Ivemark syndrome in association with congenital septum transversum defect and pancreatic divisum

Shilpa Sharma, A N Gangopadhyay, S P Sharma

Department of Paediatric Surgery, Institute of Medical Sciences, Banaras Hindu University, Varanasi 221 005

A four-month-old female baby presented with cyanosis and respiratory distress. A provisional diagnosis of congenital posterolateral diaphragmatic hernia was made but on exploration there was a defect in the septum transversum along with features of Ivemark syndrome – asplenia with visceroatrial heterotaxia, malrotation and pancreatic divisum – an association not yet reported in literature. The child did well after operative correction of the hernia. Echocardiography showed situs inversus with dextrocardia with double outlet right ventricle, atrial septal defect, ventricular septal defect, patent ductus arteriosus and pulmonary stenosis. [Indian J Gastroenterol 2006;25:94-96]

The characteristic association of asplenia with visceroatrial heterotaxia is traditionally named after the Swedish pediatrician, Ivemark. He described the implications of splenic agenesis on the pathogenesis of heart malformations in childhood. The condition has been described by terms like cardiosplenic syndrome, syndrome of visceral symmetry or heterotaxy syndrome.

The most common congenital defect in diaphragm is through the pleuropitoneal membrane (left > right).
Herniation through the septum transversum is extremely rare.

A 4-month-old girl presented with history of intermittent respiratory distress and cyanosis since birth that increased on crying. There was no history of antenatal polyhydramnios. She was the first issue of her parents and there was no history of prior miscarriage. She was exclusively breast-fed.

On examination, the baby weighed 3.5 Kg. Her pulse rate was 110/min, respiratory rate 40/min and she was afebrile. She had central and peripheral cyanosis and clubbing. Breath sounds were decreased on the left side and there was mediastinal shift to the right side. The liver was palpable 3 cm below the costal margin in the mid-clavicular line. There was no appreciable murmur and heart sounds were heard in the right hemithorax. Her blood investigations were normal. Chest skiagram suggested a diagnosis of left congenital diaphragmatic hernia.

She was operated on through a left subcostal incision. On exploration, the left hemidiaphragm was normal. On elevating the liver, a diaphragmatic defect was seen in the posterior half of the septum transversum, 3 cm x 5 cm in size, anterior to the inferior vena cava, through which abdominal contents were herniating into the chest. The contents – small intestinal loops, transverse colon, stomach and pancreas – were reduced in that order. There was asplenia, which was confirmed by searching all over the abdomen. Associated findings included malrotation and pancreatic divisum with separate ventral and dorsal enlarges. The diaphragmatic defect was closed with interrupted prolene sutures. Ladd’s procedure was done for malrotation. The baby was kept on ventilatory support for 48 hours along with vasopressor support that was gradually withdrawn.

The cyanosis persisted postoperatively, so echocardiography was done, which revealed complex heart disease. Postoperatively the baby had regular heart rhythm and did not go into congestive heart failure. Peripheral blood smear was done to confirm asplenia and it showed Howell-Jolly bodies.

The baby was discharged on the tenth postoperative day after a repeat skiagram (Fig 1) that showed part of the stomach in the chest due to congenital short esophagus. She was given pneumococcus vaccine for asplenia along with regular immunization as per schedule. She has been under medical treatment for the heart disease and is well 9 months later.

Failure of development of the septum transversum is typically associated with underlying pericardial defect as well as with lower sternal defect, upper abdominal wall deficiency and intracardiac anomalies (Cantrell’s pentalogy). Less extensive defects may be associated with some but not all the features of the pentalogy. The case presented here had partial septum transversum defect in association with congenital asplenia, congenital heart disease and large midline liver, constituents of the heterotaxy syndrome.

These patients often have severe anomalies of cardiac looping, frequently with single ventricle anatomy. Evaluation of the cardiac condition reveals evidence of asplenia such as Howell-Jolly bodies in the erythrocytes. Patients should be placed on chemoprophylaxis for asplenia as soon as the diagnosis is made.

The origin of congenital heart disease in some cases has been directly related to chromosomal anomalies or defects in a single gene. Anomalous expression of genes that break the initial embryonic symmetry induces the heterotaxia syndrome that usually courses with congenital heart disease. Digestive tract disorders associated with asplenia / polysplenia syndrome include malrotation, preduodenal portal vein, gastric volvulus, esophageal hiatal hernia and biliary atresia.

Review of literature did not reveal any earlier report of septum transversum defect with heterotaxia, malrotation and pancreatic divisum.

References
4. Gunal N, Bilgic A, Lenk MK, Yurdkul Y, Sarigul A, Ispir S. Abnormal connection of the inferior vena cava to the left atrium with double outlet right ventricle and heterotaxia: a
showed an irregular
The small intestine is the most frequent site
A 62-year-old lady presented with 3-month history
Malignant melanoma in the gut is usually meta-
The tumor cells were large, some epithelioid, with pleo-
Criteria for primary melanoma in-
Growth and differentiation was absent.
Primary malignant melanomas of the GI tract are usually metastatic. Primary malignant
They may either show abundant melanin pig-
S-100 but negative for HMB-45 (Fig). A diagnosis of ma-
Primary malignant melanoma was made. Examination of the skin, eye
malignant melanomas of the gastrointestinal (GI)
Primary malignant melanomas are very rare and occur in the anorectum and esopha-
ulceration.
The search for primary site was negative.
and weight loss of 7 Kg. During the preceding 3 months,
Investigations revealed no evidence of lymph node or other organ involvement.
The presence or absence of cutaneous melanoma was not determined.
showed ascending colon mass, subcutaneous nod-
Investigation of the brain revealed
Hemoglobin 5.5 g/dL;
loss of appetite. When admitted in hospital, the pa-
She was diagnosed to have anemia but was not investi-
Hypochromic, microcytic anemia. CT
The brain was normal without any mass lesion.
left-sided hemiparesis (muscle power grade 3/5); there
inflammatory thickening in the ascending colon
she developed pyogenic meningitis with progressive de-
Investigation of skeletal system showed involvement and subcutaneous nodules; there was no lym-
or cranial nerve deficit. She had
fossa with no clinical evidence of obstruction. She had
Histology and im-
In the skin, lack of other organ involvement, and
of right colon
metastatic mass in the right

Correspondence to: Prof. Gangopadhyay, Professor and Head. E-mail: gangulybhu@rediffmail.com
Received July 25, 2005. Accepted October 7, 2005