



## Case report

# Heterotaxy syndrome with associated agenesis of dorsal pancreas and polysplenia: A case report

Syed Althaf Ali<sup>1</sup>, Syeda Naziya P<sup>2</sup>, Anand Abkari<sup>1</sup>, GSN Murthy<sup>1</sup>, B Ravindra Prakash<sup>1</sup>

<sup>1</sup>Department of Radiology, Deccan College of Medical Sciences, Kanchanbagh, Hyderabad 500058, Telangana, India.

<sup>2</sup>Department of Anatomy, Deccan College of Medical Sciences, Kanchanbagh, Hyderabad 500058, Telangana, India.

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### Corresponding author

**Syed Althaf Ali**

Assistant Professor,  
Department of Radiology,  
Deccan College of Medical Sciences,  
Hyderabad 500058, Telangana, India.  
Phone: +91-8500859966  
Email: althafali98@yahoo.com

### Abstract

Heterotaxy syndrome is a rare embryological disorder comprising of polysplenia, partial agenesis of dorsal pancreas, malrotation of gut, cardiac and vascular anomalies resulting from failure of development of the usual left-right asymmetry of organs. We report a rare case of heterotaxy syndrome with polysplenia, partial agenesis of dorsal pancreas and malrotation of gut in a 28 year female presenting with subacute intestinal obstruction along with imaging illustrations, brief discussion and thorough review of literature.

**Key words:** Heterotaxy, polysplenia, dorsal pancreatic agenesis, malrotation of gut, embryogenesis.

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**H**eterotaxy syndrome derives its name from greek word; "heteros – different and taxis – arrangement"<sup>1</sup>. It is described as situs ambiguous as it follows deranged positioning of thoracoabdominal organs and morphological abnormalities which do not fit in the normal organ positioning of either situs solitus or its mirror image situs inversus<sup>2,3</sup>. The heterotaxy syndrome is due to a primary defect in lateralization of the thoracic and abdominal viscera leading to mal development of the asymmetric organs, sparing the symmetric structures. The syndrome complex is associated with lesions like polysplenia, asplenia, cardiac anomalies, abdominal heterotaxia, short pancreas, malrotation of gut, major vascular aberrations and bronchial malformations<sup>4</sup>. Helwig first described the Heterotaxy syndrome with polysplenia in 1929<sup>4</sup>, since then around 100 cases has been reported in world literature<sup>5</sup>. It is often found in childhood (incidence of 1 per 2,50,000 live births<sup>6</sup>) but only 10% of the patients that do not have cardiac anomalies can reach adulthood and are often diagnosed incidentally.

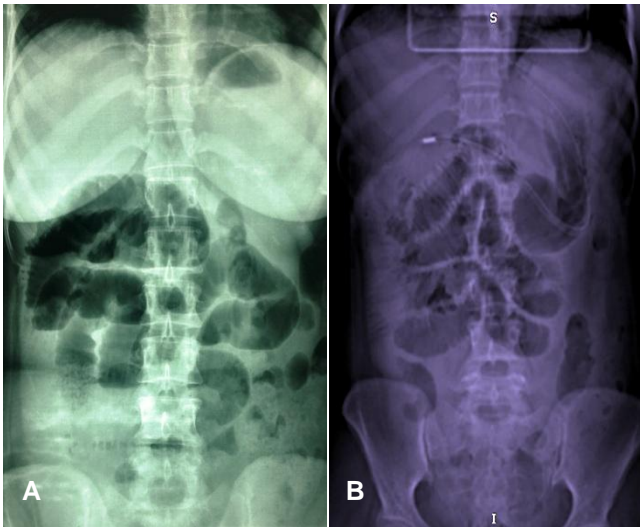
We report a unique case of heterotaxy syndrome associated with polysplenia, agenesis of the dorsal

pancreas, malrotation of gut incidentally diagnosed in a patient presenting with sub-acute intestinal obstruction. The multiplicity and diversity of findings in our case made the individualization of cases extremely valuable and it is pertinent to present this rare entity to add up to the existing world literature.

### Case report

A 28 year old female presented with complaints of pain in abdomen and vomiting since 3 days. On clinical examination there was tenderness all over the abdomen and the diagnosis of sub-acute intestinal obstruction was made. On routine blood analysis patient had hyperglycemia (RBS- 182mg/dl). The WBC count was raised with 80% neutrophils on DLC. Chest X ray was normal. X ray erect abdomen had evidence of multiple air fluid levels suggestive of sub-acute intestinal obstruction (Fig 1A).

On ultrasound the liver was seen extending into the left hypochondrium, three splenules were seen and the tail of pancreas was absent suggestive of Heterotaxy with polysplenia syndrome.



**Fig 1 A.** X ray Erect Abdomen demonstrating multiple air fluid level. **B.** CT Topogram demonstrating mildly distended jejunal loops occupying the right upper quadrant.

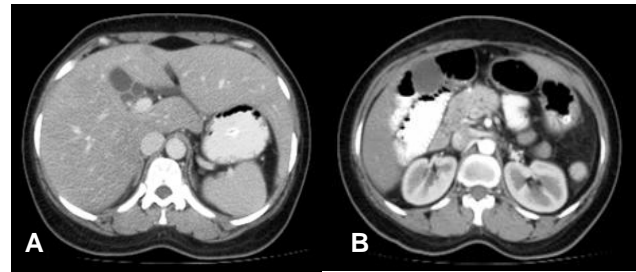
There were multiple fluid filled bowel loops and ascitis suggestive of sub-acute intestinal obstruction. Two hypo to mixed echoic lesions of size 24x20 mm and 9x8 mm were found in liver in segment IV and VII.

On CECT Abdomen (Contrast enhanced computed tomography) with oral & IV contrast, all the findings of USG were confirmed that is a midline liver with a prominent caudate lobe (Fig 2A), absence of pancreatic tail and the pancreatic bed occupied by the bowel loops suggesting partial agenesis of dorsal pancreas (Fig 2B) and presence of three splenules with splenic clefts suggesting polysplenia (Fig 3A). The duodeno-jejunal flexure was seen on right side and the jejunal coils occupy upper right quadrant of abdomen with jejunal branches of superior mesenteric artery coming off from right side, suggestive of malrotation of gut (Fig 3B). There was increased wall thickness and matting of caecum and terminal ileal loops with narrowing of lumen and minimal interloop fluid suggesting ileo-caecal kochs as the cause of subacute intestinal obstruction (Fig 4A). The isodense enhancing lesions seen in liver had persistent filling in venous phase suggestive of hemangioma (Fig 4B).

### Discussion

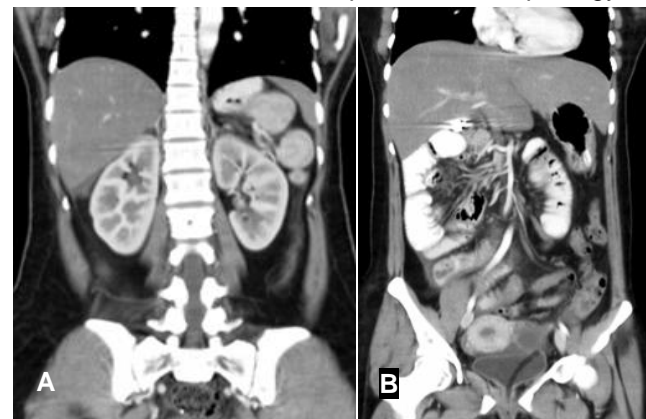
A brief account of embryogenesis is considered pertinent prior to the detailed discussion and review of literature. The visceral sidedness is determined when the blood flow of an embryo makes a transition from symmetric to asymmetric around the 25<sup>th</sup> day. Disruption of this early embryologic event explains the preponderance of common atria, single ventricles, abnormal pulmonary venous connections and

conotruncal anomalies observed in heterotaxy syndrome<sup>7,8</sup>.



**Fig 2 A.** Axial postcontrast CT demonstrating hepatomegaly with liver extending to the left hypochondrium (midline liver) and caudate lobe enlargement (arrow) **B.** Axial postcontrast CT demonstrating absence of tail of pancreas and the pancreatic bed in this region is occupied by intestinal loops (arrow) described as dependant intestinal sign.

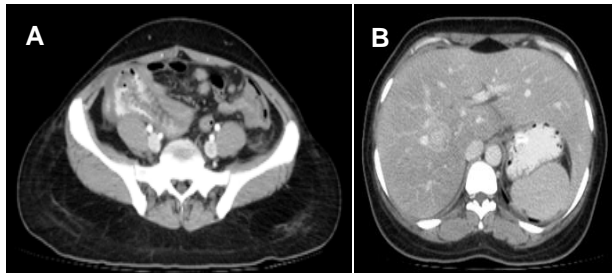
The dorsal pancreas and the spleen develop in the dorsal mesogastrum; thus, concomitant anomalies can be expected in both organs in patients with heterotaxy syndrome<sup>9,10</sup>. The exact mechanism and etiology of dorsal pancreatic agenesis are not known. However, ischemic events in the developing pancreas or primary dysgenesis are the possible explanations<sup>11</sup>. The pancreas develops from two endodermal buds arising from the caudal region of foregut at fourth week and these buds fuse at eighth week of gestation (Fig 5 A-D)<sup>11</sup>. The ventral bud gives rise to the lower portion of the head and the uncinete process whereas the dorsal bud gives rise to the upper part of head, neck, body, and tail of the pancreas<sup>12</sup>. Complete agenesis of the pancreas and agenesis of the ventral pancreas are incompatible with life<sup>13</sup>. The pancreatic development is closely associated with duodeno-jejunal rotation, thus the bowel malrotation is associated with abnormal pancreatic morphology<sup>7</sup>.



**Fig 3 A.** Coronal postcontrast CT demonstrating three splenules (arrows) separated by splenic clefts in left hypochondrium **B.** Coronal postcontrast CT demonstrating jejunal branches (arrows) arising from right side of superior mesenteric artery.

The spleen develops during the 5<sup>th</sup> embryonic week from mesenchymal cells between the leaflets of the

dorsal mesogastrium and the cells of the coelomic epithelium of the dorsal mesentery (Fig 6 A and B). The initial splenic primordia are then created as incisures on the left side of the dorsal mesogastrium and failure of fusion of these incisures leads to polysplenia<sup>14</sup>.

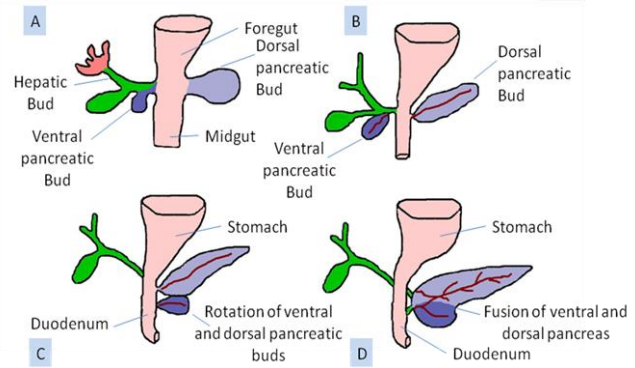


**Fig 4 A.** Axial postcontrast CT demonstrating thickening of ileocaecal walls (arrow) and pericaecal collection **B.** Axial postcontrast CT of venous phase demonstrating enhancing lesions in liver (arrow) suggestive of hemangioma.

Heterotaxy syndrome is usually asymptomatic, but patient may present with abdominal pain, hyperglycemia, diabetes mellitus and acute or chronic pancreatitis<sup>12,15</sup>. Cardiac and thoracic anomalies like atrioventricular septal defect, atrioventricular valvular abnormalities, total anomalous pulmonary venous connection, absence or hypoplasia of the suprarenal inferior vena cava, bilateral bilobed lungs and bilateral hyperarterial bronchi which are usually associated with this syndrome were not found in our case<sup>3</sup>.

The gastrointestinal abnormalities may include a mid-line liver and gallbladder, right-sided stomach and spleen, malrotation of gut, extrahepatic biliary malformations and preduodenal portal vein<sup>8</sup>. The development of diabetes may be due to the absence of the islet cells located in the body and tail of the pancreas as in our case. The increased likelihood of pancreatitis is due to either sphincter of Oddi dysfunction or higher intrapancreatic duct pressures resulting from glandular hypertrophy of the ventral pancreas<sup>3,10</sup>.

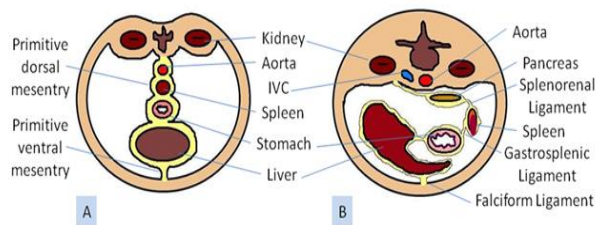
The agenesis of dorsal pancreas needs to be differentiated from pancreatic lipomatosis, pseudo-agenesis, pancreatic divisum and obstructing pancreatic tumors<sup>5,12</sup>. On imaging, fat replacement will be distinguished by abundant fat tissue seen anterior to the splenic vein, whereas agenesis of the dorsal pancreas will be characterized by filling of the distal pancreatic bed with stomach and intestines<sup>5</sup>. Pseudo-agenesis is atrophy of body and tail of the pancreas with sparing of pancreatic head secondary to chronic pancreatitis. Pancreatic divisum has normal body and tail of the pancreas but the ventral and dorsal ducts drain separately into the duodenum in contrast to the absence of dorsal duct in dorsal pancreatic agenesis<sup>5,12</sup>.



**Fig 5 (A-D).** Drawings illustrating the normal embryologic development of the pancreas. **A.** The ventral pancreatic bud arising from the hepatic diverticulum and the dorsal pancreatic bud arising from the dorsal mesogastrium. **B.** Enlargement of the ventral and dorsal pancreatic buds. **C.** The rotation of ventral and dorsal pancreatic ducts along with gut rotation. **D.** The fusion of ventral and dorsal pancreas.

Patients with associated bowel malrotation are more prone to develop volvulus due to presence of thinner mesenteric base. The orientation of the third part of duodenum, the ligament of Treitz, and the bowel helps in diagnosing intestinal malrotation. The inverted relation of superior mesenteric artery and vein along with right-sided jejunal branches can ascertain the diagnosis of malrotation of gut.

Plain radiography and USG can lead to the diagnosis of Heterotaxy syndrome, but sometimes pancreas is not adequately visualized on USG either due to morbid obesity or excessive overlying bowel gases. CT depicts the morphological abnormality of liver, spleen, pancreas, intestines and vessels very well. MRI including MRCP is a noninvasive modality to evaluate the pancreatic ductal anatomy<sup>11</sup>.



**Fig 6 (A and B)** Drawings illustrating the normal embryologic development of spleen. Relationships of spleen, stomach, liver and pancreas during the primitive embryonic stage (A) and in a fully developed stage (B).

Our patient had multiple spleens along the greater curvature of the stomach, a midline liver and partial dorsal pancreatic agenesis with hyperglycemia and malrotation of gut. Radiological evaluation of our patient made us diagnose ileocaecal kochs as a cause of sub-acute intestinal obstruction. The patient has

been conservatively managed with satisfactory follow up.

### Conclusions

The diagnosis of heterotaxy syndrome and associated polysplenia in adult patients is usually made during investigation for unrelated causes. Increased awareness of such anatomical anomalies would prevent serious complications. The importance of imaging studies lies in diagnosing the potential clinical consequences like congenital heart disease, diabetes mellitus, pancreatitis, immune deficiency (in asplenia), and catastrophic volvulus with mal rotation.

Our case is unique and rare considering the associated aberrations and the availability of different imaging modalities under one roof made us assured diagnosis and helped in proper patient care.

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**Conflict of interest:** None

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