

Coincidence of Polysplenia, Kartagener Syndrome, Dorsal Pancreas Agenesis, and Polycystic Kidney Disease in an Adult

Polispleni, Kartagener Sendromu, Dorsal Pankreas Aenezisi ve Polikistik Böbrek Hastalığı Birlikteliği

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ABSTRACT

Here we present the case of an adult male patient with the anomalies of polysplenia, Kartagener syndrome, dorsal pancreas agenesis, and adult polycystic kidney disease. Familiarity with this extremely rare coincidence may improve diagnostic accuracy and patient management.

Keywords: Polysplenia, Kartagener syndrome, dorsal pancreas agenesis, polycystic kidney disease

Öz

Polispleni, Kartagener sendromu, dorsal pankreas aenezisi ve polikistik böbrek hastalığı olan erişkin bir erkek hasta sunuyoruz. Bu çok nadir birlikteliğin iyi bilinmesi tanisal doğruluğu ve hasta tedavisini artırabilir.

Anahtar Kelimeler: Polispleni, Kartagener sendromu, dorsal pankreas aenezisi, polikistik böbrek hastalığı

Introduction

The term 'situs' refers to the position of the heart and viscera relative to the midline. Situs solitus indicates the normal position of the heart and abdominal viscera, which are asymmetric and lateralized. Situs inversus indicates the mirror-image position of the viscera relative to situs solitus. Situs inversus totalis (SIT) refers to the exact mirror image of the usual arrangement. There are multiple small accessory spleens, rather than a single, full-sized, normal spleen in polysplenia. Polysplenia may be associated with situs solitus, heterotaxy syndrome, or SIT [1, 2]. Polysplenia may sometimes occur together with dorsal pancreas agenesis or truncated/short pancreas and patients may also present with diabetes, pancreatitis, or obstructive jaundice [3, 4]. Kartagener's syndrome (KS) is a well-known entity characterized by paranasal sinusitis, bronchiectasis, and SIT. The coincidence of autosomal dominant polycystic kidney disease (ADPKD) and SIT is extremely rare, and only few pediatric cases have been reported [5, 6]. On the other hand, the coincidence of polysplenia and SIT is exceedingly uncommon [7].

This paper is the first presentation of the coincidence of polysplenia, Kartagener syndrome, short/truncated pancreas and polycystic kidney disease with their clinical significance in an adult patient.

Case Report

A 31-year-old male patient presented with headache, chronic cough, and purulent sputum. His medical history was notable for dextrocardia, frequent respiratory tract infections, and recently diagnosed diabetes. Physical examination revealed prehypertension (135/85 mmHg). Laboratory examinations showed an alanine aminotransferase (ALT) level of 65 IU/L (reference range <40 IU/L) and a serum uric acid level of 8.5 mg/dL (normal 1.8-5.4 mg/dL). A 24-h urine collection revealed microalbuminuria with a urinary albumin excretion rate of 50 mg/day. Chronic sinusitis (Figure 1a) and bronchiectasis (Figure 1b) were found on computed tomography (CT). Thoracic CT also revealed a right-sided heart, right-sided aortic arc, and right-sided descending aorta. A left trilobed lung and a right bilobed lung were also found. The liver and gallbladder were on the left side, whereas the spleen with double-layered appearance and stomach were on the right side (Figure 1c, 2). These findings were consistent with those of KS and polysplenia. A hypertrophied ventral pancreas was found in its mirror-image position, and the distal part of the pancreas



Figure 1. Kartagener syndrome and polysplenia. Coronal CT image of paranasal sinuses (a) reveals pansinus inflammatory disease. Axial chest CT image (b) reveals bronchiectasis that mainly affects the midportion of the left lung. (Coronal reformatted chest CT image (c) reveals situs inversus totalis with polysplenia. There are hyperarterial bronchus on the left side and eparterial bronchus on the right side (arrows).

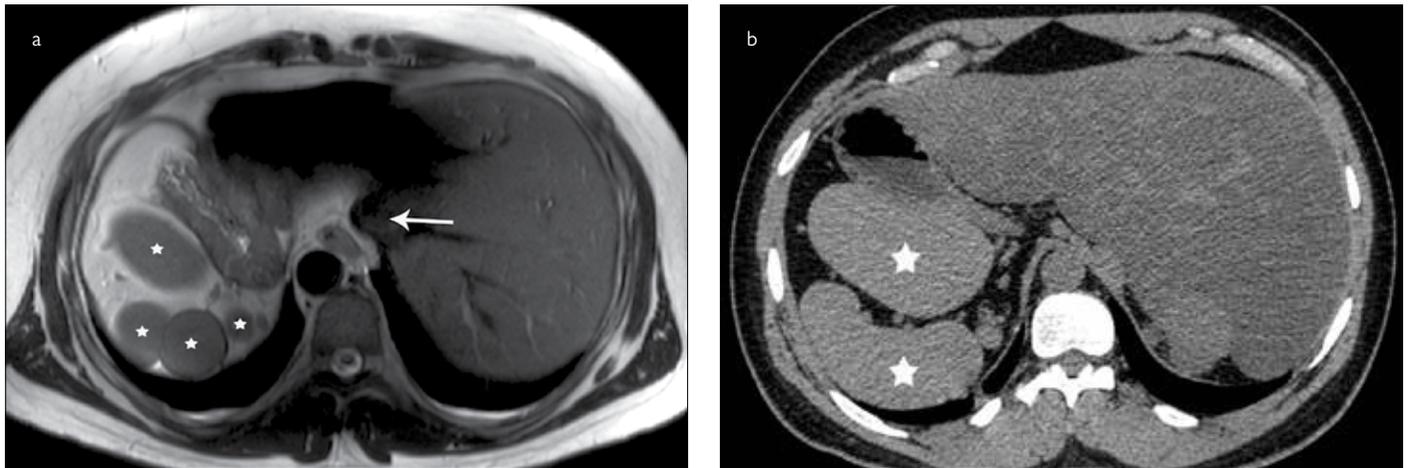


Figure 2. Polysplenia, double-layered spleen, and situs inversus totalis. Transverse T2-weighted MR image of the upper abdomen (a) reveals multiple spleens (stars) in the right upper quadrant behind the stomach and liver in the left upper quadrant with left-sided inferior vena cava (arrow). Unenhanced CT image (b) reveals two almost normal-sized spleens (stars) creating an interesting image, which we called double-layered spleen.

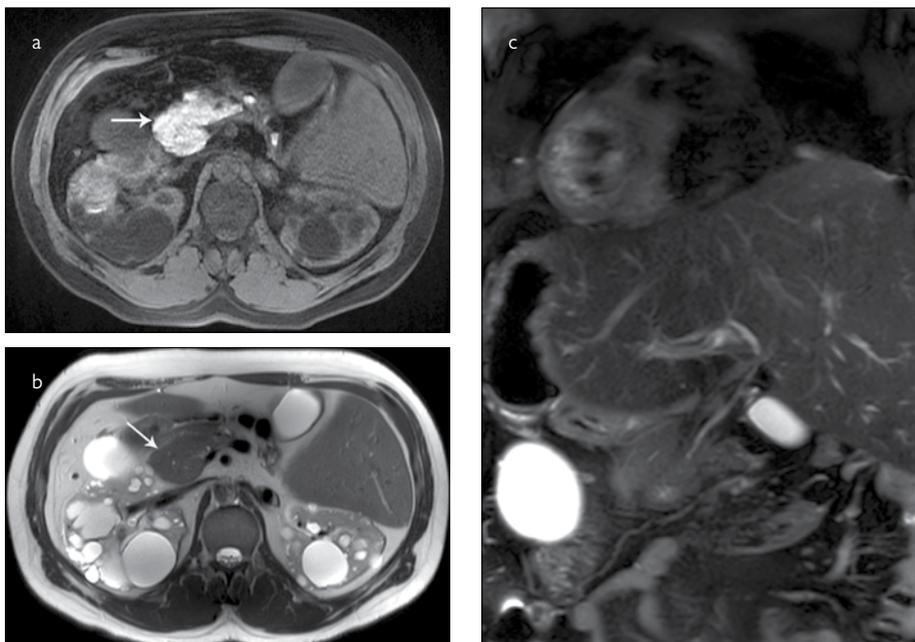


Figure 3. Dorsal pancreas agenesis, polycystic kidney disease, and situs inversus totalis. Transverse fat-saturated T1-weighted (a) and T2-weighted MR images (b) reveal a hypertrophied ventral pancreas in the mirror-image position (arrows). The distal part of the pancreas is not seen. The enlarged kidneys contain numerous cysts of variable sizes. Coronal fat-saturated T2-weighted MR image (c) reveals dextrocardia, left-sided choledochal duct, gallbladder, and liver associated with dorsal pancreas agenesis.

was not seen, consistent with dorsal pancreas agenesis or truncated/short pancreas. There were also enlarged kidneys with numerous cysts of variable sizes compatible with the findings of ADPKD (Figure 3).

Discussion

The presence of two or more spleens in a patient is called polysplenia. Polysplenia is a subtype of heterotaxy syndrome and occurs with bilateral left-sidedness (left isomerism). It is characterized by the duplication of the left-sided structures, including bilateral bilobed lungs concomitant with hyperarterial bronchi and bilateral left atria in addition to multiple spleens, not compatible with the presented case [8, 9]. However, polysplenia itself may be associated with various abnormalities such as situs inversus, pancreatic malformations, or vascular anomalies. These associations verify that polysplenia is a controversial and complex entity without pathognomonic features, although there are some embryonic hypotheses such as accelerated curvature of the embryonic body, genetic causes, and teratogenic factors. However, the exact cause of polysplenia remains unclear [10].

In this case, polysplenia was present on the left side along the greater curvature of the stomach with an interesting imaging appearance created by two spleens on axial images, which we called double-layered spleen. Generally, polysplenia includes one or two large spleens and several smaller ones with diameters of 1-6 cm. Several recent case reports describe the coincidence of situs inversus totalis (SIT) and polysplenia as rare [2, 11, 12]. Furthermore, the association of KS and ADPKD with polysplenia in an adult patient has never been reported yet. Ciliopathy is defined as any of a range of genetic disorders involving defects in the cilia or flagella of cells, and it is now recognized as a multisystem disease. Kartagener syndrome is a clinical variant of primary ciliary dyskinesia comprising a triad of situs inversus, bronchiectasis, and sinusitis. ADPKD is also an important subgroup of ciliopathies. It is expected that phenotypic parameters of ciliopathies may be used to recognize the cellular basis of a number of genetic disorders to facilitate the diagnosis and treatment of some diseases of unknown etiology and to determine the reasons of unusual associations [13].

The pancreas develops by fusion of the ventral and dorsal pancreatic buds, and splenic and pancreatic malformations develop embryonically from the dorsal bud. This clarifies why polysplenia and anomalies of the pancreas, particularly truncated/short pancreas or dorsal pancreas agenesis, tend to occur together [14].

Polysplenia and SIT are asymptomatic anomalies and are often incidentally detected in adults during imaging. In this case, symptoms of KS provided the early diagnosis of the anomalies. Dorsal pancreas agenesis may cause abdominal pain, pancreatitis, or diabetes [3, 15]. It has been hypothesized that diabetes may be caused by the abundant presence of β -cells in the agenic dorsal pancreas. An association of ADPKD may cause pain in the abdomen, renal infection, nephrolithiasis and renal colic, and hypertension. Up to 50% of patients with ADPKD require renal replacement therapy by age 60. Appendicitis, ureteropelvic junction obstruction, cholelithiasis, neoplasias such as hepatocellular carcinoma and

lymphoma, infarction of multiple spleens, splenic laceration, and splenomegaly may also be seen in SIT and polysplenia, which may create a confusing clinical picture [1].

Polysplenia, KS, dorsal pancreas agenesis, and ADPKD are rare congenital anomalies. Patients with these anomalies also present with respiratory system symptoms, prehypertension, and diabetes. To avoid misdiagnosis and misinterpretation of the diverse anomalies associated with polysplenia, proper recognition of all anomalies is crucial. A thorough and comprehensive evaluation of the whole body is essential to appreciate diverse abnormalities of this rare coincidence.

Informed Consent: Written informed consent was obtained from the patient who participated in this study.

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