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CASE REPORT

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Clinically silent heterotaxy with polysplenia syndrome and IVC azygous continuation draining to SVC: CT findings. Case report

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Summary

Background: Patients with heterotaxy syndrome often have complex cardiac and extracardiac anomalies requiring further detailed diagnostic evaluation. They often present severe cardiac failure early in life. Newer radiological modalities in the form of spiral computed tomography (CT) and three-dimensional reconstruction of spiral CT allow clear definition of the anatomy of these anomalies.

Case report: A 59-year-old woman was diagnosed with polysplenia and multiple anomalies in an abdominal ultrasonography (US) during a control medical examination due to a trivial dietary mistake. She was then referred to our institution for further examination of these anomalies and an additional thoraco-abdominal computed tomography (CT) examination. The patient was totally asymptomatic at the time of admission. There was no significant past history and no abnormal laboratory data. We performed abdominal, pelvic and thoracic CT examinations using Somatom Siemens Emotion scanner. Non-enhanced sections were obtained with 8 mm scan collimation. Then, enhanced sections were obtained after administering a bolus of 120 ml contrast agent (Ultravist) with 5 mm scan collimation. Further multiple planar (MPR), volume rendering technique (VRT) and 3D reconstructions were performed in order to better visualize present abnormalities. Abdominal CT scans showed 5 spleens located in the right upper abdomen, 2 of them joined together in the form of a horseshoe with the latter 3 small splenules around. Liver was elongated and located in the central upper abdomen, with an enlarged left lobe. The dextraposition of stomach was found. Pancreas was short with hypoplastic tail. Furthermore, there was an abnormal passage of the IVC through the abdomen, which was interrupted at the level of diaphragm with azygous continuation in the thorax then joining the SVC, forming an arch and draining directly to the right-sided atrial appendage. No other anatomic anomalies, in particular those concerning the venous pulmonary return, lung lobation, rotation of the intestine, gastrourinary or central nervous system, often found in heterotaxy syndromes, were observed.

Conclusions: Most of the patients die at an early age because of an associated heart disease or other malformations. Cases of heterotaxy in adults or adolescents are rare and can be sometimes discovered incidentally. We report a case of an asymptomatic adult patient with the polysplenia type of the syndrome and uncommon systemic venous drainage for this type of heterotaxy. Our case confirms also the frequent coexistence of midline associated defects in polysplenia and other heterotaxy syndromes. In our case, female sex, dextraposition of the stomach, short and hypoplastic pancreas and right-sided polysplenia occurred together, but we did not observe any malrotation of intestines, nor clearly seen gallbladder or biliary pathologies.

Key words: heterotaxy • polysplenia • venous anomalies • inferior vena cava • azygous continuation • stomach dextraposition

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Background

Heterotaxy refers to spatial malposition of the organs or isomerism of naturally asymmetric organs. Its distinctive feature is *situs ambigus* as opposed to *situs inversus totalis* in which all the visceral organs are mirrored from their normal positions. Heterotaxy is often accompanied by numerous cardiac and retrocardiac anomalies [1]. Among the retrocardiac abnormalities, the most common are those within the spleen – polysplenia or asplenia. Other anomalies, that affect the pulmonary, digestive, urinogenital and central nervous system, have also been described. The coexistence of a complete visceral inversion, existence of double inferior vena cava (IVC), its azygous or hemiazygous continuation, abnormalities in pancreas and gallbladder, biliary atresia, have been observed [2, 3]. In order to the fact that most of the diagnosed congenital heart diseases are serious, heterotaxy syndrome is often recognized in early childhood. However, in literature there are descriptions of cases where the heterotaxy syndrome was diagnosed fortuitously in adults without complicated heart disease [4]. Yet, such cases are rare and do not exceed 5% of all heterotaxy patients.

When the inferior vena cava continues to azygous or hemiazygous vein, it often joins the IVC in order to drain together into the left atrium or isomeric left auricular appendage [5, 6].

In our work we present a case of IVC continuation to an azygous vein, which forms an arch by joining the superior vena cava, and drains to the right auricular appendage. In addition, the described patient had polysplenia, right-sided stomach, midline liver and shortened pancreas. The diagnosis was made accidentally to a healthy woman with no clinical symptoms. According to our knowledge, no form of such drainage of SVC to IVC accompanied by a combination of various clinically silent anomalies has been described in literature so far.

Case report

Abdominal ultrasonography during a control medical check-up for a trivial dietary mistake showed polysplenia with multiple visceral anomalies in a 59-year-old female patient. She was then referred to our Clinic for further examination, i.a. the thoraco-abdominal computed tomography (CT) examination. The patient was totally asymptomatic on the day of admission. There was no significant past history and laboratory results were normal.

The patient underwent abdominal, pelvic and thoracic CT examinations with the use of Somatom Siemens Emotion scanner. The scan collimation was 3 and 8 mm. Further multiple planar (MPR), volume rendering technique (VRT) and 3D reconstructions were carried out to better visualize the existing abnormalities.

The abdominal CT scan revealed 5 spleens located in the right upper abdomen, 2 of which joined together in shape of a horseshoe with 3 surrounding additional splenules (fig. 1, 2). The liver was elongated in transverse dimension, had an enlarged left lobe and was located in the central upper abdomen. Gallbladder situated in the median line and stomach on the right side of the abdomen, were revealed (fig. 3). Pancreas was short with a hypoplastic tail (fig. 4). Moreover, there was an abnormal passage of the IVC through the abdomen, which was interrupted at the level of diaphragm with azygous continuation in the thorax (fig. 5 A B) to finally join the SVC (fig. 6) by forming an arch and drain directly into the right auricular appendage (fig. 7). Other anatomic anomalies often found in heterotaxy syndrome, in particular concerning the arrangement of pulmonary veins, lung lobation, intestinal malrotation, digestive or central nervous system anomalies, were not observed in our patient.

Discussion

Polysplenia belongs to the spectrum of abdominal heterotaxy syndromes, in other words *situs ambiguous,* which separates *situs solitus* – normal position of the internal organs, from *situs inversus* – a complete mirror image of all abdominal and thoracic organs. The heterotaxy syndromes have been generally divided into asplenia and polysplenia. The latter takes various forms of clinical image. Its manifestations include a congenital heart disease, anomalies within digestive and urinogenital systems, morphological malformations of organs [1, 2]. The most characteristic

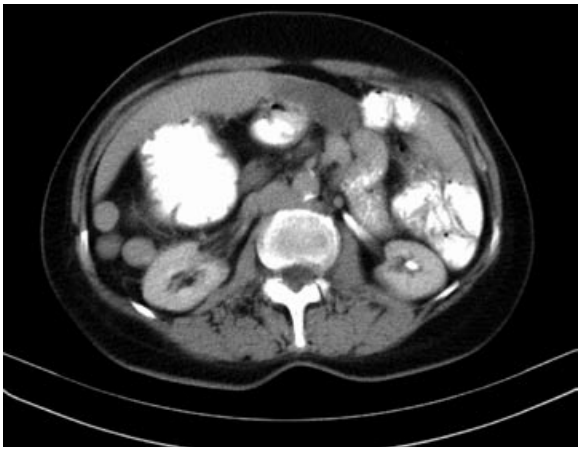

Figure 1. Three round soft tissue masses located in the right upper quadrant, anteriorly to the right kidney – splenules. Note also the dextraposition of the stomach and a midline gallbladder.

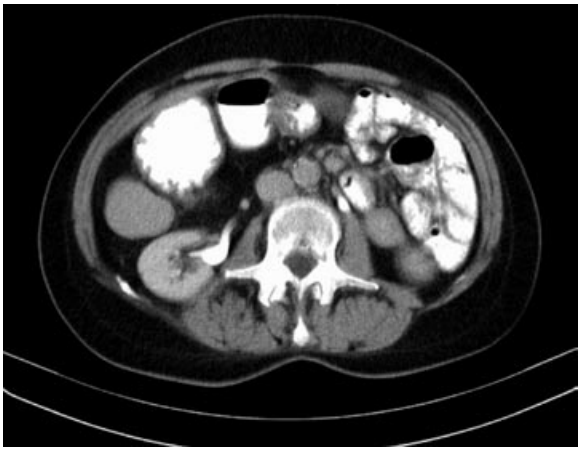

Figure 2. Lower scan of the abdomen. Another larger splenule is seen laterally and anteriorly to the right kidney at the level of its hilus.

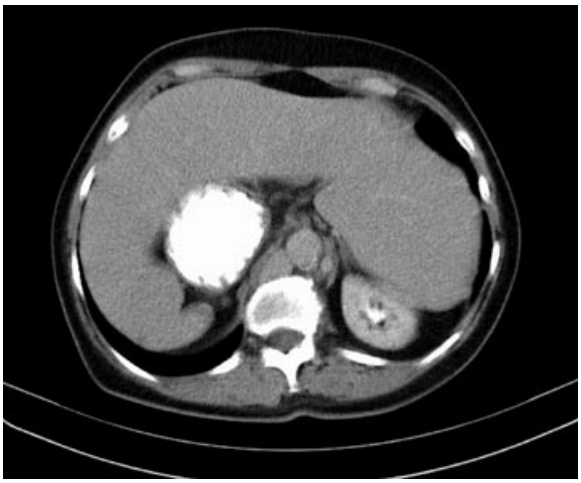

Figure 3. Right-sided stomach surrounded by a midline liver with an enlarged left lobe – a common anomaly in polysplenia syndrome.

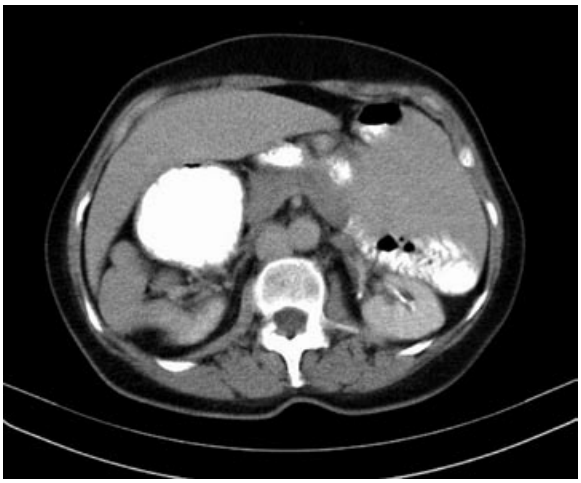

Figure 4. The horseshoe-shaped largest splenule located high in the upper right quadrant, posteriorly to the right hepatic lobe and right-sided stomach. Note also a short pancreas with the hypotrophic tail.

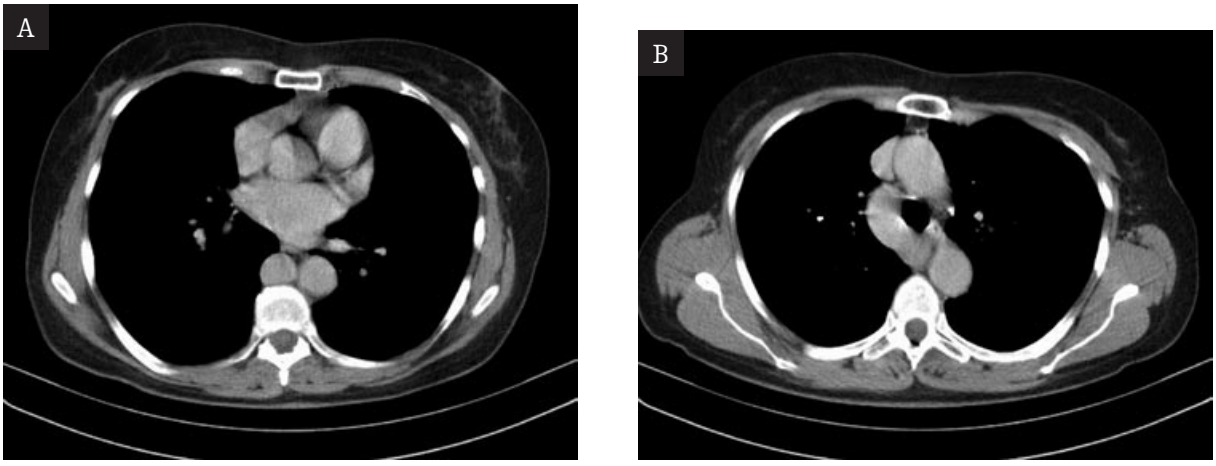

Figure 5. The intrathoracic part of an interrupted IVC on the right side of aorta (**A**) forming an arch to join the SVC (**B**).

feature of the syndrome is short IVC with azygous or hemiazygous continuation in the chest [5]. In children, the interrupted IVC usually drains to the left atrium. Most patients with heterotaxy die early in life in consequence of related heart diseases or other morphological malformations [1,7]. Cases of heterotaxy in adults and adolescents are rare and can be diagnosed incidentally [8–11]. In our study we present a case of clinically asymptomatic female patient who had polysplenia and venous arrangement unusual for this type of heterotaxy.

The most common abnormalities observed in patients with heterotaxy concern the heart. They include various intracardiac anomalies (e.g. common or hypoplastic ventricles or atria, common atrioventricular canal), heart malposition, malformed auricular appendages and arteriovenous connections [6]. The cardiac anomalies are less serious in adults and thus enable them to live to advanced age. In such cases abnormal venous connections are more frequent. One of the most common anomalies observed in adult patients is an interrupted IVC with azygous or hemiazygous continuation at the level of diaphragm. The interrupted IVC usually drains to isomeric auricular appendages, directly to the left atrium of the heart or to persistent left-sided superior vena cava [5]. In our patient we observed an interrupted IVC with its continuation in form of an azygous vein which drained to a normal SVC and then through a common arch into the right atrium. Such an anatomical anomaly is not typical for heterotaxy syndromes with polysplenia.

The presented case confirms also the presence of frequent abnormalities in the median body line which are related to polysplenia syndrome and other types of heterotaxy. Based on autopsy of 160 patients with heterotaxy, Boruch et al. corroborated a frequent relation of polysplenia with female sex, dextraposition of stomach, malrotation of intestines, absence of gallbladder, biliary atresia and abnormal morphology of the lungs [1]. In our case, the features included female sex, dextraposition of stomach, short and hypoplastic pancreas, multiple spleens located on the right side of abdominal cavity. We neither observed the malrotation of intestines nor any pathology of gallbladder or biliary ducts. In their work, Boruch et al. also prove that 92% of patients with heterotaxy syndrome have abnormal pulmonary morphology with two lobes located bilaterally in each lung, along with bronchi located below the pulmonary

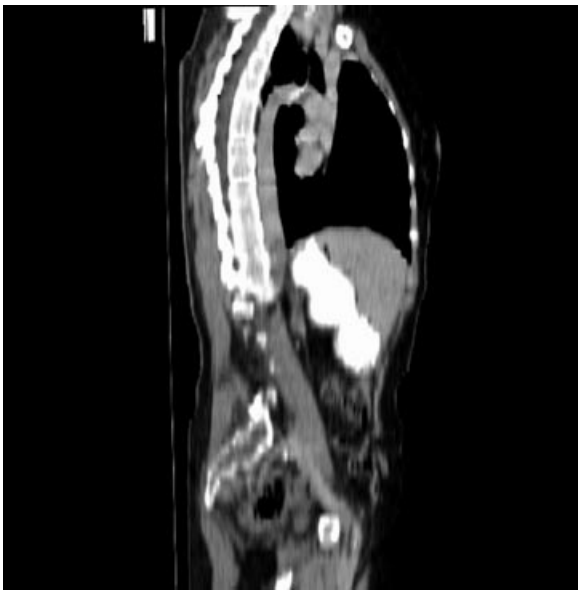

Figure 6. MPR reconstruction of interrupted IVC with azygous continuation – drainage to the SVC is well seen.

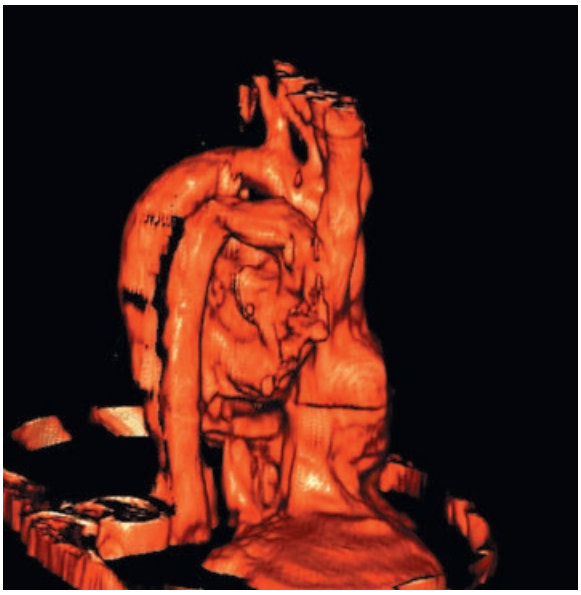

Figure 7. VRT reconstruction showing pericardial venous connection anomalies in our patient.

artery, and conclude that these features are found most frequently in patients with polysplenia [1]. We did not observe any of the morphological anomalies of the lungs in our case. Similarly, the inversion of atria is considered to be connected with the dextraposition of stomach in heterotaxy patients [1]. Although our case does not corroborate this hypothesis, it agrees with the results of Boruch et al. which indicate that most patients with polysplenia (78%) have more than one anomaly of the median line. Another interesting aspect of our case is the fact that the patient lived to advanced age with no clinical symptoms in spite of multiple congenital abnormalities in the median line, along with the fact that she was diagnosed fortuitously during a routine examination of the abdomen.

Conclusions

Certain anomalies in the median line (centrally located liver, multiple spleens and continuation of IVC in the azygous vein) can stimulate pathological processes. Therefore, the awareness of these abnormalities coexisting in one syndrome is necessary to avoid a misdiagnosis of several pathological processes. The computed tomography examination with secondary 3D reconstructions is a good method for visualizing and recognizing a wide spectrum of organ malpositions. Thus, high coincidence of related abnormalities in patients with heterotaxy imposes a comprehensive assessment and advanced diagnostics of such patients in case of finding one of the elements of the syndrome.

References:

1. Boruch S Ticho, Allan M Goldstein, Richard van Praagh. Extracardiac Anomalies in the Heterotaxy Syndromes With Focus on Anomalies of Midline-Associated Structures. Am J Cardiology 2000: 85 March, 729–734.
2. Takeshi K, Tsuyoshi T, Daisuke K et al. Anomalous portal branches associated with polysplenia syndrome: imaging findings. Comp Med Imaging Graph 2004: 28; 15–19.
3. Ruscazio M, Van Praagh S, Marass A et al. Interrupted inferior vena cava in asplenia syndrome and a review of the hereditary patterns of visceral situs abnormalities. Am J Cardiology 1998, 81 January 1.
4. Muneta S, Sakai S, Fukuda H et al. Polysplenia syndrome with various visceral anomalies in an adult: embryological and clinical considerations. Intern Med 1992; 31: 1026–31.
5. Hideki U, Siew Y, William AD et al. Atrial Appendages and venoatrial Connections in Hearts from Patients with Visceral Heterotaxy. Ann Thorac Surg 1995: 60; 561–9.
6. Shyh-lye C, Yiu-Wan L, Jou-Kou W et al. Usefulness of Electron Beam Computed Tomography in Children With Heterotaxy Syndrome. Am J Cardiology 1998: 81; 188–194.
7. Koonosuke N, Fumio K, Munechika W. Digestive Tracy Disorders Associated With Asplenia / Polysplenia Syndrome. J Pediat Surg 1997: 32, 1; 91–94.
8. Ito H, Ohgi S, Kanno T, Ishibashi T. Heterotaxy syndrome with pancreatic malrotation: CT features. Abdom Imaging. 2003 Nov-Dec; 28(6): 856–8.
9. Vanhoenacker FM, De Ruysscher D, De Backer AI, De Schepper AM. Heterotaxy syndrome in an adult, with polysplenia, visceral and cardiovascular malposition. JBR-BTR. 2001 Feb; 84(1): 1–4.
10. Abut E, Arman A, Guveli H, Bolukbas C, Kendir T, Dalay R, Ovunc O. Malposition of internal organs: a case of situs ambiguous anomaly in an adult. Turk J Gastroenterol. 2003 Jun; 14(2): 151–5.
11. Gayer G, Apter S, Jonas T, Amitai M, Zissin R, Sella T, Weiss P, Hertz M. Polysplenia syndrome detected in adulthood: report of eight cases and review of the literature. Abdom Imaging. 1999 Mar-Apr; 24 (2): 178–84.