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Case Report

Polysplenia syndrome associated with multisystem malformation: A rare case report ☆

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ABSTRACT

Polysplenia syndrome is a rare congenital disease with multiple systemic developmental abnormalities. Their occurrence and development are closely related to embryonic development. The prognosis of the disease depends on its anatomical structure, and the presence or absence of concomitant cardiac malformations also has a prognostic impact. Here, we report a case of an adult female who showed the presence of epigastric pain and was diagnosed with polysplenia syndrome after an abdominal CT scan. Imaging further revealed the presence of developmental malformations in several systems, including cardiovascular, respiratory, and digestive systems. This case is rare and therefore has some clinical reference value.

Introduction

Polysplenia syndrome is a rare congenital disease, often accompanied by anatomical abnormalities of multiple systems. Research indicates that multiple system developmental abnormalities are most likely to occur between the 5th and 7th weeks of embryonic development [1]. Over 50% of the polysplenia syndrome patients do not survive beyond 5 years, with the majority of deaths due to severe cardiovascular malformations. Polysplenia syndrome is more common in females and presents directly with several splenic nodules of varying sizes, mostly in the right upper abdomen and occasionally bilaterally. Polysplenia syndrome often involves developmental malformations of multiple organs, including pancreatic hypoplasia, heterotaxy syndrome, situs inversus totalis, absence of the hepatic segment of the inferior vena cava, and left-sided inferior vena cava changes [2]. Here, we describe a patient with typical manifestations of polysplenia syndrome.

Case presentation

A case of a 46-year-old female who presented with abdominal pain. The chief complaint was pain in the thoracic back, lower back, and upper abdomen for 1 week. There was no history of

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surgery, no family history of genetic disorders, and a 2-year history of allergic rhinitis. Physical examination: abdominal tenderness, mild pressure pain in the epigastrium and left lower abdomen without rebound pain, and mild percussion pain in the lumbar region bilaterally. In laboratory tests, mean corpuscular volume, haemoglobin volume, and mean corpuscular haemoglobin volume were reduced, and the rest of the indicators were within normal limits. On follow-up ultrasonography, it was suggested that the uterus was enlarged and multiple fibroids were seen in the uterine myometrium. The patient was found to have several systemic anatomical malformations on computerized tomography. Afterward, we consulted the patient's attending physician and deduced that the uterine fibroids were causing the patient's abdominal pain. Patients are treated with medication, taking orally Kangfuyan capsules and diclofenac sodium sustained release tablets. After that, the patient's abdominal pain has improved somewhat. During this period, the patient has no history of hospitalization or surgery.

The chest CT scan images of the patient revealed multiple anatomical abnormalities (Fig. 1). In the mediastinum window, the abnormal left-sided inferior vena cava is situated behind the descending aorta. The azygos vein joins the left inferior vena cava at the level of the 8th thoracic vertebra. The hepatic segment of the left inferior vena cava is absent, with the hepatic veins directly joining the right atrium. In the lung window, bilobar changes in the right lung. The right oblique fissure is more symmetrical with the left oblique fissure, but there is no right horizontal fissure, no right middle lobe bronchus seen, and heterotopia on the left side of both lungs.

The abdominal CT scan images of the patient also revealed multiple anatomical abnormalities (Fig. 2). There are multiple nodules in the spleen area in the upper left abdomen, with the density of these nodules consistent with the nor-

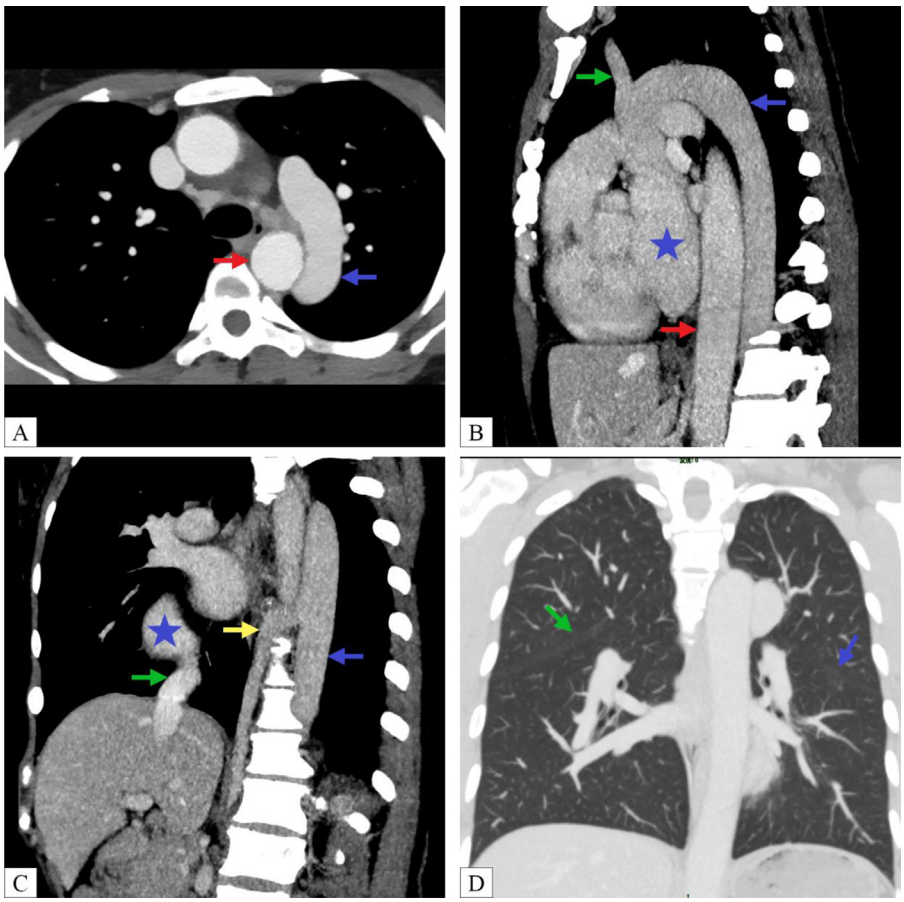

Fig. 1 – The patient's chest CT scan images. (A) CT-enhanced Mono E40kev arterial phase image (axial plane). The left inferior vena cava (blue arrowhead) lies to the left of the descending aorta (red arrowhead). (B) CT MIP image (sagittal plane). The left brachiocephalic vein (green arrowhead) joins the left inferior vena cava (blue arrowhead) and finally joins together into the right atrium (blue stars). The left inferior vena cava runs posterior to the descending aorta (red arrowhead). (C) CT MIP image (sagittal plane). The azygos vein (yellow arrowhead) drains into the left inferior vena cava (blue arrowhead) at the level of the 8 thoracic vertebra. The hepatic segment of the vena cava is absent, and direct drainage of the hepatic veins (green arrowhead) into the right atrium (blue stars). (D) CT MIP image (coronal plane). Heterotopia on the left side of both lungs, the right oblique fissure (green arrowhead) was more symmetrically positioned to the left oblique fissure (blue arrowhead), absence of the right horizontal fissure, and bilobar changes in the right lung.

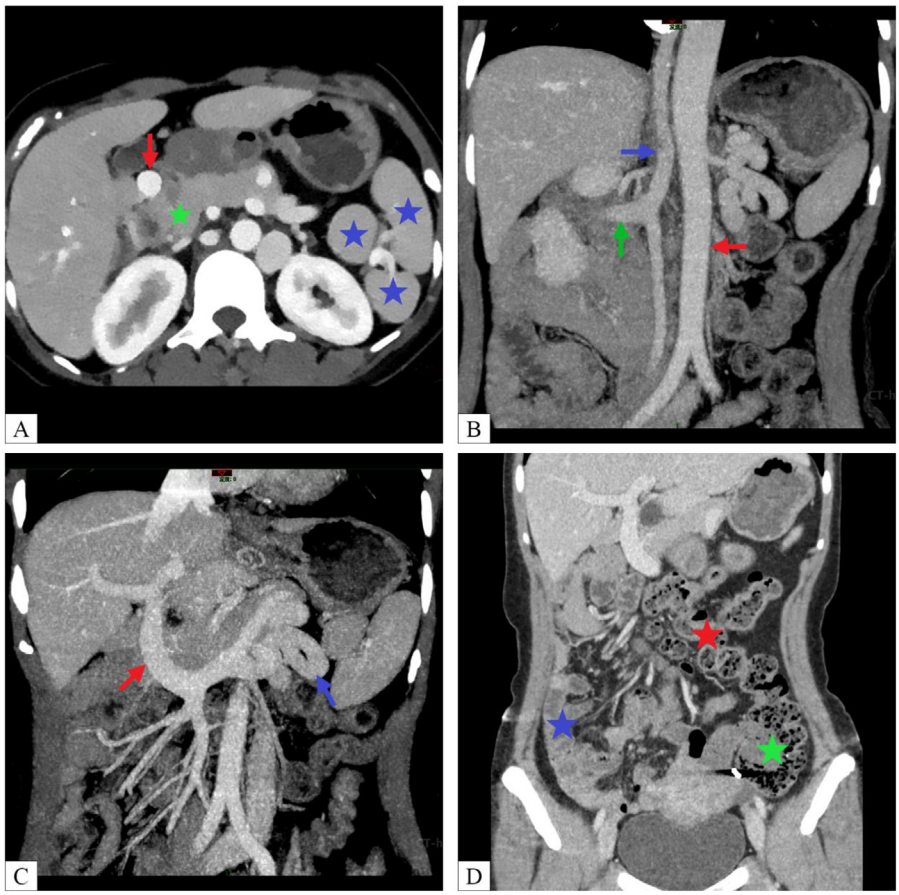

Fig. 2 – The patient's abdomen CT scan images. (A) CT-enhanced Mono E40kev arterial phase image (axial plane). Multiple splenic nodules (blue stars) of varying sizes in the splenic area of the left upper abdomen. Part of the head and neck of the pancreas (green star) is hypertrophied and spherical, and the tail of the gland is short. The portal vein (red arrowhead) is located anterior to the duodenum. (B) CT MIP image (coronal plane). The right renal vein (green arrowhead) flows back into the azygos vein (blue arrowhead), and the azygos vein is located to the right of the abdominal aorta (red arrowhead). (C) CT MIP image (coronal plane). The anterior duodenal hepatic portal vein (red arrowhead) is enlarged and thickened, and the splenic vein (blue arrowhead) is enlarged and varicose. (D) CT MPR image (coronal plane). The horizontal segment of the duodenum does not cross the midline, the small intestine loops (blue star) are located in the right abdomen. The colon (red star) is in the left abdomen, and the ileocecal region (green star) is in the left lower abdomen.

mal spleen, with splenic vessels entering the nodules. The enhancement pattern after dynamic enhancement is consistent with the normal spleen. The head and neck of the pancreas are enlarged and spherical, and the tail of the gland is short, indicating changes suggestive of a short pancreas. The right renal vein does not drain into the inferior vena cava but instead drains into the azygos vein. The portal vein in front of the duodenum is enlarged and thickened, with splenic veins also dilated. The horizontal segment of the duodenum does not cross the midline, with the small bowel loops located in the upper right abdomen, the colon in the left abdomen, and the cecum in the lower left abdomen, indicative of malrotation of the midgut. Finally, we performed Volume Rendering (Fig. 3) and showed the location of the inferior vena cava and the abdominal aorta directly.

The patient has multiple splenic nodules on anatomical examination, accompanied by anatomical abnormalities in multiple systems such as the vascular, respiratory, and digestive systems. We can make a full diagnosis of polysplenia syndrome.

Discussion

Polysplenic syndrome, a rare congenital disorder whose roots can be traced back to developmental anomalies during embryonic life, has a probability of incidence of only one in forty thousand [2]. The exact cause of polysplenic syndrome is not yet clear. It is currently believed that embryonic, genetic, and teratogenic factors are all pathogenic factors for polysplenic syndrome [3].

The anatomical complexity of the disease is remarkable for a series of developmental malformations of the abdominal viscera, such as a large midline liver, inversion of the viscera, midgut malrotation, atresia of the bile ducts and duodenum,

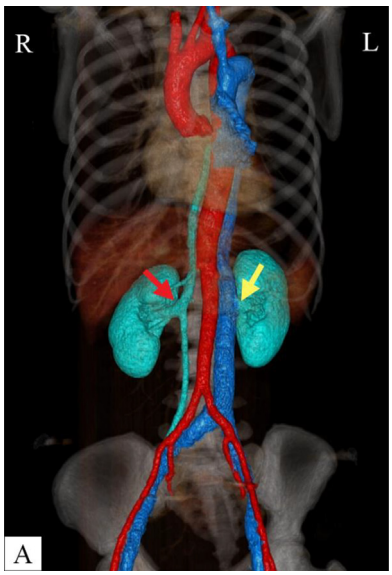

Fig. 3 – The patient's CT VR images. (A) The right renal vein (red arrowhead) flows back into the azygos vein, and the left renal vein (yellow arrowhead) drains directly into the left inferior vena cava. The left inferior vena cava runs behind the left side of the descending aorta.

and shortening of the pancreas. In addition, the patient is often associated with left-sided heterotaxy of both lungs and a variety of cardiovascular malformations, such as absence of the hepatic segment of the inferior vena cava, left inferior vena cava variant, atrial septal defect, single atrium, and double outlet of the right ventricle [4].

The number of spleens varies from 2 to 6, with diameters ranging from 1 to 6 centimeters. The spleen is located in the upper left or upper right quadrant [2]. In the fifth week of embryonic development, the initial splenic primordia forms a notch on the left side of the dorsal mesogastrium. When the notch fails to fuse, 2 or more spleens may form [5].

The most common vascular abnormality in polysplenic syndrome is the incomplete development of the inferior vena cava [5]. The embryonic development of the inferior vena cava is complex. The normal inferior vena cava is composed of 4 segments: hepatic, suprarenal, renal, and infrarenal. Due to developmental abnormalities, the hepatic segment of the inferior vena cava usually exists and separately drains into the right atrium [6]. In this case, the patient not only has the absence of the inferior vena cava liver segment but also has abnormal changes in the left inferior vena cava. Most cases of ectopic left inferior vena cava are associated with visceral ectopia syndrome and abnormal retrograde flow in the azygos venous system [7].

The normal pancreas is formed by the fusion of ventral and dorsal pancreatic buds. The ventral pancreatic bud produces the uncinate process and the head, and the dorsal pancreatic bud produces the body and tail. Most polysplenic syndromes are associated with short pancreas anomalies. Abnormal development of the pancreas may lead to pancreatic insufficiency, which can easily cause pancreatitis and diabetes [3].

The present case involved intestinal malrotation, characterized by the horizontal segment of the duodenum not crossing the midline, small bowel loops located in the upper right abdomen, colon situated in the left abdomen, and the cecum positioned in the lower left abdomen. Even in the absence of symptoms, it is difficult to predict the risk of torsion in patients [8]. Considering that the patient in this case is an adult and has no obvious clinical symptoms, there is no need for surgery.

The case combines the bilaterally left lung malformation. It presents as a defect in the right horizontal fissure, an absence of the right middle lobe bronchus, and a 2-lobed change in the right lung. Patients with incomplete lung development may live healthy lives or may experience atypical symptoms. Most cases of hypoplastic lung are related to bilobed lungs, liver transposition, congenital heart disease, azygous continuation of the inferior vena cava, and polysplenia [9].

Polysplenia syndrome combined with congenital heart malformations is also quite common, such as atrial septal defect, ventricular septal defect, bilateral superior vena cava, right aortic arch, partial anomalous pulmonary venous return, transposition of the great arteries, pulmonary valve stenosis, and coarctation of the aorta [4]. Research shows that most patients with polysplenia syndrome do not live to adulthood due to associated congenital heart anomalies. However, 5%-10% of polysplenia syndrome patients do survive into adulthood, and it may be incidentally diagnosed, with most not having severe heart malformations [8], just like our patient.

The patient presented with abdominal pain. A series of ultrasound and CT scans were performed to rule out appendicitis and ovarian torsion. Subsequently, multiple spleens were discovered, but their positioning was acceptable, ruling out splenic torsion [8]. Finally, we deduced that the cause of the patient's abdominal pain was not related to polysplenic syndrome, but was due to uterine fibroids. After symptomatic treatment for uterine fibroids, the patient's abdominal pain improved. Polysplenia syndrome is a congenital developmental anomaly, and the discovery of polysplenia syndrome in this case was incidental. The patient in this case is currently an adult, with several anatomical abnormalities but no anatomical abnormalities involving the heart, and no obvious clinical symptoms, indicating no need for corrective surgery [8]. We speculate that the patient has adapted to this ectopic blood circulation and may have made compensatory changes. The patient in this case did not undergo splenectomy and the spleen has been preserved functionally, so it was not necessary to vaccinate the patient.

The prognosis of polysplenic syndrome depends on age. In children with concomitant heart disease, the prognosis is usually poor and generally requires surgical treatment. In adults, symptomatic treatment is usually adopted [8]. Therefore, for this adult with polysplenic syndrome, no special treatment has been given clinically, and there are no necessary preventive measures. There are no apparent anatomical abnormalities of the heart involved, and it can be speculated that her prognosis is good.

Although polysplenic syndromes often coexist with developmental malformations of multiple systems, their specific clinical manifestations are not specific. In addition, the prognosis of patients with polysplenism is directly related to their

anatomical structure. And their mortality rate is significantly increased when combined with severe congenital heart disease. Therefore, it is important to perform exhaustive imaging studies to assess the developmental malformations in patients fully.

Conclusion

To this end, we present a case of polysplenic syndrome with multiple systemic malformations. This is a case of a middle-aged female patient. She presented with abdominal pain symptoms caused by uterine fibroids. During the subsequent examination process, the presence of polysplenic syndrome was accidentally detected. The diagnosis of polysplenic syndrome was made with a series of rare anatomical variations, including short pancreas, midgut malrotation, left-sided heterotaxy of both lungs, absence of the hepatic segment of the inferior vena cava, and left-sided inferior vena cava. Despite the presence of various malformations in this patient, these malformations did not significantly affect the patient's daily life. For incidentally detected polysplenic syndrome in adults, there is no need for specific treatment in clinical practice. This case not only deepens our understanding of the complexity of polysplenic syndrome but also provides a valuable reference for radiologists and clinicians in the management of similar cases.

Patient consent

Written informed consent was obtained from the study participant.

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