

A very unusual cause of severe right upper quadrant pain: splenic infarct in a patient with heterotaxy syndrome

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DESCRIPTION

A 31-year-old woman with a prior history of morbid obesity presented to a local facility with severe right upper quadrant pain for 1 day. The patient had work-up at the outside hospital that showed elevated liver enzymes. A right upper quadrant ultrasound was done; however, they were not able to locate her gallbladder. The patient was subsequently transferred to hospital. At presentation to our hospital she was complaining of severe 10/10 right upper quadrant pain. Her vital signs were stable, and an abdominal exam revealed significant right upper quadrant tenderness. The patient had repeat basic labs performed that showed elevated alanine transaminase (ALT) and aspartate transaminase (AST). The patient subsequently had a CT abdomen and pelvis that showed findings consistent with heterotaxy syndrome with polysplenia (figure 1). There were multiple small splenules in the right upper quadrant with an infarct in one of them that was likely the cause of the severe right upper quadrant pain (figure 2). Some other findings evident in our patient were a midline liver, presence of the stomach in the right upper quadrant. The intestines were malrotated with the caecum in the

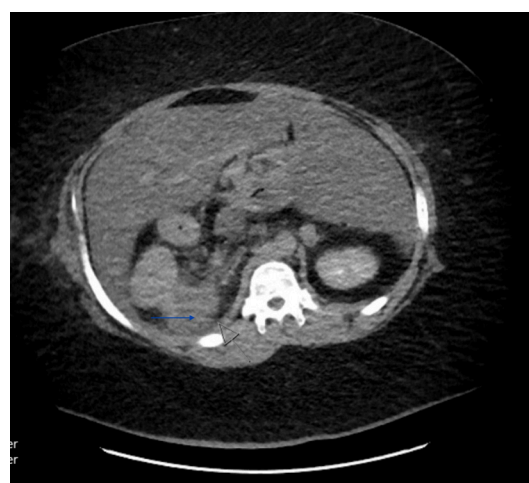


Figure 2 CT of the abdomen and pelvis showed an infarct in one of splenules that was likely the cause of the severe right upper quadrant pain (black arrow).

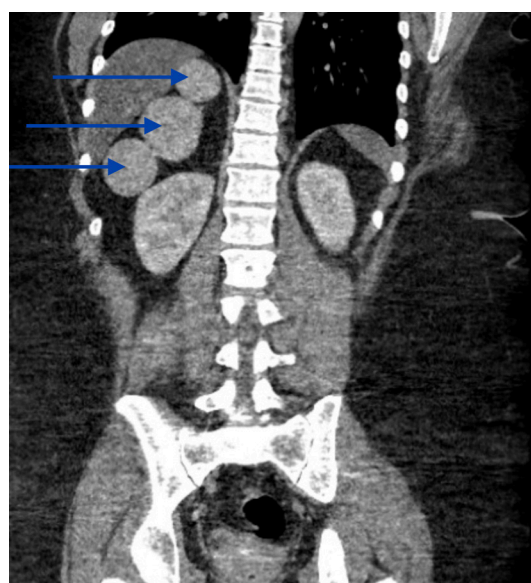


Figure 1 CT of the abdomen and pelvis was consistent with heterotaxy syndrome with polysplenia. There were multiple small splenules (blue arrows) in the right upper quadrant.

midline and terminal ileum in the right lower quadrant. The inferior vena cava was interrupted with azygos continuation. She had further work-up but no cause for the splenic infarct was found. She was managed conservatively. Her elevated liver function tests (LFTs) were found to be chronic and attributed to fatty liver disease.

Heterotaxy syndrome is a condition in which the different organs in the thoracoabdominal cavity can be abnormal or malpositioned. It is a broad term with many different variants. Our patient had visceral heterotaxy which can be classified into heterotaxy with asplenia or polysplenia, as was in our case. Polysplenia syndrome usually involves the presence of multiple small splenules without a parent spleen. Other associations include midline liver, intestinal malrotation, bilateral hyparterial bronchi, bilobed lungs, bilateral left atria, and azygos or hemiazygos continuation of the inferior vena cava. Polysplenia syndrome typically does not involve congenital cyanotic heart defects and thus patients with this syndrome have better outcomes. Asplenia syndrome is characterised by the absence of a spleen. It also involves a midline liver and intestinal malrotation but in contrast to the polysplenia syndrome these patients typically have bilateral right atria, trilobed lungs and eparterial bronchi. Asplenia syndrome typically involves severe cyanotic congenital heart defects.¹



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Images in...

The exact incidence of this syndrome and its variants is not known due to underdiagnosis. Patients are either diagnosed at birth especially if they have a variant with congenital heart defects, or later in life incidentally after they undergo imaging for different reasons. The underlying pathophysiology involves genetic mutations that have been identified in as many as 60 different genes.¹

Learning points

- ▶ Heterotaxy syndrome is a condition in which the different organs in the thoracoabdominal cavity may be abnormal or malpositioned.
- ▶ It can be classified based on malpositioning of abdominal or thoracic viscera. In the first case, it is divided into heterotaxy with polysplenia or asplenia. In the second case, it is divided into isomerism of the right atrial appendages or isomerism of the left atrial appendages.
- ▶ In case of heterotaxy with polysplenia, there may be multiple small splenules and any pathology can have a very atypical presentation.

Due to the wide heterogeneity in underlying organ dysfunction, the treatment varies according to the organs involved. Patients with cardiac involvement usually require surgery early in childhood. Patients with heterotaxy involving the abdominal viscera may also develop complications after birth like bowel obstruction. However, they are more likely to be asymptomatic and be diagnosed later in life.¹

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

REFERENCE

- 1 Kim S-J. Heterotaxy syndrome. *Korean Circ J* 2011;41:227–32.

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