ANNALES
UNIVERSITATIS MARIAE CURIE-SKŁODOWSKA
LUBLIN – POLONIA
VOL. LXII, N 1, 43 SECTIO D 2007

1st Department of Radiology, Department of Human Anatomy, Department of Cardiology
Medical University of Lublin

ELŻBIETA CZEKAJSKA-CHEHAB, GRZEGORZ J. STAŚKIEWICZ, ANDRZEJ TOMASZEWSKI, KAMIL TORRES, ANNA TORRES, SEBASTIAN UHLIG, ANDRZEJ DROP

Visceral heterotaxy syndrome with polysplenia diagnosed with ECG-gated multislice computed tomography – a case report

Situs ambiguus, or visceral heterotaxy is a rare congenital condition, resulting from disorders of right-left symmetry. Typically, heterotaxy is accompanied by disorders of morphology of the spleen, with two basic types: asplenia or polysplenia. Both these conditions are accompanied by typical syndromes of additional anomalies. The paper deals with a case of a patient with polysplenia diagnosed with multislice computed tomography (MSCT).

CASE REPORT

P.M., 22-year-old male was referred to the 1st Department of Radiology for MSCT cardiac examination because of anomalous vascular structure visualized in superior mediastinum on echocardiography. Medical history of the patient included episodic arrhytmias. The ECG gated MSCT examination was performed using standard cardiac CT protocol with 64-row LightSpeed scanner (GE Medical Systems). The series of scans were reconstructed at every 10% of ECG R-R segment in 5–95% option. Multiplanar and 3D reconstructions were used to visualize heart and extracardiac structures. Scanning range included epigastric region.

The MSCT examination findings showed that anomalous vascular structure diagnosed with echocardiography was widened hemiazygos vein. Abdominal findings were: polysplenia, midline liver (Fig. 1), short pancreas (Fig. 2) and interruption of the inferior vena cava (IVC) with hemiazygos continuation to persistent right-sided superior *vena cava* (Fig. 1, 3). Concomitant thoracic anomalies were: high origin of coronary arteries from distal parts of aortic sinuses, bicuspid aortic valve (Fig. 4) and additional fissures of the lower lobes of both lungs (Fig. 5). Basing on the findings the patient was diagnosed of heterotaxy–polysplenia syndrome.

DISCUSSION

Situs anomalies are rarely diagnosed in adults. Typically they are identified in children, in cases of severe congenital heart diseases. *Situs ambiguus* is a rare condition (1/6,000–1/20,000 births (7), where a disturbed symmetry of internal organs is observed, opposed to *situs solitus* (normal position of internal organs) or *situs inversus* (mirror image of situs solitus, with normal relation of organs of

the same side). *Situs ambiguus*, or heterotaxy is typically accompanied by polysplenia or asplenia, with typical sets of concomitant anomalies in both conditions. Heterotaxy–polysplenia syndrome is accompanied by congenital heart diseases less common (50–90%) than heterotaxy with asplenia (congenital heart disease in 99% cases) (6).

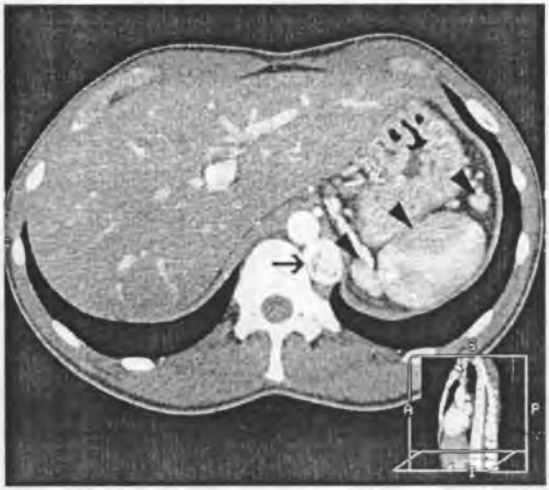

Fig. 1. Axial scan of epigastrium. Arrow – widened hemiazygos vein, arrowheads – multiple spleens. Enlarged left lobe of liver as indicator of disturbed right-left symmetry. Interruption of inferior *vena cava* – lack of intrahepatic portion

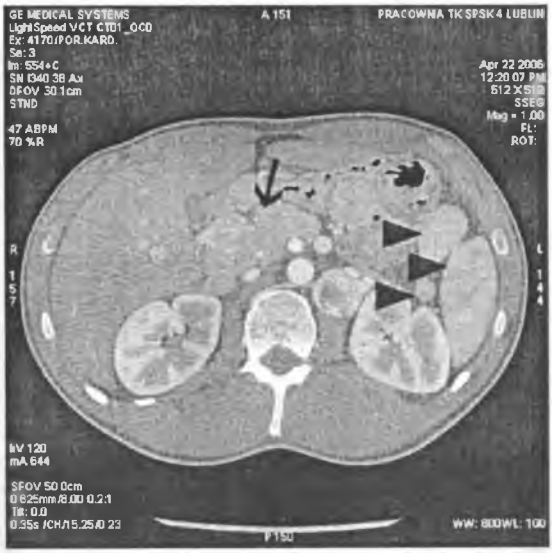

Fig. 2. Axial scan of epigastrium. Arrow – short pancreas: hypoplasia of tail, arrowheads – multiple spleens

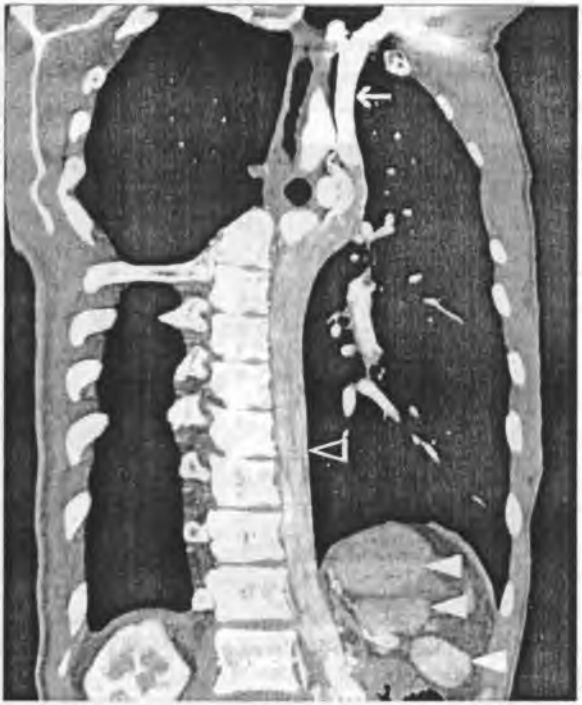

Fig. 3. Multiplanar curved line reformation of thorax. Arrow – persistent right superior *vena cava*, blank arrowhead – hemiazygos vein, arrowheads – multiple spleens

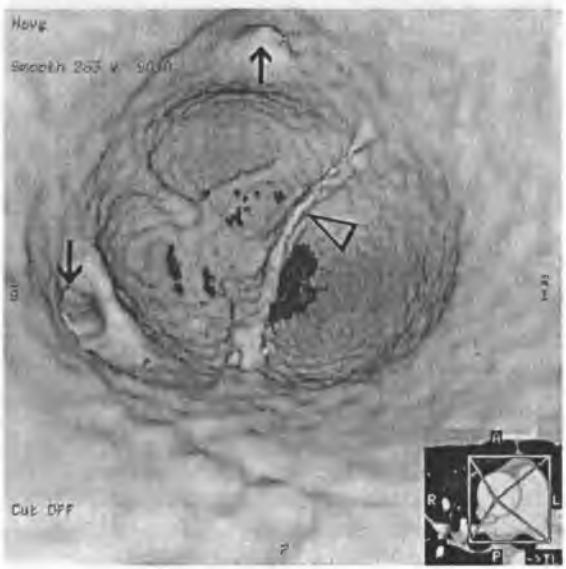

Fig. 4. Virtual angioscopy of aortic valve. Arrowhead – bicuspid aortic valve, arrows – high origin of coronary arteries from distal parts of aortic sinuses

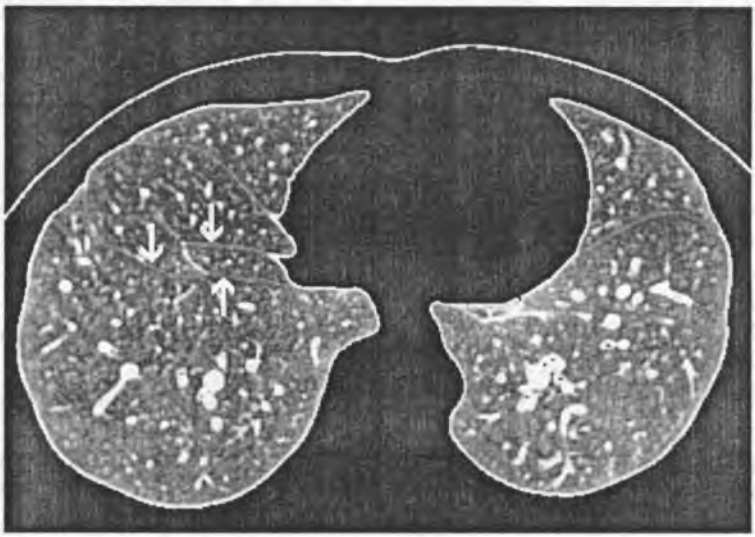

Fig. 5. Adjusted pulmonary window, axial view. Arrows – additional fissures of the lower right lobe

Heterotaxy-polysplenia syndrome, also defined as left isomerism or bilateral left-sidedness (3) is related with midline or ambiguus position of abdominal organs and multiple spleens. Typical anomalies of abdominal organs include midline liver (50–67% of cases), interrupted IVC with azygos continuation (58–100%), anomalies of vertebrae, congenital short bowel, atresia of duodenum, hypoplasia of pancreas. Thoracic anomalies typical of heterotaxy–polysplenia syndrome include bilateraly bilobar lungs (72–88%), duplication of superior *vena cava* and cardiac anomalies: common atrium (25–30%), hypoplasia or absence of one ventricle (37%), and ventricular outflow tract anomalies (2).

Heterotaxy–polysplenia syndrome is a complex anomaly, with no single anomaly pathognomonic for this condition (8). In the presented case, multiple features of this sydrome were identified with MSCT. In the patient, both thoracic and abdominal anomalies typical of heterotaxy–polysplenia syndrome were identified. Cardiac features of the syndrome were slightly expressed in the patient, however, with medical history of arrhytmia, he remains under ambulatory cardiological care. Tendency to heart arrhytmias is common in patients. with heterotaxy–polysplenia syndrome. Complete heart block is found in 22% of them, and, when accompanied by the structural defect of the heart, is the main cause of interuterine death in this group (1).

Etiology of heterotaxy syndromes remains unclear. Some researchers suggest its relation to pregnancy diabetes (4), however most popular theories explain the heterotaxy with single gene mutations, with recent interest in PA26 (6q21) gene (5).

The presented case shows that heterotaxy–polysplenia syndrome may remain undiagnosed until adulthood. Because of typical sets of anomalies related to both polysplenia and asplenia, each of those conditions should be thoroughly diagnosed. In the presented case, MSCT examination allowed a reliable identification of the syndrome, and ECG-gating used for cardiac protocol was particularly useful for identification of cardiac anomalies.

REFERENCES

1. Atkinson D. E., Drant S.: Diagnosis of heterotaxy syndrome by fetal echocardiography. Am. J. Cardiol., 82, 1147, 1998.
2. Bartram U. et al.: Heterotaxy Syndrome – Asplenia and polysplenia as indicators of visceral malposition and complex congenital heart disease. Biol. Neonate, 88, 278, 2005.
3. Fulcher A. S., Turner M. A.: Abdominal manifestations of situs anomalies in adults. Radio Graphics, 22, 1439, 2002.
4. Lin A. E. et al.: Heterotaxy: Associated conditions and hospital-based prevalence in newborns. Genet. Med., 2, 157, 2000.
5. Peeters H. et al.: PA26 is a candidate gene for heterotaxia in humans: identification of a novel PA26-related gene family in human and mouse. Hum. Genet., 112, 573, 2003.
6. Peoples W. M. et al.: Polysplenia: a review of 146 cases. Pediatr. Cardiol., 4, 129, 1983.
7. Tonkin I. L.: The Definition of Cardiac Malpositions with Echocardiography and Computed Tomography. W. F. Friedman, C. B. Higgins (ed.): Pediatric Cardiac Imaging. Saunders, 157, Philadelphia 1984.
8. Winer-Muram H. T., Tonkin I. L. D.: The spectrum of heterotaxic syndromes. Radiol. Clin. North Am., 27, 1147, 1989.

SUMMARY

Heterotaxy–polysplenia syndrome is one of rare anomalies of right-left symmetry of internal organs. In patients with the syndrome, polysplenia is accompanied by multiple anomalies of thoracic and abdominal organs, including congenital heart defects, bilobar lungs and widening of azygos vein resulting from interruption of inferior *vena cava*. Abdominal manifestations include right-or left-sided multiple spleens, bilateral liver and short pancreas. Heterotaxy–polysplenia syndrome is rarely diagnosed in adults, as typically congenital heart anomalies result with early signs and symptoms. The paper presents a case of 22 years old male in whom the heterotaxy–polysplenia syndrome was diagnosed with multislice computed tomography. Retrospective ECG-gating allowed a reliable visualization of accompanying cardiac anomalies.

Zespół heterotaksji–polisplenii rozpoznany w wielorzędowej tomografii komputerowej z bramkowaniem EKG – opis przypadku

Zespół heterotaksji-polisplenii jest rzadkim zaburzeniem symetrii narządów wewnętrznych. W zespole tym prawo- lub lewostronnej polisplenii towarzyszą typowo liczne zaburzenia położenia i budowy narządów wewnętrznych klatki piersiowej i jamy brzusznej, obejmujące wrodzone wady serca, obustronnie dwupłatowe płuca i poszerzenie żyły nieparzystej, wynikające z przerwania ciągłości żyły głównej dolnej, powiększenie lewego płata wątroby oraz hipoplazja ogona trzustki. Zespół heterotaksji–polisplenii jest nieczęsto rozpoznawany w wieku dorosłym. Często występujące wady serca powodują zwykle wystąpienie objawów w okresie dziecięcym. W pracy przedstawiono przypadek 22-letniego mężczyzny, u którego rozpoznanie zespołu heterotaksji–polisplenii postawiono w badaniu wielorzędowej tomografii komputerowej z bramkowaniem EKG, wykonanym po stwierdzeniu w badaniu echokardiograficznym nieprawidłowej struktury naczyniowej w śródpiersiu górnym. Dzięki zastosowaniu bramkowania EKG badanie umożliwiło rozpoznanie cech zespołu polisplenii–heterotaksji nie tylko w obrębie narządów jamy brzusznej i płuc, lecz również anomalii struktur serca.