A Case of Polysplenia Syndrome Presenting with Ascites

Asit ile Gelen Bir Polispleni Sendromu Olgusu

Sami FİDAN, MD, Msc,[a]
Murat ERKUT, MD, Msc,[a]
Adem DEMİREL, MD, Msc.,[b]
Ali AHMETOĞLU, MD, Prof.,[c]
Arif Mansur COŞAR, MD, Assis.Prof.,[a]
Orhan ÖZGÜR, MD, Prof.[a]

Departments of
[a]Gastroenterology,
[b]Internal Medicine,
[c]Radiology,
Karadeniz Technical University,
Faculty of Medicine,
Trabzon

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Yazışma Adresi/*Correspondence:*
Sami FİDAN, MD, Msc
Karadeniz Technical University
Faculty of Medicine,
Department of Gastroenterology,
Trabzon,
TÜRKİYE/TURKEY
fidansami@yahoo.com

ABSTRACT Polysplenia syndrome is a rare congenital anomaly frequently associated with various visceral anomalies. The most frequent manifestations of this syndrome, in addition to polysplenia are malrotation, congenital heart diseases and gastrointestinal, genitourinary and vascular abnormalities. In this report, we present an adult case of ascites due to prehepatic portal hypertension associated with portal cavernous transformation. The patient had a left-sided liver, right-sided multiple splenules and stomach, interrupted inferior vena cava with azygos continuation and extensive ascites. The portal vein was not observed and collateral vascular structures of cavernous transformation were identified in the hepatic hilus. Portal cavernous transformation with various congenital abnormalities has been reported in the literature; however, there are no cases of polysplenia syndrome presenting with ascites associated with portal cavernous transformation.

Key Words: Ascites; situs inversus; spleen

ÖZET Polispleni sendromu sıklıkla çeşitli organ anomalileri ile ilişkili nadir bir konjenital anomalidir. Polispleniye ek olarak bu sendromun en sık görülen belirtileri malrotasyon, konjenital kalp hastalıkları, genitoüriner, gastrointestinal ve vasküler anomalilerdir. Bu yazıda, portal kavernöz transformasyon ile ilişkili prehepatik portal hipertansiyona bağlı asit gelişmiş olan erişkin bir olgu sunulmuştur. Hastada sol taraflı karaciğer, sağ taraflı çok sayıda aksesuar dalak ve mide, azygos devamlılığı ile kesintiye uğramış inferior vena kava ve yaygın asit vardı. Portal ven gözlenmedi ve karaciğer hilusunda kavernöz transformasyonlu kollateral vasküler yapılar tespit edildi. Literatürde portal kavernöz transformasyon ile beraber çeşitli konjenital anomaliler mevcuttur, ancak asit ile gelen portal kavernöz transformasyon ile ilişkili polispleni sendromu olgusu henüz bildirilmemiştir.

Anahtar Kelimeler: Asit; situs inversus; dalak

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Polysplenia syndrome (PS) is a rare congenital abnormality frequently associated with various visceral abnormalities including multiple splenules, malrotation, congenital heart diseases and gastrointestinal, genitourinary and vascular abnormalities.[1,2] The incidence of polysplenia syndrome is reported as 1 in 20000 live births.[3] Here, we describe the first case of ascites due to prehepatic portal hypertension caused by portal cavernous transformation and abnormalities associated with PS.

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CASE REPORT

A 28-year-old female was admitted to our Gastroenterology Department with complaints of abdominal swelling and fatigue. Eleven years ago, when she was admitted to our clinic with complaints of abdominal swelling and diarrhea, the patient was finally diagnosed with ascites due to portal hypertension and urolithiasis. During that period, the patient was discharged upon her request while the medical evaluation was still continuing. The patient underwent a surgical operation in the urology clinic for a right renal stone at the same period. The patient's general condition was good; she was conscious, oriented and cooperative on the physical examination performed on her last visit. On admittance, the blood pressure was 100/70 mmHg, pulse was 76/min, body mass index was 23. She had no history of alcohol consumption. There were no abnormal physical findings, apart from pretibial edema and extensive ascites in the abdomen. The laboratory tests revealed the following: serum alanine aminotransferase 34 U/L (normal 5-40 U/L); albumin 2.9 g/dL (normal 3.4-4.8 g/dL); thyroid-stimulating hormone 1.14 μIU/mL (normal 0.27-4.2 μIU/mL); prothrombin time 14.6 seconds and negative viral hepatitis serology. Complete blood count and urine analysis were normal. Esophagogastroduodenoscopic findings were normal; esophageal or gastric varices were not detected. Colonoscopic exeamination revealed varices in the sigmoid colon and rectum. Examination of the paracentesis fluid demonstrated a leukocyte count of 300/uL (46% neutrophils); albumin: 0.4 g/dL; adenosine deaminase 5.2 U/L (normal 0-40 U/L) and Acid-Fast Bacilli smear was negative. Serum-ascites albumin gradient (SAAG) was 2.5 g/dL, consistent with ascites due to portal hypertension. Paracentesis culture was negative. Electrocardiographic findings were normal. Echocardiographic examination showed normal structure and localization of the heart. Cardiac index was measured as 2.9 L/min/m^2 (normal range: 2.8-4 L/min/m^2). Computerized tomography (CT) revealed that liver was located on the left and stomach was located on the right side of the abdomen, portal vein was not observed, collateral vascular structures of cavernous transformation were in the hepatic hilus, there was extensive ascites in the abdomen, and small spleens, 4x4 cm in diameter, were observed in the right side of the abdomen (Figure 1A). Dilatation was observed in the submucosal veins of the small intestine. The hepatic segment of the inferior vena cava was absent and the inferior vena cava was continuing with the azygos vein. The left kidney was atrophic, whereas the size of the right kidney was normal. Marked dilatation of the right renal pelvis and a renal stone, 1 cm in diameter was observed (Figure 1B). Hepatic venous blood was draining into the remnant of suprahepatic inferior vena cava. The dilatation of the azygos vein was shown on the thoracic tomography (Figure 1C). The patient rejected liver biopsy to detect the degree of liver damage. Medical treatment was scheduled to manage the ascites, and the patient was scheduled for follow-up.

DISCUSSION

PS generally manifests as a fatal condition during early childhood due to major cardiac abnormalities and biliary or intestinal atresia. It has been reported that one-half of the patients are dead by fourth month of age, whereas 25% of the patients live until the fifth year.[4] Patients who reach adulthood comprise only 5-10% of the cases.[4,5] The adult cases are generally asymptomatic during adulthood and diagnosed incidentally during radiological examination. Although a number of congenital abnormalities are observed in cases of PS, none of them are pathognomonic for the syndrome. Multiple spleens, interruption in the continuity of the vena cava inferior, and continuity with the azygos or hemiazygos veins are well-known abnormalities for this syndrome.[5-7] Double-lobe lungs in both hemithoraces and congenital cardiac anomalies are frequently encountered thoracic abnormalities. Genitourinary abnormalities, such as absence of the kidneys, double ureter and hypoplastic kidneys have rarely been reported.[5] On the other hand, gastrointestinal abnormalities observed in this syndrome are right-sided localization of the stomach and multiple spleens, left or mid-line localization of

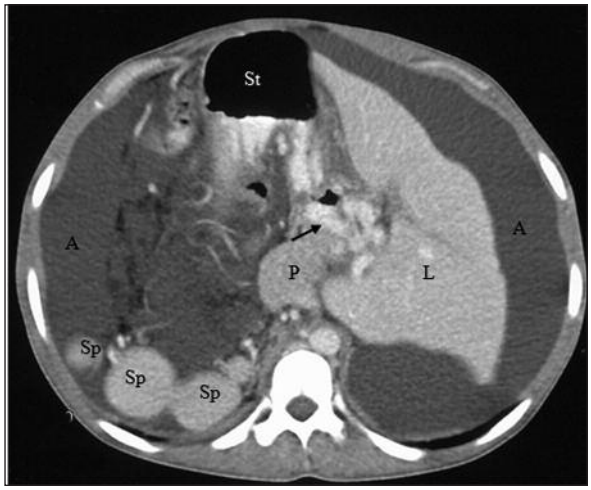

FIGURE 1A: The liver (L) was observed in the left side of the abdomen whereas the stomach (St), the pancreas (P), multipl spleens (Sp) and extensive ascites (A) were observed in the right side of the abdomen. Cavernous transformation was observed in the hepatic hilus (arrow) on the axial section of the computerized tomography.

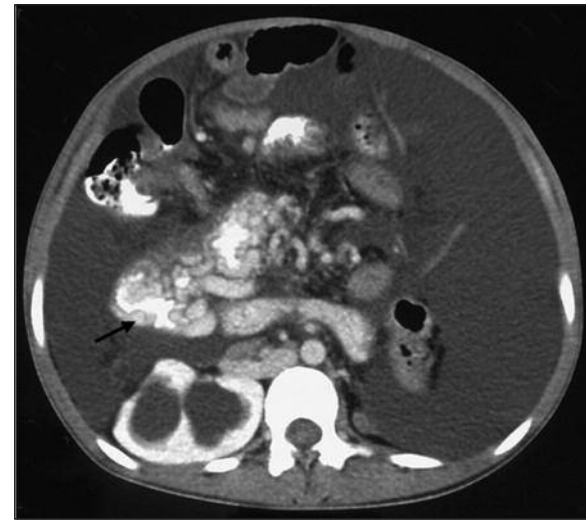

FIGURE 1B: Extensive ascites in the abdomen and dilatation of the submucosal veins in the small intestine (arrow).

the liver, rotation abnormalities of the small intestine and colon, short pancreas and vascular abnormalities.[1,2,5] Thoracic and abdominal abnormalities may contribute to the pathology in different degrees.

Interruption in the continuity of the inferior vena cava and continuity with the azygos vein, and preduodenal portal vein are well-known vascular anomalies of this syndrome.[2,4] Other vascular anomalies which are rarely described in cases of PS are transhepatic portal vein, hepatic artery originating from the superior mesenteric artery, circumaortic renal vein, portal vein hypoplasia or the absence of portal vein.[8-11]

The hepatic segment of the inferior vena cava was not observed in this case and the inferior vena cava demonstrated continuity with the dilated azygos vein. The dilated azygos vein results from the increased blood flow to the inferior vena cava. The portal vein was absent and collateral vascular structures of cavernous transformation were observed in the hepatic hilus. Congenital absence of the portal vein and its association with polysplenia is very rarely encountered in the literature.[11-13] Congenital absence of the portal vein leads to a complete interruption of mesentericoportal flow and development of a large portosystemic shunt, but not portal

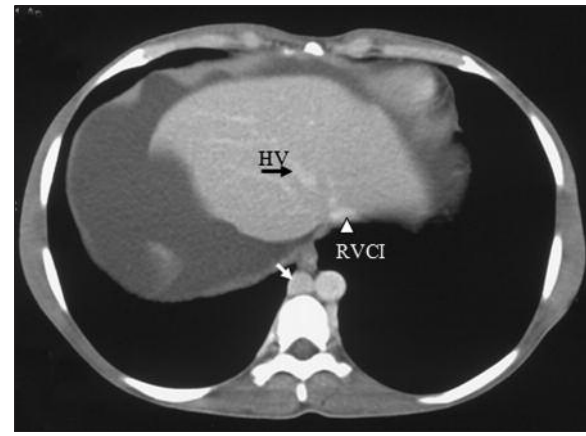

FIGURE 1C: Dilated azygos vein adjacentto the aorta was observed on the thoracic tomography (arrow), remnant of suprahepatic inferior vena cava (RVCI) (arrow head) and hepatic vein (HV) (black arrow). Hepatic venous blood drains into the remnant of suprahepatic inferior vena cava.

cavernoma.[14] In the cases with a large portosystemic shunt, there is no portal hypertension or ascites. Absence of portal vein might be due to postpartum portal venous thrombosis. Portal cavernoma develops in response to portal vein thrombosis in later life, with a partial but ineffective restoration of portomesenteric flow and prehepatic portal hypertension. On the long term, in portal vein thrombosis, the thrombosed portal vein shrinks and the remnant scar might feint to be clearly visible on CT, especially when abundant

collaterals develop. Therefore, there is presumably a portal vein thrombosis, but not congenital absence of the portal vein in this case. Angiographic imaging or the findings of the absence of the portal vascular structures in the liver biopsy may be useful for the differential diagnosis. Additionally, the patient has ascites, prolonged prothrombin time and hypoalbuminemia. Liver biopsy was planned in order to exclude chronic parenchymal liver disease. However, we could not perform the biopsy because of the patient's refusal.

Patient's left kidney was atrophic. Pelvicalyceal dilatation and renal calculi were detected in the right kidney; therefore angiographic imaging could not be performed due to the risk of renal toxicity.

Presence of cavernous transformation may result in the frequent development of symptoms of pre-hepatic portal hypertension since adequate drainage of the splenomesenteric blood flow could not be achieved. Patients manifest with hemorrhagefrom varicose veins, splenomegaly, portosystemic collaterals or ascites.[15,16] A case of portal cavernous transformation with various congenital abnormalities without polysplenia (and two cases of PS presented with ascites, the etiologies were intrauterine heart failure due to cardiac abnormalities and chylous ascites caused by intestinal malrotation) have been reported.[16-18] However, there are no cases of PS that present with ascites due to prehepatic portal hypertension caused by portal cavernous transformations in the literature.

In conclusion, PS is a very rare syndrome, in which the majority of cases die at an early age and the cases that reach adulthood are incidentally diagnosed during radiological examinations. It is also accompanied by various abnormalities that are not pathognomonic for the syndrome. Identification of the effects of these anomalies on various organ systems would be beneficial in explaining their pathologic processes on these systems.

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