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Ivemark Syndrome- A Rare Syndrome with Constellation of Anatomical Defects

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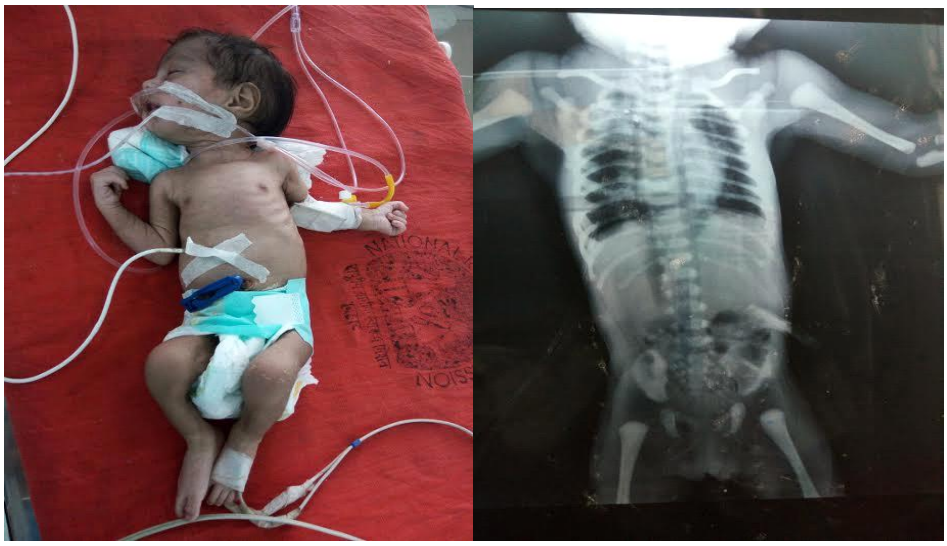
Abstract- *Ivemark syndrome is classified as a heterotaxy disorder or a laterality disorder. These terms refer to the failure of the internal organs of the chest and abdomen to be arranged in the proper location within the body. It is characterized by the absence (asplenia) or underdevelopment (hypoplasia) of the spleen, malformations of the heart and the abnormal arrangement of the internal organs of the chest and abdomen. We report a 7 day old female neonate with this syndrome with complex congenital cardiac defects with right isomerism with asplenia with midline liver.*

I. Introduction

Congenital heart diseases (CHD) affect approximately 0.75-0.9% of newborns and are the leading cause of death in neonates and infants[1]. Among the various kinds of CHD, heterotaxy syndromes (HS) are the most complex. They appear in approximately 1 to 5,000-7,000 of live births with CHD[2,3,6]. Right-atrial isomerism (RAI) or Ivemark syndrome (IS) is a heterotaxy and plurimalformative syndrome with modification of the left–right axis. The result is a complex heart malformation and anomalies of the thoracic and abdominal organs. The revealing element for the diagnosis is asplenia. This condition is rarely seen in adults, because the majority of children with RAI do not survive beyond their first year of life[9,10].

II. Case Report

A 7 days old, full term female was born after an apparently normal pregnancy and labour but with meconium stained liquor. The birth weight of the baby was 2.5kg .The mother was G1P1A0 with no significant family history. Baby had delayed cry after birth with meconium aspiration for which she was admitted in a private NICU for 6 days and presented to us on 7th day of life with complaints of difficulty in breathing. On examination bilateral crepts were present with cardiac murmur and liver was palpable continuosly both on right and left hypochondrium. A plain chest X-ray with abdomen was advised which depicted no fundal gas shadow with opacity in both right and left hypochondrium with consolidation in right upper lung. For this ultrasound of whole abdomen was advised along with echocardiography. Ultrasound revealed asplenia with both lobes of liver enlarged giving bilateral right sided appearance with evidence of fistula between portal vein and an abnormal vessel on right side of Inferior vena cava. Echocardiography showed complex congenital heart disease with dextrocardia with ventricular septal defect with single atrium with transposition of great vessels.



III. Discussion

Heterotaxy syndromes are rare congenital anomalies with multi-system involvement. Grouped under the broad category of situs ambiguous defects, these often pose diagnostic difficulties due to their varied and confusing anatomy. They include a wide range of malformations including both cardiac and extracardiac . In the majority of cases major cardiac malformations reveal this condition in newborns. This is the reason for the high mortality rate (60%) before reaching the 1st year of age[9,10]. Only 5 to 10% of the patients with Ivemark syndrome reach adulthood[10]. It is seen more in males, presents mostly with cyanotic heart disease and patients are immunocompromised due to absent spleen. Neonates with right isomerism typically show a single atrium, a single right ventricle and an atrioventricular connection often associated with atrioventricular valve regurgitation. Pulmonary venous obstruction due to total anomalous pulmonary venous drainage should be precisely diagnosed. The most frequently found hepatic anomaly is a median liver. The complete common mesentery consists of the mesentery rotation being stopped at 90°. The colon is entirely situated on the left side, the small intestine and stomach are placed on the right side of the median line.

Ivemark syndrome is primarily induced by disorders of left-right axis determination during early embryological development. This condition probably arises from a defect in the lateralization, thus causing development disorder in asymmetric organs. Right isomerism is a condition where production of left determinants is low and/or nodal flow is abnormal. As a result, the left signal is not activated in both sides. The exact cause of Ivemark syndrome is unknown. Most cases seem to occur randomly for no apparent reason (sporadic cases). Researchers believe that multiple factors (e.g., genetic and environmental) play a role in the development of the disorder. Ivemark syndrome has occurred in multiple members of the same family suggesting that an inherited genetic predisposition may have been a factor in the development the disorder in these cases.

Most patients with right isomerism succumb within the first year of life due to cardiovascular compromise. Moreover, susceptibility to infection still remains a problem. Survival into adulthood can be explained by less severe pulmonary stenosis, absence of arrhythmias or overwhelming infections. Sepsis due to asplenia, sudden death and cardiac arrhythmias may potentially be related to major causes of late mortality. This case confirms that it is one of the most severe forms of CHD. The prognosis remains poor despite modern surgical techniques. Prenatal diagnosis will allow appropriate counseling for families and will facilitate the prompt treatment immediately after birth.

IV. Conclusion

Therefore, given the high mortality rate in patients with right atrial isomerism it seems prudent to reassess the management of these patients. It is crucial to reveal the anatomical features in these patients by using imaging modalities because being aware of them prior to surgery and invasive intervention prevents the possible risks and complications.

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