Case Report / *Olgu Sunumu*

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Heterotaxy Polysplenia Syndrome Causing Intermittent Vomiting Due to Malrotation of the Duodenum in an Adult

Yetişkin Bir Hastada Duodenumun Malrotasyonuna Bağlı İntermitan Kusmaya Neden Olan Heterotaksi Polispleni Sendromu

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Abstract

Heterotaxy syndrome (situs ambiguus) is a rare condition in which the internal organs are abnormally arranged in the chest and abdomen. It occurs from an early embryological developmental disturbance with most cases being sporadic. In this report, we present a case of a 43-year-old female with heterotaxy syndrome who had anomalous right renal artery of high origin and preduodenal portal vein, anomalous orientation of the third portion of the duodenum causing intermittent symptoms and abnormal localization of intestinal and colonic segments. To the best of our knowledge, this the first report of association of these anomalies with heterotaxy syndrome.

Keywords: Heterotaxy, polysplenia, situs ambiguus

Öz

Heterotaksi sendromu (situs ambiguus), iç organların abdomen ve göğüs içerisinde anormal yerleşimi ile ilgili bir bozukluktur. Embriyonel dönemin erken gelişimsel bozukluğuna bağlı olup çoğu olgu sporadik olarak oluşur. Bu yazıda heterotaksi ile birlikte anormal yüksek orjinli sağ renal arter, preduodenal portal ven, intermitan semptomlara neden olan duodenumun üçüncü kısmının anormal oryantasyonu, intestinal ve kolonik segmentlerin anormal lokalizasyonu olan 43 yaşında bir kadın hastayı sunduk. Bilgilerimize göre, olgu heterotaksi sendromunda tanımlanan anomalilerin birlikteliğini gösteren ilk olgudur.

Anahtar Sözcükler: Heterotaksi, polispleni, situs ambiguus

Introduction

Heterotaxy is a rare condition in which the internal thoraco-abdominal organs and vascular structures show abnormal arrangement in the left-right axis of the body (1). As being the part of heterotaxy syndrome, polysplenia refers to presence multiple splenules without a parent spleen (2). The reported incidence of polysplenia is 1 per 250,000 live births (3). Embryologically the pancreatic bud and uncinate process arise from the ventral pancreatic bud whereas the dorsal pancreatic bud gives rise to the anterior head, body and tail. As the dorsal pancreas and spleen develop from the dorsal mesogastrium, coexisting anomalies of these structures can be expected (4). Although the precise etiology is not known, any pathology causing faulty formation of the portal vein from the vitelline veins leads to vascular anomalies such as preduodenal portal vein (PDPV) (5). We report a case of polyplenia syndrome with anomalous right renal artery of high origin, PDPV, anomalous orientation of the third portion of the duodenum causing intermittent symptoms and abnormal localization of intestinal and colonic segments with heterotaxy syndrome.

Case

A 43-year-old female with a 2-year history of intermittent epigastric pain and vomiting was referred to our clinic for the evaluation of suspected extrinsic compression of the second part of the duodenum detected on a gastroscopy procedure.

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She underwent a contrast-enhanced abdominal computed tomography (CT) which revealed several tiny accessory spleens in the left hypochondrium, PDPV, and a near-midline gallbladder (Figure 1,2). The head of the pancreas was visualized with the absence of the body and tail (short pancreas) (Figure 3). The right renal artery was originating from the abdominal aorta at the same level with superior mesenteric artery (Figure 4) and a hypoplastic hepatic artery was present. There was azygous continuation of the inferior vena cava (IVC) and median arcuate ligament compressing the celiac artery (Figure 5,6). The portal vein was running anteriorly to the duodenum, but the third portion of the duodenum was not crossing the midline; it was turning anteriorly over the second portion of the duodenum causing compression of the second part with continuation of small intestines on the right side whereas the colonic segments were detected on the left side (Figure 2,7).

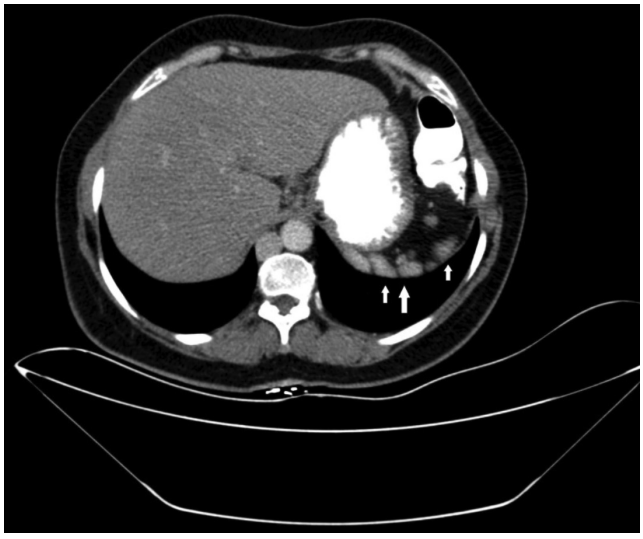

Figure 1. Several tiny accessory spleens in the left hypochondrium (white arrows) are seen on an abdominal contrast-enhanced CT image
CT: Computed tomography

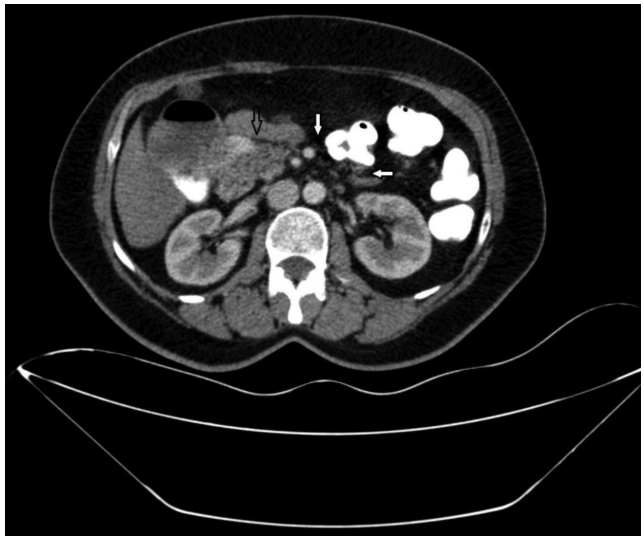

Figure 3. Axial images of abdominal contrast-enhanced CT showing short pancreas without body and tail
CT: Computed tomography

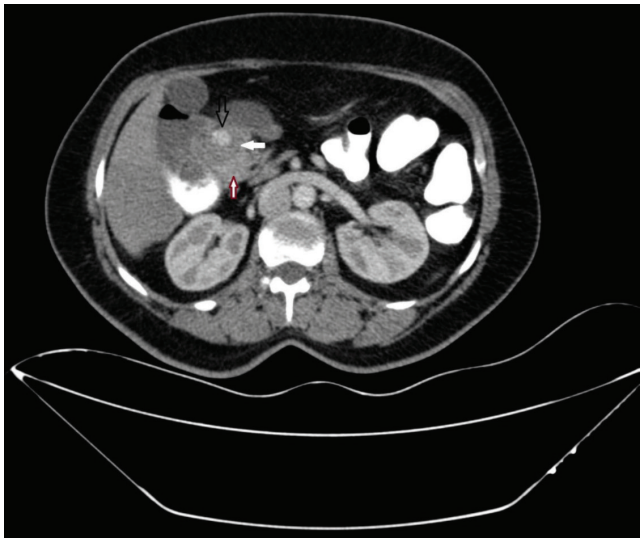

Figure 2. Contrast-enhanced CT of the abdomen shows a PDPV (black open arrow). Also, third portion of the duodenum (white solid arrow) is seen turning anteriorly over the second portion (red-lined arrow) instead of crossing the midline and causing compression of the second portion
CT: Computed tomography, PDPV: Preduodenal portal vein

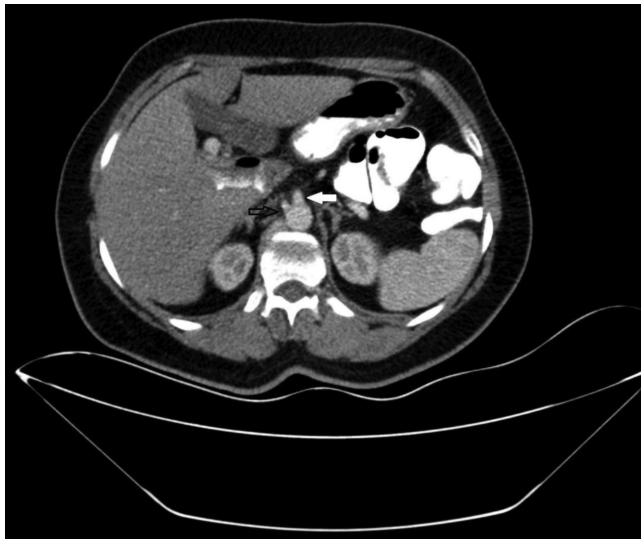

Figure 4. Abdominal contrast-enhanced CT showing right renal artery (black open arrow) originating from the abdominal aorta at the same level with superior mesenteric artery (solid white arrow)
CT: Computed tomography

Discussion

Heterotaxy syndrome or situs ambiguus is a disturbance in the usual left and right distribution of the thoracic and abdominal organs which does not entirely correspond to the situs solitus (normal symmetrical arrangement) or situs inversus (reversal or mirror-image of the normal arrangement). Rose at al. (6) reported that the incidence of situs ambiguous was one per 40,000 live births. Heterotaxy syndrome is classified into polysplenia (left isomerism, bilateral left sidedness) and asplenia (right isomerism) syndrome. There is a broad spectrum of anomalies in asplenia and polysplenia. Classical left isomerism (polysplenia) is described frequently with bilateral bilobed lungs with hyparterial bronchi and bilateral left atria in addition to the presence of multiple spleens/splenules while right isomerism (asplenia) is characterized by bilateral right atria, bilateral trilobed lungs, bilateral epiarterial bronchi and absence of spleen (7). Polysplenia is associated with cardiac anomalies which are less common in asplenia. Only 5-10% of patients with polysplenia syndrome reach adulthood without symptoms (8).

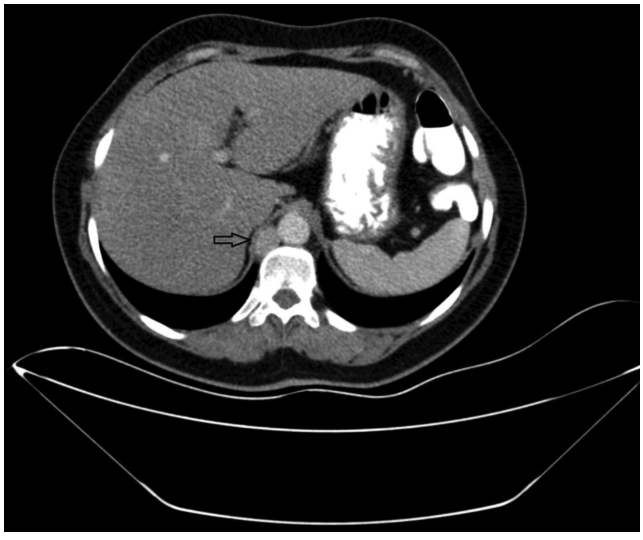

Figure 5. Azygos continuation of the IVC in the retrocrural area is seen on an abdominal contrast-enhanced CT image
IVC: Inferior vena cava, CT: Computed tomography

The spleen is considered to play an important role in normal development and lateralization of visceral organs. Embryologically spleen develops in the dorsal mesogastrium which also gives rise to dorsal pancreas (4). Thus, it is not surprising that any anomalies causing polysplenia may also cause developmental anomalies of the dorsal pancreas. Since it is asymptomatic mostly, polysplenia is often diagnosed incidentally on various other examinations and patients usually present with symptoms related to other coexisting pathologies (9). The present patient had polysplenia with various tiny accessory spleens 1-2 cm in diameter and complete agenesis of the dorsal pancreas presenting with absence of body and tail portions. Short pancreas forms because of developmental failure of the body and the tail and of the pancreas during the development stage of the pancreas (10).

Azygous continuation of IVC, which is a well-known vascular anomaly, was present in our case. Presence of azygous continuation of IVC was seen in 92 of 142 (65%) patients with polysplenia in a large-scale review (7).

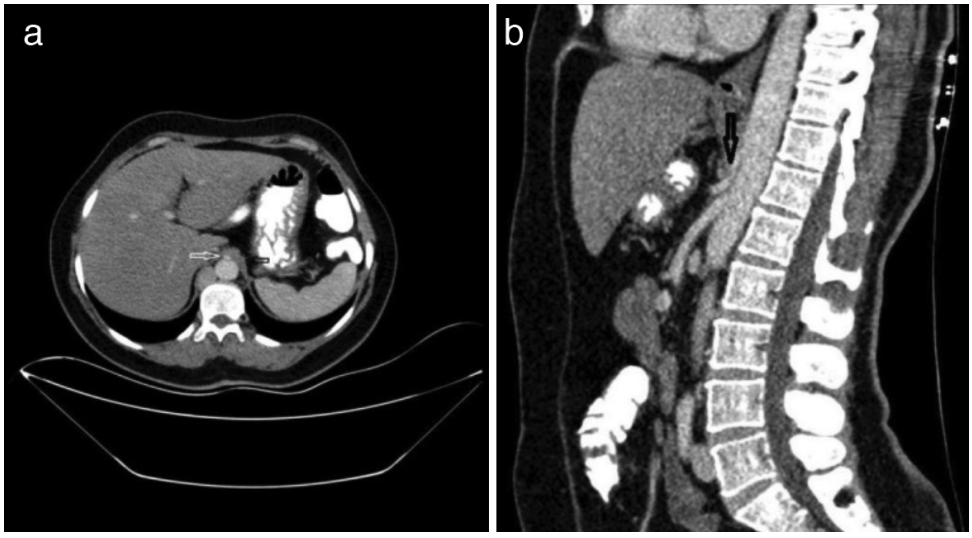

Figure 6a, b. Axial **(a)** and sagittal **(b)** images of contrast-enhanced CT of the abdomen shows median arcuate ligament compressing the celiac artery
CT: Computed tomography

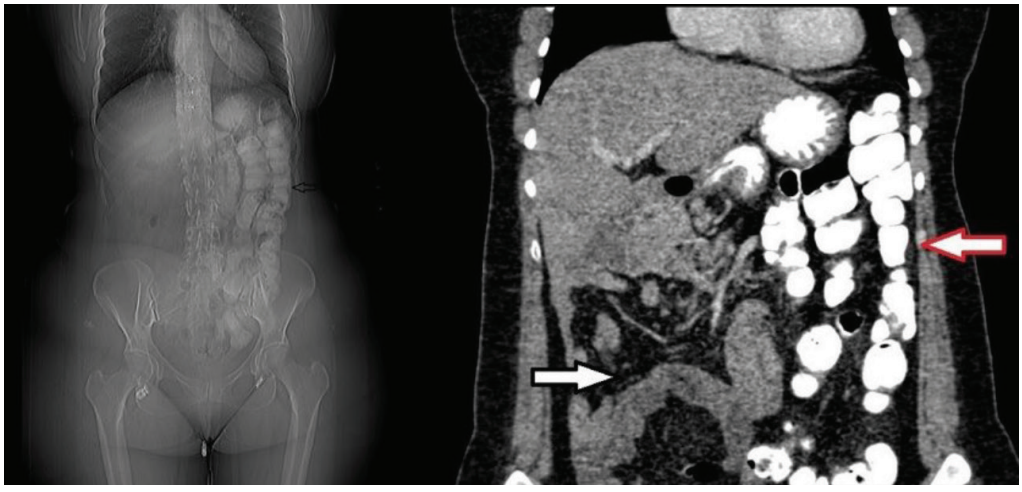

Figure 7. CT scenogram (left) and axial contrast-enhanced CT (right) images showing contrast enhanced colon segments on the left side of the abdomen and non-enhanced intestinal segments on the right side of the abdomen
CT: Computed tomography

A PDPV was detected in our patient on CT examination which is another common anomaly reported in the literature (11-13). PDPV first described by Knight in 1921, is a portal vein running anteriorly to the duodenum and head of pancreas. It might be associated with mechanical obstruction of the duodenum and related symptoms or might interfere with pancreatic development that gain importance during pancreaticobiliary operations (10,14).

In our case, the symptoms were not related to duodenal obstruction by a PDPV, but were due to intermittent obstruction of the second part of the duodenum by the third portion which abnormally turned anteriorly over the second portion.

In addition to vascular anomalies, such as azygos continuation of the IVC and PDPV, there existed an anomalous right renal artery of high origin from the abdominal aorta at the same level with the superior mesenteric artery, a median arcuate ligament compressing the celiac artery and a small caliper hypoplastic hepatic artery in our patient which were not reported previously in relation with heterotaxy and polysplenia syndromes with these combinations.

As mentioned, and studied before, malrotation of the gut is a part of spectrum of findings of heterotaxy syndromes (10). In our present case, in addition to malposition of the third part of the duodenum over second portion which was the cause of symptoms, small intestines were lying on the right side whereas the colonic segments were on the left side.

Whereas polysplenia syndrome usually remains asymptomatic, PDPV may present with obstructive symptoms, thus, the preferred treatment is duodeno-duodenostomy (15). Surgery was not performed in our patient since her symptoms were intermittent and were not due to mechanical obstruction of PDPV itself.

Conclusion

Polysplenia syndrome was considered to be a rare congenital anomaly, but by the advent of non-invasive imaging techniques, their incidence appeared to be increased. Radiologist should be alert about possible co-existing pathologies. Recognition of these anomalies is important in case of a need for surgery in order to prevent vascular damage or in gaining access to the target organ.

Authorship Contributions

Consept: T.S.C., B.K.Y. Design: T.S.C., B.K.Y., Y.S. Data Collection or Processing: T.S.C., B.K.Y. Analysis or Interpretation: T.S.C., B.K.Y., Y.S. Literature Search: T.S.C., B.K.Y. Writing: T.S.C., B.K.Y.

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