx2, are implicated in the development of heterotaxia syndrome.[3]

In right isomerism the heart is likely to reside to the left of midline. Ventricular-great Arterial connections are usually discordant. Amongst Intracardiac malformations majority of cases have a common Atrioventricular valve, functionally single ventricle and severe pulmonary atresia or stenosis, double outlet right ventricle, and total anomalous pulmonary venous connection (TAPVC).[5] In Ivemark syndrome infracardiac TAPVC (>80%) is most common as opposed to supracardiac type (50%) in isolated cases of TAPVC.[6] In our case, the rare presentation of supracardiac TAPVC with obstruction was seen.

Right isomerism is usually identified first in the neonatal period due to complex cardiac defects. Around 60% of cases present with cyanosis and occasionally present with respiratory distress and cyanosis in cases of obstructed TAPVC in the neonatal period as seen in our case.

Asplenia is the main extracardiac feature; because of this patient experience recurrent and sometimes life threatening infections. Gastrointestinal manifestations include biliary atresia and intestinal malrotation that predisposes to volvulus are common. Midline defects also include meningomyelocele, cerebellar agenesis, cleft lip, cleft palate, horseshoe kidney, kyphoscoliosis and pectus deformity.[7]

The haematological manifestations are mainly due to asplenia which include presence of Howell jolly bodies, target cells and Heinz bodies.[8] In our case, peripheral smear showed Howell jolly bodies.

Treatment of Ivemark syndrome requires a multidisciplinary approach with efforts of a team of specialists such as paediatricians, surgeons, paediatric

cardiologists, paediatric gastroenterologists, pulmonologists, and other healthcare professionals. Before starting PGE1 infusion, obstruction of pulmonary veins should be ruled out, because it leads to increased pulmonary blood flow and reduces the pulmonary vascular resistance and may exacerbate the pulmonary venous congestion when obstructed.[9]

In supracardiac type, a large side-to-side anastomosis is made between the common pulmonary venous sinus and the LA. A systemic-PA shunt may be needed in infancy but the mortality rate is high in asplenia patients than with other defects, and is probably related to regurgitation of the common AV valve and undiagnosed obstructive TAPVC.[6] A Fontan-type operation can be performed. Regurgitation of the AV valve is a high-risk factor, requiring repair or replacement of the valve.[6]

Prophylactic antibiotic therapy can help to reduce the incidence of infection in these patients. Routine immunisation along with optional vaccines against capsulated organisms such as pneumococcal, meningococcal and H influenza B vaccine should be given. Overall survival estimates were 70% at 1 month, 50% at 1 year, and 35% at 5 years.[10]

CONCLUSION

Ivemark syndrome is a rare disorder with complex cardiac disease, especially obstructed supracardiac TAPVC along with asplenia requires prompt diagnosis for early intervention of cardiac correction and use of prophylactic antibiotics to prevent death from recurrent infections. It should also be noted that, not every cyanosed neonate with congenital heart disease benefits from PGE1. This rare disorder should be known to diagnose prenatally and importance of genetic counselling should be emphasised to parents and prevention of recurrence. Not every cyanosed neonate with congenital heart disease benefits from PGE1.

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