

*Case Report***Ivemark Syndrome with Asplenia and Multiple Complex Cardiac Defects - A Case Report**Gaurav Jain^{1*}, Sujata R Kanetkar²¹Senior Resident, Department of Pathology, Hindu Rao Hospital, Delhi, India.²Professor and Head, Department of Pathology, Krishna Institute of Medical Sciences University, Karad, India.

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*Received: 26/07/2013**Revised: 28/08/2013**Accepted: 03/09/2013***ABSTRACT**

Ivemark Syndrome is a very rare sporadic or autosomal recessive congenital disorder that affects multiple organ systems of the body. It is characterized by the absence or underdevelopment of spleen, malformations of the heart and the abnormal arrangement of the internal viscera of the chest & abdomen. Ivemark syndrome is classified as a heterotaxy disorder which refers to the failure of the internal organs of the chest and abdomen to be arranged in the proper location within the body. We present this extremely rare disorder in a full term live born male baby delivered normally to a twenty years old female, married in consanguinity, having severe oligohydramnios. During the immediate postnatal period, baby developed cyanosis and was shifted to NICU for intensive care. However, baby succumbed to cyanotic congenital heart disease on the fourth day. Autopsy was performed which revealed situs ambiguous, asplenia and multiple complex cardiac anomalies. Heart was present in normal situs on left side. Large atrial septal defect was noted forming uniautrial chamber. Other cardiac defects seen were overriding of aorta, pulmonary artery atresia and patent ductus arteriosus. Liver was present on left side and spleen was absent. Microscopic examination of liver, kidney and rest of the viscera showed no significant pathology. Based on above mentioned findings postmortem diagnosis of classical Ivemark syndrome was made. This case highlights importance of genetic counseling to parents for prevention of recurrence in the next child. Role of perinatal autopsy as an important tool in the developmental research is also emphasized.

Keywords: Ivemark Syndrome, Asplenia, Complex cardiac anomalies.**INTRODUCTION**

Ivemark Syndrome is a very rare sporadic or autosomal recessive congenital disorder that affects multiple organ systems of the body. It is characterized by the absence or underdevelopment of spleen, malformations of the heart and the abnormal arrangement of the internal viscera of the chest & abdomen. Ivemark syndrome is classified as a heterotaxy disorder which

refers to the failure of the internal organs of the chest and abdomen to be arranged in the proper location within the body.

CASE REPORT

Our patient was a full term male child born to a twenty years old non-diabetic female married in consanguinity, presenting to labour room with nine months amenorrhea, labour pains and severe

pregnancy induced hypertension. Antenatal care was not received by the mother. There was no history of drugs intake, infection or family history of any congenital heart disease. USG abdomen revealed oligohydramnios and a single live intrauterine fetus in cephalic lie with evidence of situs inversus in abdomen, large atrial septal defect in heart and evidence of single umbilical artery. Baby was delivered by vaginal route with episiotomy. During the immediate postnatal period, baby developed cyanosis and was shifted to NICU for intensive care, where cyanosis deepened and baby developed systolic murmur. A 2D ECHO was done on which revealed congenital cyanotic heart disease with hypoplastic left ventricle, dilated right ventricle and absence of interatrial septum.

The baby died on fourth day and a clinical autopsy was requested.

Autopsy findings

Baby weighed 2330 grams and showed cyanosis. After taking midline incision, the thoracic organs were found in their normal anatomical position, while abdominal organs showed situs inversus with liver on left side and stomach on right side. Spleen was absent (asplenia). Heart showed large atrial septal defect forming uniatrial chamber (cor triloculare biventricularis), hypoplastic (rudimentary) left ventricle, aorta arising from right ventricle, overriding of aorta with pulmonary artery atresia and patent ductus arteriosus. Microscopic examination of liver, kidney and rest of the viscera showed no significant pathology.

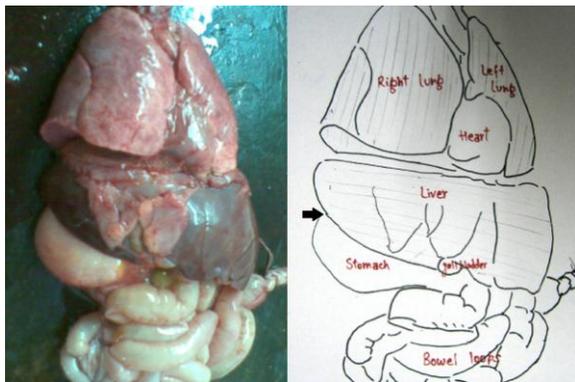


Fig 1 – Situs inversus in abdominal cavity with asplenia (arrow).

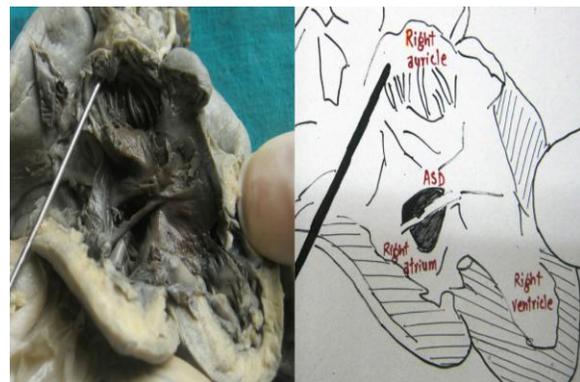


Fig 2 – Large atrial septal defect (ASD) from right atrial view (uniatrialization).



Fig 3 – Overriding of aorta, Aorta arising from right ventricle, Hypoplastic left ventricle, Patent ductus arteriosus and Pulmonary artery atresia (arrow)

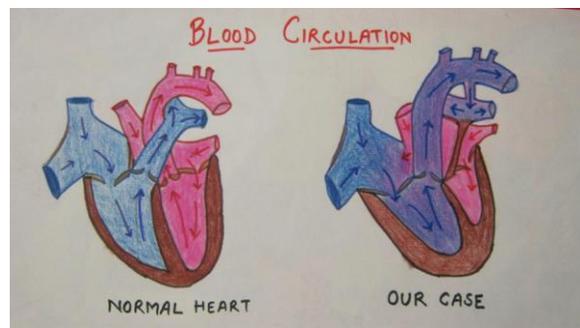


Fig 4 – Aberrant blood circulation in our case was incompatible with life

Based on above mentioned findings diagnosis of Ivemark syndrome was made and cause of death given was congenital cyanotic heart disease with multiple complex cardiac defects, incompatible with life.

DISCUSSION

Ivemark syndrome is named after the Swedish paediatrician, Ivemark, who described the implications of splenic agenesis on the pathogenesis of heart malformations in childhood.^[1] The condition has been described by terms like asplenia syndrome, cardiosplenic syndrome, right isomerism, syndrome of visceral asymmetry or heterotaxy syndrome. The incidence is very low, estimated at 1 in 10,000 to 40,000 live births.^[2] Ivemark syndrome consists of viscerotaxia, congenital heart defects and asplenia.^[3] However this should not be confused with hepato-reno-pancreatic dysplasia syndrome^[4] (or Ivemark II syndrome) also described by Ivemark. As no histological abnormality was detected in liver, kidney or pancreas, our case is of Ivemark Syndrome and not Ivemark II syndrome. The cause of Ivemark syndrome still remains obscure, however researchers believe it has multifactorial inheritance i.e. genetic and environmental. The origin in some cases has been directly related to chromosomal anomalies or defects in a single gene.^[5] Reports of multiple affected siblings, equal male and female incidence and reports of several instances of parental consanguinity suggests autosomal recessive type inheritance. Some sporadic cases are also reported. Heterotaxy results from failure of developing embryo to establish normal left-right asymmetry.^[6]

Various abnormalities that can be seen in this syndrome are, congenital absence of spleen (asplenia), congenital cardiac malformations, cardiac malpositions, maldevelopment of abdominal organs, situs

inversus etc.^[7] Our case showed multiple cardiac malformations with asplenia and situs inversus of abdominal organs. Diagnostic investigations are echocardiography for congenital heart defects and diagnosis of asplenia by technetium sulphur colloid scan. Prenatal diagnosis can be made by antenatal high frequency ultrasound and fetal echocardiography. Prognosis in these cases is related to severity and complexity of cardiac malformations. Anoxia and cardiac insufficiency in severe cases can lead to death in very early life in cyanotic neonates. Similarly in our case the prognosis was poor due to severe congenital cyanotic heart disease. Those with minimal cardiac defects, who survive the first twelve months, there is greater risk of infection and sepsis leading to sudden death due to asplenia.

The treatment of Ivemark syndrome is directed towards the specific manifestations (corrective cardiac surgery). Prophylactic antibiotic therapy can help to reduce the incidence of infection in these patients. Termination of pregnancy can be offered if prenatal diagnosis of severe cardiac anomalies is confirmed in early gestation period. Genetic counselling may be offered to parents of such neonates.^[8]

CONCLUSION

We present this case of Ivemark Syndrome, product of a consanguineous marriage, born to a non diabetic mother who did not receive antenatal care, for the complex cyanotic congenital heart disease with asplenia, which was incompatible with life.

This case highlights the importance of genetic counselling, as there may be recurrence in the next child and significant role of autopsy as a major tool in the developmental research.

REFERENCES

1. Ivemark BI. Implications of agenesis of the spleen on the pathogenesis of conotruncus anomalies in childhood: an analysis of the heart malformations in the splenic agenesis syndrome, with fourteen new cases. *Acta Paediatr* 1955; 44 (Suppl 104):7-110.
2. <http://www.sonoworld.com/TheFetus/page.aspx?id=407>
3. Ferlicot S, Emile JF, Le Bris JL, Cheron G, Brousse N. Congenital asplenia. A childhood immune deficit often detected too late. *Ann Pathol* 1997; 17:44.
4. Ivemark BI, Oldfelt V, Zetterstrom R: Familial dysplasia of kidneys, liver, and pancreas: a probably genetically determined syndrome. *Acta Paediatr Scand* 1959, 48:1-11.
5. Icardo JM, Garcia Rincon JM, Ros MA. Congenital heart disease, heterotaxia and laterality. *Rev Esp Cardiol* 2002; 55:962-74.
6. Bartram U, Wirbelauer J, Speer CP. Heterotaxy syndrome -- asplenia and polysplenia as indicators of visceral malposition and complex congenital heart disease: *Biol Neonate* 2005; 88(4):278-90. Epub 2005 Aug 18.
7. <http://www.rarediseases.org/rare-disease-information/rare-diseases/byID/740/viewAbstract>
8. Cesko I, Hajdu J, Marton T, Tarnai L, Zs Toth E. Familial heterotaxy syndrome. Case report and review of the international literature. *Orv Hetil*. 1998 Nov 15; 139(46):2775-8.

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