

# Heterotaxy syndrome with accompanying azygos continuation of the inferior vena cava, patent ductus arteriosus and replaced common hepatic artery

Candan Güngör, Emrah Akay, Sena Yılmaz, Bahar Yanık

Department of Radiology, Balıkesir University  
Faculty of Medicine, Balıkesir, Turkey

## ORCID ID of the author(s)

CG: 0000-0003-1793-8823  
EA: 0000-0002-8586-3571  
SY: 0000-0002-1066-6585  
BY: 0000-0003-3358-7330

## Abstract

Abnormal anatomical organization of thoraco-abdominal visceral organs and vascular structures is called "Heterotaxy Syndrome." Heterotaxy is generally classified under two main headings as polysplenia and asplenia syndromes. Complex cardiac anomalies are closely associated with poor prognosis in the patient group with asplenia syndrome. On the other hand, cardiac anomalies are less common in polysplenic syndrome. It is particularly important to define the findings accurately in the planning of surgical and interventional treatment. We present the findings of a patent ductus arteriosus (PDA) patient with co-existent heterotaxy syndrome associated with abdominal anomalies, such as polysplenia, azygos continuation of interrupted inferior vena cava, liver with midline localization, short pancreas, preduodenal portal vein, replaced common hepatic artery (HA) originating from the superior mesenteric artery (SMA) and left sided superior mesenteric vein to the SMA. To the best of our knowledge, there is no previous report of HA originating from the SMA found together with PDA and heterotaxy syndrome.

**Keywords:** Heterotaxy, Polysplenia, PDA, Replaced HA, SMA, IVC interruption

## Introduction

Heterotaxy syndrome, also known as situs ambiguous, is a condition where the thoracoabdominal organs demonstrate abnormal arrangement across the left-right axis of the body [1]. Patients with heterotaxy syndrome have complex birth defects affecting the cardiac, respiratory, gastrointestinal, genitourinary and other systems [2]. The severity of heterotaxy syndrome varies depending on the specific abnormalities involved [1, 2]. High mortality rate is seen in patients with severe cardiac abnormalities while some patients have only mild health problems [1-3]. Radiological examinations are useful to accurately assess the abnormalities in patients with heterotaxy syndrome [4-7]. Herein, the CT findings of a 5-year-old patient who underwent radiological examination for suspicion of PDA and who was incidentally diagnosed with heterotaxy syndrome are presented.

## Corresponding Author

Candan Güngör  
Department of Radiology, Balıkesir University  
Faculty of Medicine, Balıkesir, Turkey  
E-mail: candan\_gungor@hotmail.com

## Informed Consent

The authors stated that the written consent was obtained from the parents of the patient presented with images in the study.

## Conflict of Interest

No conflict of interest was declared by the authors.

## Financial Disclosure

The authors declared that this study has received no financial support.

## Published

2021 October 26

Copyright © 2021 The Author(s)

Published by JOSAM

This is an open access article distributed under the terms of the Creative Commons Attribution-Non Commercial-NoDerivatives License 4.0 (CC BY-NC-ND 4.0) where it is permissible to download, share, remix, transform, and build upon the work provided it is properly cited. The work cannot be used commercially without permission from the journal.



**How to cite:** Güngör C, Akay E, Yılmaz S, Yanık B. Heterotaxy syndrome with accompanying azygos continuation of the inferior vena cava, patent ductus arteriosus and replaced common hepatic artery. J Surg Med. 2021;5(10):1072-1075.

### Case presentation

A five-year-old female patient was referred to the cardiology department of our hospital after recognizing a continuous murmur during cardiac auscultation in routine physical examination. In the echocardiographic examination, PDA was suspected. Contrast enhanced CT was performed for further evaluation. Late arterial images were obtained using a 64-slice CT scanner (Aquilion 64; Toshiba Medical System) with the following parameters: 80 kV tube voltage, 150-200 mA tube current, 1280.625 mm collimation, 0.67 mm section thickness, 1 mm increment, 0.2 mm/s table feed, 0.4 seconds rotation time. The patient was examined supine from suprasternal notch to the level of kidneys during free breathing, and a PDA with a diameter of 1.3 mm was detected (Figure 1 A-B). No anomaly was observed in the cardiac chambers and lung bronchi or lobes. The azygos vein was dilated in the posterior mediastinum and appeared to be the continuation of suprarenal IVC segment and drained into the superior vena cava (Interrupted IVC with azygos continuation) (Figure 2 A-B). The spleen was small in size with multiple adjacent accessory spleens (polysplenia) (Figure 3 A). The liver was located in the midline of abdomen and preduodenal portal vein (PDPV) was detected lying anterior to head of pancreas and duodenum (Figure 3 B). The pancreas was small in size. The common hepatic artery originated from the superior mesenteric artery (SMA) and the superior mesenteric vein (SMV) was observed on the left side of the SMA (Figure 4 A-B-C). In light of these findings, we diagnosed the patient with polysplenia syndrome. Written informed consent was obtained from the patient's father for the publication of this case report and tomography images.

Figure 1: Axial (A) and sagittal oblique (B) CT maximum intensity projection images. Patent ductus arteriosus, through which a slight passage of contrast material is observed (hollow arrow).

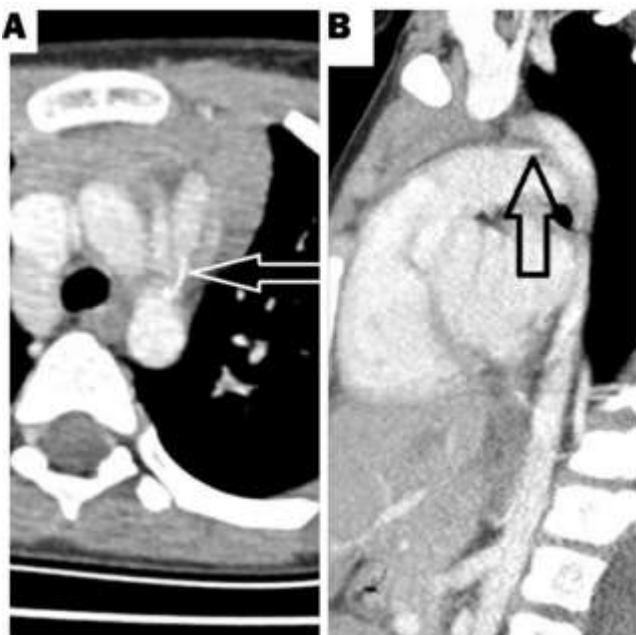


Figure 2: Sagittal reformat (A) and axial (B) CT images. Azygos continuation of interrupted inferior vena cava and opening superior to the vena cava.

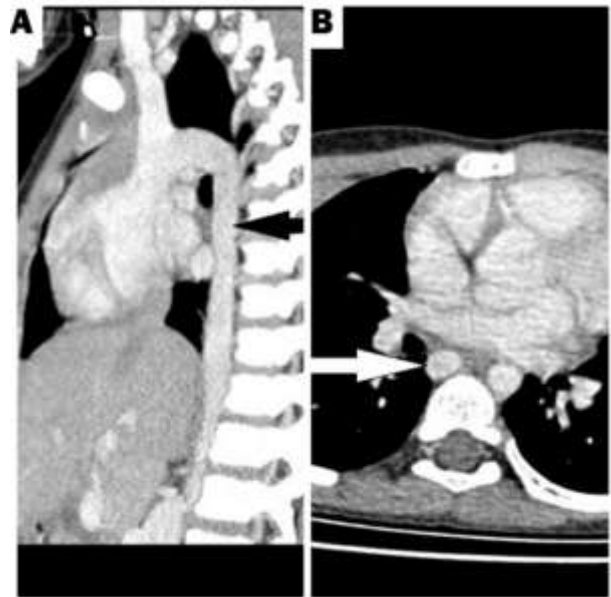


Figure 3: Axial CT images: A There are two nodular solid formations with similar density to the spleen indicating polysplenia. B. The falciform ligament and the liver is located in the midline (hollow arrow). Preduodenal portal vein is shown (solid arrow).

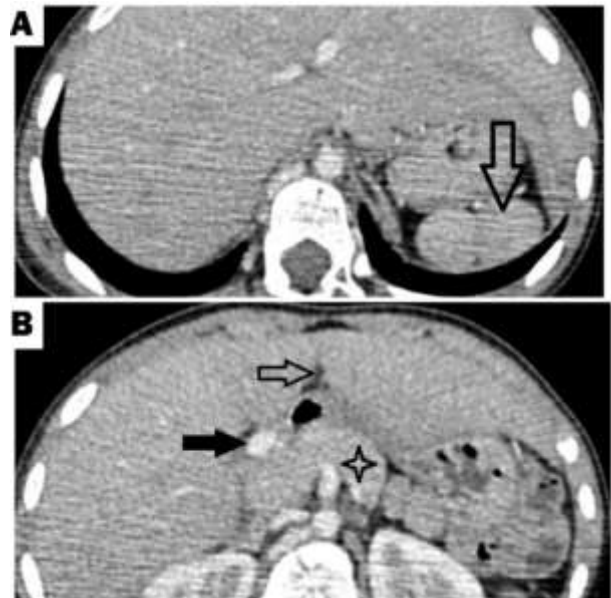
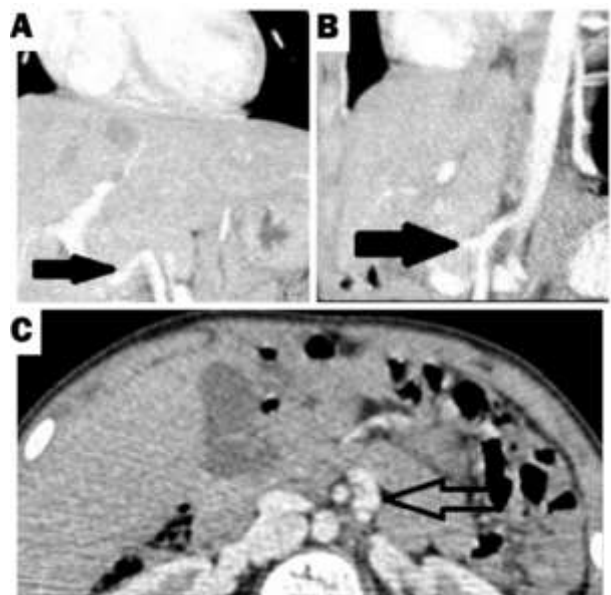


Figure 4: Coronal (A), sagittal (B) and axial (C) CT images. Hepatic artery arises from the superior mesenteric artery (solid arrow) and the superior mesenteric vein is located on the left side of superior mesenteric artery (hollow arrow).



## Discussion

The exact cause of heterotaxy syndrome is unknown. It is thought to occur due to disruption of left-right axis determination in the early embryonic period. A single pathognomonic anomaly was not described for this rare entity. Recent studies have shown that more than 80 genes are required for normal asymmetric left-right organ development [3]. In general, there is a confusing classic method that classifies heterotaxy into two major syndromes, polysplenia (left isomerism) and asplenia (right isomerism) syndromes, where there are too many combinations of possible malformations, which is still a matter of debate today [1, 3, 8]. It is recommended to use the definition of "heterotaxy syndrome" instead and to describe associated findings afterwards [9]. However, this case will be discussed over the classical classification of heterotaxy syndrome. Our case had polysplenia without definite isomerism.

Polysplenia syndrome is a complex entity with a broad spectrum of abnormalities of thoracic and abdominal organs. The incidence of polysplenia syndrome is reported as 1 per 250,000 live births [3]. It is the condition in which there are multiple (2 to 16) spleens congenitally. There may be multiple spleens on the right or left side or in the midline, according to severity of the primary defect in lateralization. One of the other abdominal features in the polysplenia syndrome is a midline liver. This is a finding diagnosed in about half of patients with polysplenia [10]. In our case, the liver of the patient is located close to the midline. PDPV, stomach malposition, dorsal pancreatic agenesis, annular pancreas, short pancreas, duodenal atresia, biliary atresia, malrotation, and mobile cecum are among other abdominal findings diagnosed in cases with polysplenia [4, 9, 11-13]. In our case, while the stomach was located in the normal position, a short pancreas and PDPV were identified. Polysplenia syndrome with a variation of HA and the SMA is often diagnosed incidentally in patients by CT [14]. A limited number of articles were published in the literature, in which the HA originates from the SMA with heterotaxy syndrome, as in our case [3, 14-16]. In addition, the SMV was located on the left side of SMA, which is a suspicious finding in terms of intestinal malrotation. However, our patient had no abdominal complaints. Thus, a whole abdominal CT examination was not performed.

Polysplenia syndrome involves bilateral bilobed lungs, bilateral hyperarterial bronchi and bilateral left atria in the thorax [1, 3]. Tracheobronchial structures, pulmonary vascularity and lungs segmental anatomy were normal in our patient. One of the common findings is azygos continuation of interrupted IVC [8]. As in our case, because of the developmental interruption of the suprarenal IVC segment, venous blood in the lower extremity flows into the superior vena cava (SVC) through azygos. Cardiovascular congenital anomalies such as atrial and ventricular septal defects, PDA, absent coronary sinus, abnormal location of the cardiac apex, common atrioventricular canal pulmonic stenosis, pulmonary atresia, an anomalous pulmonary venous connection, and an anomalous systemic venous return were reported in association with this syndrome [2, 17]. While no intracardiac pathology or main thoracic vascular anomalies were diagnosed in our case, the presence of PDA was confirmed. Peoples et al. [8] reported that 45 of the 127 autopsied cases of

polysplenia had accompanying PDA. To the best of our knowledge, this is the first case report concerning the association between PDA, replaced common hepatic artery and heterotaxy syndrome.

Enhanced multislice CT is excellent radiological method for determining heterotaxy syndrome. Especially arterial and venous phase dynamic CT imaging series facilitate the identification of vascular and cardiac anomalies. Moreover, multiplanar and three-dimensional reconstructions can define congenital anomalies to an even greater advantage [4, 5, 7]. Technical factors can be adjusted to minimize the radiation dose to children. Ultrasonography may demonstrate abdominal anomalies such as midline localization liver, and multiple spleens. However, abnormally localized hollow organs can be mistakenly evaluated as masses [4]. MR imaging is an alternative method for detection of these anomalies [6]. Nevertheless, we did not perform additional MR imaging as our patient's family did not give their consent on the sedation process required for MR imaging of their 5-year-old child.

Heterotaxy syndromes with severe cardiac abnormalities are less frequent in adults because of the high mortality during early stages of life. In adult patients, heterotaxy syndrome is usually detected incidentally during radiologic examination or vascular interventions. It is important for surgeons or radiologists to detect and know these anomalies and anatomical characteristics before conducting surgical and interventional procedures [2, 14]. Thus, possible complications such as hemorrhage, vascular ligation, and organ injury may be prevented.

## Conclusion

Heterotaxy syndromes encompass of a wide range of abnormalities affecting the thoracoabdominal organs and vascular structures. It has no characteristic radiological pattern, or typical laboratory findings. Since cardiovascular pathologies can have fatal complications, patients with a congenital heart disease (ASD, VSD, PDA, etc.) or those with an atypical position of the organs should be carefully examined in terms of other anomalies that make up the heterotaxy spectrum. Accurate diagnosis is also essential for proper planning of surgical and interventional procedures without the risk of vital organ injury.

## References

- Kim SJ. Heterotaxy syndrome. *Korean Circ J*. 2011 May;41(5):227-32.
- Lagrotta G, Moises M. Heterotaxy Polysplenia Syndrome in Adulthood: Focused Review and a Case Report. *Cureus*. 2020 Jan 30;12(1):e6822.
- Rameshbabu CS, Gupta KK, Qasim M, Gupta OP. Heterotaxy polysplenia syndrome in an adult with unique vascular anomalies: case report with review of literature. *J Radiol Case Rep* 2015;9(7):22.
- Gayer G, Apter S, Jonas T, Amitai M, Zissin R, Sella T, et al. Polysplenia syndrome detected in adulthood: report of eight cases and review of the literature. *Abdom Imaging*. 1999 Mar-Apr;24(2):178-84.
- Wolla CD, Hlavacek AM, Schoepf UJ, Bucher AM, Chowdhury S. Cardiovascular manifestations of heterotaxy and related situs abnormalities assessed with CT angiography. *J Cardiovasc Comput Tomogr*. 2013 Nov-Dec;7(6):408-16.
- Murat SN, Yalcinkaya D, Yarlioglu M, Yigit H, Bozkurt U, Celik IE, et al. Diagnosis of Heterotaxy Syndrome in a Patient With Multiple Congenital Cardiac Malformations Using Magnetic Resonance Imaging. *Circ Cardiovasc Imaging*. 2020 Sep;13(9):e010307.
- Burton EC, Olson M, Rooper L. Defects in laterality with emphasis on heterotaxy syndromes with azygos continuation, patent ductus arteriosus, and aortic branches variations: a case report. *J Thorac Dis*. 2018 Feb;10(2):E138-E141.
- Peoples WM, Moller JH, Edwards JE. Polysplenia: a review of 146 cases. *Pediatr Cardiol*. 1983 Apr-Jun;4(2):129-37.
- Applegate KE, Goske MJ, Pierce G, Murphy D. Situs revisited: imaging of the heterotaxy syndrome. *Radiographics*. 1999 Jul-Aug;19(4):837-52;discussion853-4.
- Burton EC, Olson M, Rooper L. Defects in laterality with emphasis on heterotaxy syndromes with azygos continuation, patent ductus arteriosus, and aortic branches variations: a case report. *J Thorac Dis*. 2018 Feb;10(2):E138-E141.
- Fulcher AS, Turner MA. Abdominal manifestations of situs anomalies in adults. *Radiographics*. 2002 Nov-Dec;22(6):1439-56.
- Tawfik AM, Batouty NM, Zaky MM, Eladally MA, Elmokadem AH. Polysplenia syndrome: a review of the relationship with viscerocranial situs and the spectrum of extra-cardiac anomalies. *Surg Radiol Anat*. 2013 Oct;35(8):647-53.
- Emekli E, Gündoğdu E, Kebapçı M. Coincidence of left isomerism, malposition of cecum, dorsal pancreatic agenesis, and retroaortic left renal vein: A case report. *J Surg Med*. 2019; 3(11):812-814.

14. Liu J, Yang K, Wang J. Polysplenia syndrome with a rare variation between the common hepatic artery and the superior mesenteric artery in adults. *Arch Med Sci.* 2020 Aug 10;16(5):1263-1266.
15. Tsutsumi R, Nagata Y, Enjoji A, Ohno Y, Kamito H, Kanematsu T. Situs Ambiguus with Gastric Cancer: Report of a Case. *Surgery Today*, 2006;37:676-9.
16. De Wailly P, Metzler P, Sautot-Vial N, Olivier D, Meunier B, Faure JP. Pre-duodenal portal vein in polysplenia syndrome: clinical effects and surgical application. *Surg Radiol Anat.* 2011 Jul;33(5):451-4.
17. Celik M, Celik T, Iyisoy A, Guler A. An unusual combination of congenital anomalies in an adult patient: patent ductus arteriosus, Kommerell's diverticulum with aberrant right subclavian artery, and heterotaxy syndrome. *Hellenic J Cardiol.* 2011 Sep-Oct;52(5):469-72.

This paper has been checked for language accuracy by JOSAM editors.

The National Library of Medicine (NLM) citation style guide has been used in this paper.