Polysplenia Syndrome with Various Visceral Anomalies in an Adult: Embryological and Clinical Considerations

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A 26-year-old female with polysplenia syndrome is reported. She had numerous visceral anomalies including polysplenia, a short pancreas, a preduodenal portal vein, malrotation of the bowel, azygos continuation of the inferior vena cava, bilateral hyparterial bronchi and symmetrical liver lobation. Embryological and clinical considerations of polysplenia syndrome are described. (Internal Medicine 31: 1026-1031, 1992)

Key words: short pancreas, preduodenal portal vein, azygos continuation of the inferior vena cava, bilateral hyparterial bronchi, malrotation of the bowel

Introduction

Polysplenia syndrome is a rare congenital anomaly frequently associated with various visceral anomalies including multiple spleens, impaired visceral lateralization, congenital heart diseases, gastrointestinal abnormalities and azygos continuation of the inferior vena cava (IVC) (1, 2). However, short pancreas and preduodenal portal vein (PPV) are unusual congenital anomalies in association with polysplenia syndrome. A patient with numerous congenital anomalies including short pancreas and PPV in association with polysplenia syndrome is described in this paper. The embryogenesis and the clinical implications of this anomaly are also discussed.

Case Report

A 26-year-old female began to feel dull pain in the right upper quadrant of the abdomen six years ago. The pain occurred several times a year and disappeared spontaneously within a few minutes. At a local hospital, she was noted to have an abnormal mediastinal shadow on a chest X-ray film and an abnormality of the IVC on an abdominal ultrasonogram a few years ago. A definitive diagnosis, however, could not be made at that time. She visited our hospital for a complete medical evaluation.

Chest X-ray films revealed a convexity in the right superior mediastinum (Fig. 1A) and an absence of the retrocardiac IVC shadow on the lateral view (Fig. 1B). Contrast-enhanced chest computed tomographic (CT) scan revealed a dilated azygos vein in the mediastinum, which drained into the superior vena cava (SVC) through an azygos arch at the level of the aortic arch (Fig. 2A–C). Bilateral hyparterial bronchi were also demonstrated (Fig. 2B). Contrast-enhanced abdominal CT scan demonstrated absence of the hepatic portion of the IVC and a dilated azygos vein which parallels the aorta anterior to the spine in the retrocrural space (infrahepatic interruption of the IVC with azygos continuation) (Fig. 2D and E). On T1-weighted (530/31/2, TR/TE/excitations) coronal magnetic resonance imaging (MRI), an enlarged anomalous collateral vein originated from the right renal vein, ascended in front of the right kidney (Fig. 3B) and emptied into the azygos vein at the level of the intervertebral disk between Th10 and Th11 (Fig. 3A). The hepatic vein drained directly into the right atrium (Fig. 2C). There were several small spleens, 1 to 3 cm in diameter, in the left hypochondrium (polysplenia) (Fig. 2D and 3A). The liver extended across the midline and tended to be symmetrical. An accessory lobe of the liver, which connects to the right lobe, was also seen (Fig. 2E). The portal vein was positioned anterior to the duodenum (PPV). A short round pancreas and a midline-positioned gallbladder were present (Fig. 2F). These visceral anomalies were also seen on abdominal ultrasonograms (Fig. 4). X-ray examinations of the upper gastrointestinal tract and the colon revealed malrotation of the bowel (Fig. 5). The echocardiogram showed no abnormality.

Discussion

Polysplenia syndrome is known to be frequently associated with cardiopulmonary, gastrointestinal, geni-
Fig. 1. A) A posteroanterior chest X-ray film shows a round mass in the right superior mediastinum (arrow), which was subsequently shown to represent a dilated azygos arch. B) The retrocardiac IVC shadow is absent on a lateral view of the chest.

This syndrome also has a strong tendency for normally asymmetric organs to develop symmetrically and has been called "bilateral left-sidedness" (3). Anomalies associated with polysplenia are interesting from the embryogenesis point of view. Since splenic anomalies such as splenic agenesis, hypogenesis and polysplenia, are known to be associated with many congenital anomalies of other visceral organs, the spleen is considered to play an important role in the normal development and lateralization of visceral organs (4-6). The present patient had polysplenia, a short pancreas, a PPV, an accessory liver lobe, a tendency toward symmetrical liver lobation, a midline-positioned gallbladder, malrotation of the bowel, azygos continuation of the IVC with an anomalous collateral vein, and bilateral hyparterial bronchi (bilateral left-sidedness of the lung). Although some of these anomalies are well known in association with polysplenia syndrome, the combination of these numerous visceral anomalies including short pancreas, PPV and azygos continuation of the IVC as seen in the present patient has not yet been reported. In particular, short pancreas and PPV are unusual associated anomalies. Most patients with this syndrome have symptoms in childhood as a result of associated cardiac anomalies (2). The other anomalies may be discovered incidentally in adulthood because they do not present any pathognomonic symptoms. The association between abdominal pain and polysplenia syndrome in the present patient is not clear, but malrotation of the bowel may be involved.

The association of polysplenia with congenital short pancreas was first reported in 1984 by Hatayama and Wells (7). Thereafter, only three case reports of a short pancreas in polysplenia syndrome have been published in 1991 (8-10). The present patient showed a short pancreas measuring 4.0 × 2.3 cm in diameter on abdominal CT scan. Short pancreas is also known to be caused by pathologic conditions including chronic pancreatitis, ischemic atrophy of the pancreatic tail, kwashiorkor and generalized viral infection. None of these pathologic conditions were found in the present patient. The cause of congenital short pancreas has been presumed to be agenesis of the dorsal pancreas. Tanaka et al (11) reported that endoscopic retrograde pancreatography (ERP) reveals the presence of the accessory pancreatic duct in two-thirds of the patients...
Fig. 2. Contrast-enhanced CT scan. A) A dilated azygos arch (white arrow) and the aortic arch (open arrow), forming a collar around the trachea, are seen at the level of the carina. B) A dilated azygos vein (Az) paralleles the aorta (Ao) anterior to the spine. Bilateral hyparterial bronchi are seen. C) The hepatic vein (arrow) enters directly into the right atrium. D) Four small spleens (asterisk) are present posterior to the stomach (St). D and E) An enlarged azygos vein (Az) adjacent to the aorta (Ao) and an enlarged collateral vein (C) in front of the right kidney are seen. The IVC is not identified within the liver. The liver tends to be symmetrical. E) An accessory liver lobe (asterisk) is present. F) A preduodenal portal vein (Pv), a short pancreas (P) and a midline-positioned gallbladder (GB) are present. A collateral vein (C) originates from the right renal vein. The IVC shadow (arrowhead) can be seen at the level of the hilus of the kidney. D: duodenum, Sv: splenic vein.

with short pancreas, suggesting that short pancreas may be caused by hypoplasia rather than complete agenesis of the dorsal pancreas. While most patients with short pancreas in normal situs had diabetes mellitus occasionally accompanied by pancreatitis, none of the patients with short pancreas in polysplenia syndrome had diabetes mellitus and pancreatitis. Therefore, the embryogenesis of short pancreas in polysplenia syndrome may be different from that of
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Fig. 3. T1-weighted (530/31/2, TR/TE/excitations) magnetic resonance imaging (coronal section). A) Multiple spleens (asterisk) are present between the diaphragm and the left kidney. A collateral vein (arrow) empties into the azygos vein (arrowhead) at the level of the intervertebral disk between Th10 and Th11. B) An enlarged anomalous collateral vein (arrow) originates from the right renal vein and ascends in front of the right kidney.

Fig. 4. An ultrasonogram of the abdomen shows a preduodenal portal vein (Pv), a short pancreas (P), a midline-positioned gallbladder (GB) and an azygos vein (Az). The splenic vein (arrow) and the superior mesenteric artery (arrowhead) are also seen. Ao: aorta, D: duodenum, L: liver.

because PPV is frequently found in combination with various congenital anomalies including intestinal malrotation, situs inversus, duodenal atresia, annular pancreas, biliary atresia and splenic anomalies (21). Malposition of the abdominal viscera may be another cause in the formation of PPV. Clinically, PPV is an anomaly of considerable surgical importance (22, 23). It is unlikely that PPV itself could cause duodenal obstruction because it is a thin-wall, low-pressure vessel (24). The indication for operation is often due to other concomitant anomalies like intestinal obstruction caused by malrotation, annular pancreas, duodenal diaphragm or biliary atresia. The presence of PPV causes technical difficulties at the time of upper abdominal surgery. Failure to recognize PPV during operation easily results in serious complications like thrombosis or profuse hemorrhage. Correct preoperative diagnosis of PPV is critical to avoid potential hazards during operation.

Azygos continuation of the IVC is a well-known vascular anomaly in association with polysplenia syndrome. Peoples et al (2) reported 92 (65%) out of the 142 patients with polysplenia presented this anomaly. The embryogenesis of the IVC was well described by Chuang et al (25). Useful signs on chest X-ray film of this IVC anomaly are convexity in the right tracheobronchial angle on the posteroanterior view and an absence of the IVC shadow on the lateral view as shown in the present patient (26). On CT examination, the enlarged azygos vein may resemble retroperitoneal lymphadenopathy or paravertebral mass. However, the azygos vein is easily identified when the tubular structure, the intense contrast enhancement to the same degree as the aorta and the continuity with the SVC through the azygos arch are seen (27).
Polysplenia syndrome has been considered to be a rare congenital anomaly. However, case reports of polysplenia syndrome and its associated anomalies appear to have increased with the advent of noninvasive imaging techniques such as CT and MRI. Most patients with this syndrome may be overlooked because they are asymptomatic, and thus polysplenia syndrome may be more frequent than is generally considered. Unawareness of this syndrome entails extensive investigations including angiography and even unnecessary thoracotomy because of its associated unusual anomalies. Physicians should be familiar with the spectrum of anomalies that occurs in patients with polysplenia syndrome.

Acknowledgements: The authors wish to thank Professor Dr. Kunio Hiwada of the Second Department of Internal Medicine, Ehime University School of Medicine, for his helpful suggestions for this manuscript.

References

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