Case Report

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Intestinal malrotation with levocardia and Spondylo-costal dysostosis: A case report

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KEYWORDS

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Spondylo-costal dysostosis,
Jarcho-Levin Syndrome

ABSTRACT

Background: Intestinal malrotation with midgut volvulus is usually present in the neonatal period but can present at any age, and can be associated with various congenital anomalies. These congenital defects increase the morbidity of the disease per se and require a multidisciplinary approach.

Case Presentation: The index case was an eight-month-old child with intestinal malrotation, situs inversus, levo-cardia, single ventricle, and Spondylo-costal dysostosis. Here, we discuss its presentation, treatment, challenges during management, and review of pertinent literature.

Conclusion: The association of intestinal malrotation with situs inversus, levocardia, and SCD requires further reporting as well as genetic studies to understand its embryo pathogenesis. The multidisciplinary team is essential for the ideal treatment of this type of complex case.

INTRODUCTION

Intestinal malrotation (IM) with symptoms of intestinal obstruction can present at any age.[1] IM is associated with many congenital malformations like congenital diaphragmatic hernia, omphalocele, congenital heart disease, intestinal atresia, and anorectal malformations.[2] These associated congenital malformations adversely affect the outcome in these children as well as create a diagnostic and surgical challenge to manage the condition. Here, we discuss a case of IM with midgut volvulus in an infant with situs inversus (SI), levo-cardia, complex congenital heart anomaly, and Spondylo-costal dysostosis (SCD). The index case may be the first reported case with a combination of all these entities, association of IM with levo-cardia and situs inversus in isolation as well as with SCD are well documented in the literature although rare.

CASE REPORT

An eight-month-old female child presented at paediatric emergency with complaints of feed intolerance and bilious vomiting for 2 days. The child was full term, born via normal vaginal delivery, and was apparently alright previously. On examination, the child had abnormal posture due to scoliosis (since birth) otherwise had a normal cry, tone, and activity. The patient had central as well as peripheral cyanosis with an oxygen saturation of 70% on room air. There was an ejection systolic murmur over the right 2nd intercostal space. The abdomen was soft, and non-distended with no palpable organomegaly. After resuscitation with intravenous fluids and nasogastric tube drainage, an X-ray chest and abdomen showed fused multiple thoracic vertebrae, abnormal costovertebral alignment, mediastinal shift, and stomach on the right side (Figures 1A and 1B). Ultrasound abdomen showed a left-sided liver and right-sided spleen. The superior mesenteric artery was on the left side of the superior mesenteric vein with a swirling of mesenteric vessels, which was suggestive of midgut volvulus. Echocardiography showed a single ventricle with pulmonary stenosis with possible vegetation on the aortic valve.

After explaining the prognosis in view of cardiac defects and weighing the risks and benefits of the treatment, the child underwent laparotomy via left upper quadrant transverse incision. Intraoperative findings showed a distended stomach on the right side and a volvulus of 180 degrees in an anti-clockwise direction. Duodeno-jejunal (D-J) junction was on the left side with Ladd's band between D-J to

the ileocecal junction which was in the midline (Fig. 1C). The mesenteric vessels were dilated and the intestine was congested even after the correction of volvulus. (Fig.1D) The Ladd's procedure was done in opposite directions i.e. small bowel was placed on the left side of the abdomen and large bowel was placed over the right side, and appendicectomy along with the widening of the mesentery was also done. The child was mechanically ventilated for two days.

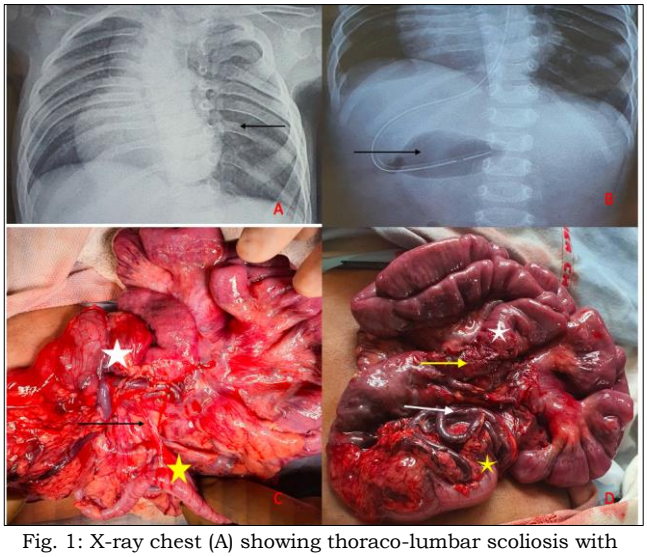

Fig. 1: X-ray chest (A) showing thoraco-lumbar scoliosis with multiple costo-vertebral abnormality (black arrow).X-ray abdomen (B) showing stomach on the right side with nasogastric tube in-situ (black arrow). Intraoperative finding (C) showing Ladd's band (black arrow) between duodenojejunal junction (white star) and ileo-colic junction (yellow star) with dilated congested blood vessels (white arrow) and bowel after widening of mesentery (D) and division of Ladd's band (yellow arrow).

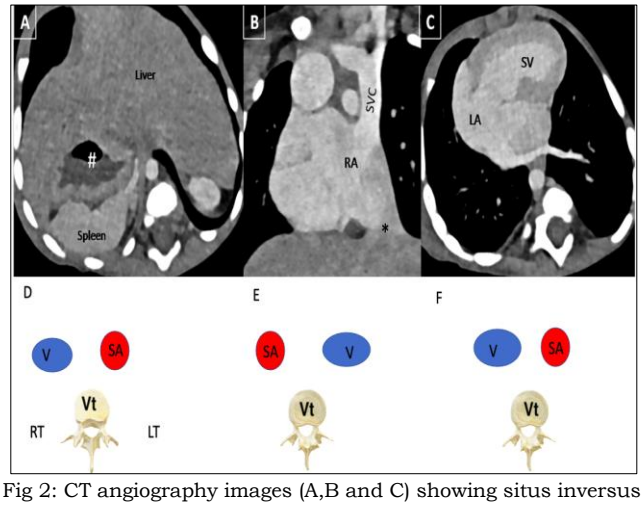

Fig 2: CT angiography images (A,B and C) showing situs inversus with liver on the left side, stomach (#) and spleen on right side, left sided superior (SVC) and inferior (*) vena cava with levocardia and single ventricle (SV) with ventricular apex towards left. The images D,E, and F are showing the relationship of superior mesenteric artery (SA) and superior mesenteric vein (V) in normal individuals, during malrotation in normal individuals and malrotation in situs inversus respectively. [LA=left atrium, Lt= left side, RA= right atrium, Rt= right side, Vt= Vertebra]

Computerized tomography with angiography thorax (Fig 2A) was done after stabilization as it was suggested by the cardiologist to plan future cardiac intervention. It was suggestive of levo-cardia and single ventricle physiology with pulmonary stenosis. The cardiologist advised intravenous antibiotics for 6 weeks because of congenital heart disease and the risk of infective endocarditis. At the same time, an orthopaedic opinion was also taken for abnormal costovertebral alignment and the plan was close observation for respiratory distress. The child had prolonged ileus and oral feeds were started on postoperative day 10 and discharged on day 12. At 6 months follow up the child was under paediatric surgery, cardiology, and orthopaedic team and doing fine.

DISCUSSION

The index case was presented with features of intestinal obstruction and cyanosis with low oxygen saturation, so the initial provisional diagnosis was vascular pathology/ thrombosis. The abnormal posture due to Spondylo-costal Dysostosis (SCD) had created challenges for radiologists to do USG as well as for the cardiologists to perform echocardiography. After investigations, the diagnosis was clear but still, the treatment part was under consideration. A major point of contention was regarding the long-term prognosis of the child related to cardiac anomalies. The cardiologist prognosticated the disease then the child was taken for surgery with due cardiac risk.

Intestinal malrotation (IM) is a rare congenital abnormality affecting 0.2-1% of individuals. Approximately more than 85% of malrotation children present within two weeks of life contrary to the current case presenting at 8 months of age.[3] IM is reported in 2.8-4.1% of all patients with congenital cardiac defects, mainly in single ventricle disease.[4]

Spondylo-costal Dysostosis (SCD), also known as Jarcho-Levin Syndrome is a rare heritable autosomal-recessive growth and developmental disorder of the thoracic skeletal system. SCD can be associated with congenital heart disease, urogenital anomalies, and limb anomalies.[5] These patients have a characteristic 'crab-like' appearance of chest on radiological images due to multiple ribs fused at the costovertebral junction. The restricted development of the chest wall in these children frequently causes respiratory distress with a mortality rate of 32% in childhood.[6] In the index case, the diagnosis of SCD was suggested based on radiographic features as per literature and genetic testing to identify pathogenic variants was not done because of the family's financial constraints.[7] There was no history of similar complaints in the first and second-degree relatives. There was no respiratory distress and so the child was managed conservatively and kept under follow-up for possible future intervention whenever needed.

Situs inversus (SI) is a congenital anomaly with inverted transposition of the internal organs and can be associated with IM and cardiac anomalies.[8] SI is strongly associated with dextrocardia but on the contrary, this child had levo-cardia (cardiac apex to the left) which is extremely rare.[9] Almost 100% of children of SI with levocardia have associated congenital heart disease.[10] SI is also associated with a congenital deformity of the spine such as tethered spinal cord, and scoliosis.[11]

In children with IM, the superior mesenteric artery (SMA) lies to the right of the superior mesenteric vein (SMV). But in children with IM along with situs inversus (SI), SMA is situated to the left of SMV (Fig 2E, F, and G) similar to the index case. The preferred treatment for intestinal obstruction due to IM is Ladd's procedure. In the current case, all steps were followed but in the opposite direction. Intraoperatively, the mesenteric vessels were dilated and the intestine was also congested, the possible explanation could be the long-term intermittent volvulus that could create these types of changes.

The co-segregation of SCD and the laterality defects has been only reported in a large extended Arab family by Sparrow et al in the year 2013. They reported that a mutation in the gene HES7 causes segmental defects of the vertebrae with complete penetrance and dextrocardia with situs inversus with incomplete penetrance.[12] The exact genetic etiology is yet to be established for this perturbation. Dextrocardia with situs inversus is the least pathological type and occurs in 1/8000 live births. In contrast, situs inversus with levocardia in combination with SCD as in our index case has never been reported in the literature earlier.[13] Table 1 illustrates a few case reports with varying presentations with none having a similar combination as the index case.

Table 1: Shows case reports of children with situs inversus in varying combinations of anomalies such as dextrocardia, levocardia, congenital heart defects, malrotation with midgut volvulus, small bowel atresias, and segmental vertebral defects. No case has been reported with the combination of anomalies as in the index case

Study	Diagnosis	Patient details	Comments
G D Ruben et al (1983)[14]	Situs inversus with levocardia and acute abdominal emergencies (n=7)	midgut volvulus (n=1). Malrotation (n=4).	All underwent Ladd's procedure. 2 out of 3 deaths were due to major cardiac defects
Budhiraja S et al (2000)[15]	Situs inversus with levocardia with congenital heart defects (tricuspid valve stenosis)	A 6-week-old male child with malrotation and midgut volvulus.	Ladd's procedure with inversion appendectomy was done.
Rahul Gupta et al (2017)[16]	Situs inversus with dextrocardia with malrotation and volvulus.	One-month-old male infant with associated small jejunal segment gangrene. A four-day-old female patient with associated multiple jejunal atresia with bilateral renal dysplasia	Ladd's procedure with resection and anastomosis of jejunal segments. Both patients expired due to sepsis and multiorgan failure respectively.
A Nawaz et al (2005)[17]	Situs inversus with levocardia with congenital duodenal obstruction	A two-day-old female child with tetralogy of Fallot with duodenal atresia. A four-day-old female child with duodenal web.	Duodenoduodenostomy was done and both neonates were discharged in stable condition.
Sparrow et al (2013) [12]	Situs inversus with dextrocardia with segmental vertebral defects.	Two distantly related individuals	Both were homozygous for the mutation in the HES7 gene.

To conclude, the association of intestinal malrotation with situs inversus, levo-cardia, and SCD requires further reporting as well as genetic studies to understand its embryo pathogenesis. The prognosis depends upon the severity of associated anomalies especially cardiac, the multidisciplinary involvement is crucial to manage this type of complex cases.

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