Heterotaxia: radiological and surgical observations in a case of polysplenic syndrome

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Riassunto

Gli Autori descrivono il caso di una paziente asintomatica portatrice di una sindrome polisplenica diagnosticata mediante immagini ultrasonografiche, radiografiche e di risonanza magnetica, e confermata all'atto chirurgico. Una malformazione della milza è frequentemente associata al quadro dell'eterotassia ed ad altre malformazioni di organi toracici ed addominali, ma non può essere evidenziata nessuna caratteristica patognomonica legata a tale condizione. Nel caso descritto, le indagini radiologiche hanno evidenziato la presenza di una milza polilobulata in una sede atipica, al di sotto del fegato, associata al malposizionamento del fondo gastrico e della giunzione gastroesofagea, con interruzione della vena cava inferiore ed in assenza di malformazioni congenite cardiache. In conclusione, la sindrome polisplenica rappresenta una rara condizione clinica la cui diagnosi è spesso occasionale perché asintomatica nell'adulto. L'utilizzo di più metodi radiologici permette di distinguere la presenza di masse patologiche in sede mediastinica piuttosto che addominale e di determinare l'esatta anatomia delle strutture coinvolte onde poter programmare in maniera idonea un eventuale atto chirurgico, qualora dovesse rendersi necessario.

Parole chiave: eterotassia, polisplenia, vena cava inferiore

Summary

Heterotaxia: radiological and surgical observations in a case of polysplenic syndrome.
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We describe a case of asymptomatic polysplenic syndrome as suggested by ultrasonography, gastrointestinal series, computed tomography, magnetic resonance imaging scans and arteriography, and confirmed at surgery. Spleen malformation is frequently associated with heterotaxia and other malformations of the thoracic and abdominal organs, but no pathognomonic features can be detected in relation to this condition. In our patient, imaging studies revealed the presence of a polylobulated spleen in an atypical position

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below the liver, associated with malpositioning of the gastric fundus and gastro-oesophageal junction with interruption of the inferior vena cava, but no congenital heart disease. In conclusion, polysplenic syndrome is a rare clinical condition, occasionally found in asymptomatic adults. Radiological detection could be mistaken for mediastinal or abdominal pathological masses, and only a thorough study performed with several different radiological methods can determine the precise anatomy of the structures involved so as to be able to plan surgery where necessary.

Key words: heterotaxia, polysplenia, inferior vena cava.

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Introduction

The heterotropy or heterotaxia syndrome is an abnormal position of organs and great vessels[1], probably due to the abnormal rotation of some anatomical structures in a direction different from that of the principal axis, or to their position on the side opposite to the one normally occupied.

In this case the new position is different from that in the situs solitus or in the situs inversus[2], and is defined as "situs ambiguus", where normally asymmetric structures can become symmetric[1-3].

Heterotropy is a rare genetic syndrome with autosomal recessive inheritance, characterised by an incidence of 1-5/10000[4]. It is a polymorphic syndrome, since there is no single pathognomonic abnormality.

One form of heterotaxy is isomerism characterised by a symmetric position of the abdominal and chest organs, associated with congenital abnormalities. Usually, during embryogenesis, development of the hemithorax is asymmetric: in isomerism, the two hemithoraxes have a mirror image and the right or left hemisomus arrangement can prevail[1-4,5].

Right isomerism, also defined as "asplenic syndrome", is more common in male patients and occurs with severe cyanosis, transposition of the great vessels, pulmonary stenosis or atresia and systemic venous abnormalities, splenic agenesia and a transverse liver. Most of these patients die during their first year of life[6].

Left isomerism, also defined as "polysplenic syndrome", is more common in females and is generally characterised by the presence of several spleens or a single lobulated spleen, with a less severe clinical picture and prognosis, in that the patients are sometimes asymptomatic and therefore have a better chance of survival[6].

The case reported here is that of an asymptomatic female patient with a polysplenic syndrome, which could only be diagnosed with the aid of imaging studies.

Case report

A 51-year-old female patient was admitted to our hospital for elective umbilical hernia. She had undergone appendectomy when she was 29 years old. Routine laboratory examination findings were normal .

During hospitalisation, following an acute abdomen episode with colic pain and bilious emesis, routine radiological studies were performed. Chest X-rays did not reveal any abnormality. A plain film of the abdomen showed a gastric bubble under the diaphragm, left colic flexure and intestinal loops in their normal sites and radiopaque gallstones.

Abdominal ultrasonography revealed a midline liver and two smooth masses located between the liver and the right kidney, with rounded margins, moderately hypoechoic as compared to the liver parenchyma and with a diameter not exceeding 3-4 cm. (Fig. 1).

The spleen was not evident in its typical location and the pancreas could not be explored because of meteorism.

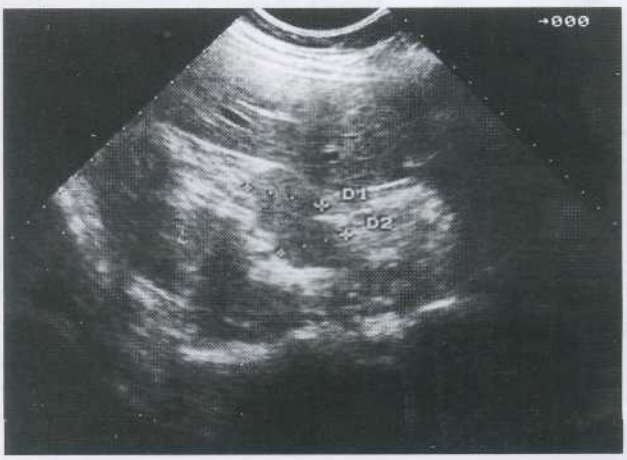

Fig. 1. Abdominal ultrsonography showing two splenules under the liver parenchyma.

In view of these findings, other examinations were required, consisting in a contrast study of the gastrointestinal tract, computed tomography and magnetic resonance imaging.

The contrast study of the gastrointestinal tract confirmed the malpositioning of the gastro-oesophageal junction and of the superior gastric portion; neither oesophageal hiatal hernia nor gastro-oesophageal reflux were detected. The duodenum formed its normal backward "C", and the duodenojejunal junction was in its typical site (Fig. 2).

Computed topography (CT) and magnetic resonance imaging (MRI) confirmed the presence of a smooth textured mass below the liver with rounded margins, which was interpreted as the spleen in an atypical location, and an abnormal vascular pedicle with correct positioning of the splenic artery and vein, the latter regularly draining into the superior mesenteric vein (Fig. 3).

Both imaging methods showed a considerable dilation of the azygous vein (azygous continuation), a dilated right diaphragmatic vein and the absence of the inferior vena cava with independent suprahepatic veins draining directly into the right atrium.

The correct position of the gastric fundus was confirmed.

Since the patient was also scheduled for cholecystectomy, angiography was performed in order to detect other possible vascular malformations. It showed a common hepatic artery originating from the superior mesenteric artery , the absence of the inferior vena cava with an enlarged azygous vein and the coeliac trunk with the left gastric and splenic arteries running together to the right side.

Cholecystectomy revealed a single lobulated spleen with its vessels between the superior pole of the right kidney and the liver, while the gastro-oesophageal junction was found in its normal position.

Discussion

In addition to multiple splenules (2 to 10) located along the greater gastric curvature, various types of abnormalities can be observed in polysplenic syndrome: partial situs inversus, cardiovascular malformations (dextrocardia, defects in the atrial or ventricular septum and absence of the coronary sinus), interruption of the inferior vena cava with collateral drainage via an azygous or hemiazygous vein and independent drainage of the hepatic vein into the right atrium, left chest isomerism with bilateral bilobated lungs, absence of a gallbladder, biliar atresia, a transverse liver, and an annular pancreas or lack of its caudal portion[1,2,4,7,8].

The complex malformations occurring in this

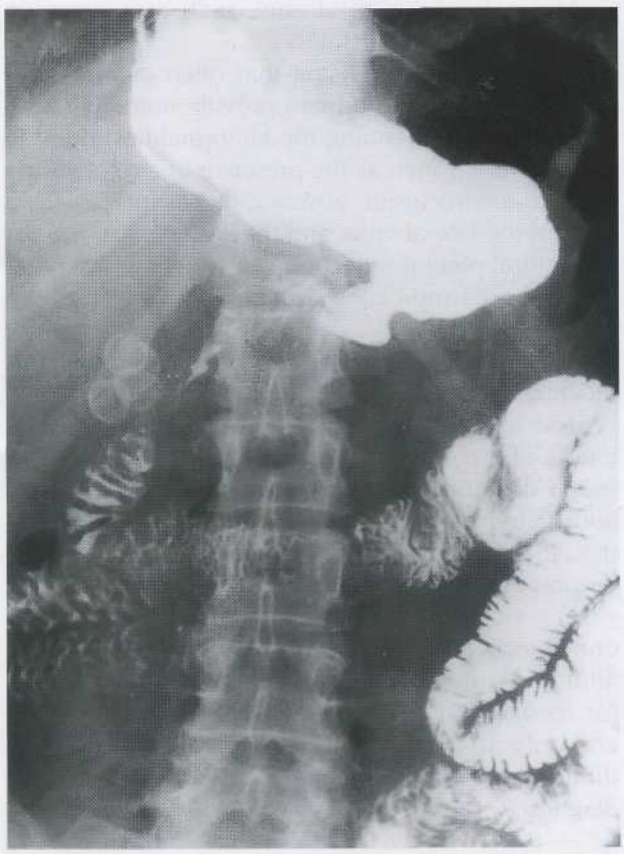

Fig. 2. Contrast study of the gastrointestinal tract demonstrating malpositioning of the gastro-oesophageal junction and of the superior gastric portion and stones in the gallbladder.

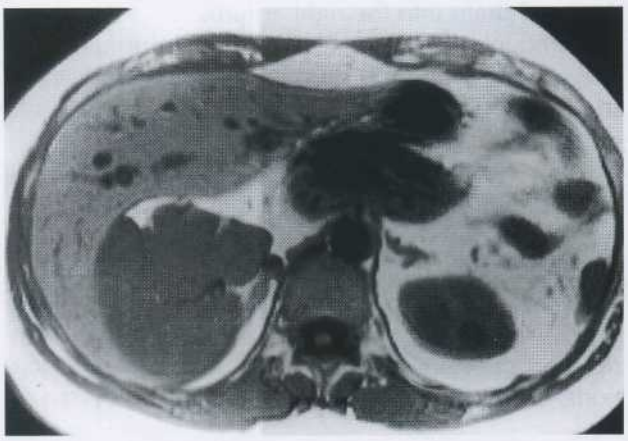

Fig. 3. T1-weighted MR image of the upper abdomen showing a right-sided spleen with its own vessels under the liver and a retrocrural dilated azygos vein.

syndrome are the result of development abnormalities during the embryonal period.

The fact that heart and intestinal rotations occur during the 4th week of the embryonal period explains the frequent coexistence of both cardiovascular and abdominal abnormalities. Total or partial lack of rotation causes the abnormal position of the anatomical structures[9].

During the 5th week, the spleen develops from a mesodermic proliferation between the two leaves of the dorsal mesogastrium. With the formation of the lesser peritoneal sac, the dorsal part of the mesogastrium, between the spleen and the dorsal midline, fuses with the posterior abdominal wall, while the residual portion of the mesogastrium forms the splenorenal ligament, where the vessels fixing the spleen in the upper left abdominal quadrant run[9].

Usually, during the 4th-5th week, the stomach rotates clockwise to the left, while the liver rotates to the right; in these cases the stomach rotates counterclockwise, thus determining the abnormal position of the organs in the mesogastrium, as in the case reported here[10].

As far as the inferior vena cava malformations are concerned, it should be noted that during embryogenesis the inferior vena cava is composed of five segments, each of different origin[4]; at times, some of the segments of the inferior vena cava either fail to form or fail to connect in the usual manner.

The lack of a hepatic-subcardial anastomosis causes the absence of the hepatic segment of the inferior vena cava; thus, the blood returns from the lower part of the body to the right atrium through the azygous and hemiazygous veins and the superior vena cava. The suprahepatic veins are independent and drain into the right atrium.

Although the presence of all the abnormalities in polysplenic syndrome constitutes a very characteristic condition, there is no single pathognomonic abnormality[1].

The left chest isomerism, the situs ambiguus of the abdominal organs, the absence of the hepatic segment of the inferior vena cava and the multiple spleens strongly suggest a polysplenic syndrome, but their concomitant presence is not found in all patients. Moreover, unlike the asplenic syndrome, there is no constant association with heart malformations, and even when these are present, they are often only minor heart defects, such as dextrocardia or septal defects[5].

For these reasons polysplenic syndrome can be detected in asymptomatic adults as an incidental finding during evaluation for other disorders, as in the case in point[1].

In actual fact no cardiovascular malformations worthy of note were found in our patient. CT, MRI and ultrasonography can provide accurate information about chest morphology, abdominal organs and their anatomical relationships. All morphological abnormalities, malpositionings and vascular malformations can be clearly detected using these methods, particularly CT scans with contrast enhancement and color-Doppler ultrasonography[5]; MR imaging, with the multiplanarity and peculiar ability to visualise the main vessels, is particularly suitable for the evaluation of the abnormalities associated with polysplenic syndrome.

At present, the possibility of obtaining angiographic-type images through MRI and spiral CT angiography makes it possible, at times, to avoid conventional angiography, as, for instance, in the study of minor vascular malformations which may be present in polysplenic syndrome, such as a common coeliac-mesenteric vessel[3,5].

It is worth emphasising that often even a standard X-ray examination can provide numerous useful elements for defining the abnormalities found in the syndrome, such as the presence of a right gastric bubble, dextrocardia, absence of the inferior vena cava in the lateral view and the widening of the paravertebral pleural reflection in the right or left paratracheal area indicating a dilated azygous and hemiazygous system[1,11], or the absence of the minor fissure and hypoarterial position of both bronchi, indicating a left isomerism[1,3,5,11].

The role of ultrasonography in the prenatal diagnosis of this syndrome should also be stressed, through detection of the aorta and an enlarged azygous vein running together in a paraspinal retrocardiac location (double vessel sign) caused by hypertension of the inferior vena cava[12].

In conclusion, the polysplenic syndrome is a rare condition, occasionally found in asymptomatic adults[5]. Radiological detection could be mistaken for mediastinal or abdominal pathological masses, and only an accurate study performed with several different radiological methods can provide a firm diagnosis[5].

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