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Acute appendicitis in a patient with heterotaxy syndrome

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ABSTRACT

Heterotaxy syndrome, also known as situs ambiguous, is a congenital condition that results in an abnormal distribution of visceral organs in the chest and abdomen. Heterotaxy syndrome has generally been categorized into two forms: a left-atrial isomerism form with a polysplenic association and a right-atrial isomerism form with an asplenic association. The polysplenic form has a high degree of co-occurrence (80%–90%) with fatal and non-fatal cardiac abnormalities. Of patients afflicted with the polysplenic form of heterotaxy syndrome, only 10% reach adulthood without complications. However, situs anomalies can cause a confusing clinical picture for the physician when the patient's pain does not correlate with the expected location due to organ dislocation. The differential diagnosis can prove to be unclear, incorrect, and unfitting when the physician does not know that certain organs are situated in abnormal locations. We present a case of a young male that was unaware of his heterotaxy syndrome. He presented with atypical signs of appendicitis secondary to an abnormal position of his appendix. Incidentally, his rare and fairly unheard of condition, heterotaxy syndrome, was only discovered because he was afflicted with a well-known and common condition, appendicitis.

1. Introduction

Situs solitus is defined as a normal arrangement of internal organs along a left-right axis. Although this arrangement is normal physiologically, organs are often asymmetric when compared to their "mirror image" on the other side of the axis [1]. This asymmetry is in stark contrast to what is seen in the embryonic period in which all vertebral embryos are bilaterally symmetric in the early stages of development [2]. The exact opposite of situs solitus is defined as sinus inversus, in which internal organs mirror the normal arrangement exactly, including a finding of dextrocardia. Patients with sinus inversus are usually clinically normal pointing towards the conclusion that as long as the internal organs turn in a specific pattern, the actual direction of the turn is irrelevant [3]. Heterotaxy syndrome (synonymous with situs ambiguous [4]) is then defined as not only an abnormal pattern of anatomical organization of internal organs, but as a disruption of the aforementioned specific turn pattern [4]. Heterotaxy can be further divided into two subsets; right (atrial) isomerism which is associated with asplenia and a higher mortality rate. While left (atrial) isomerism is associated with polysplenia and a significantly lower mortality rate [1,5,6]. Situs anomalies do not directly causes symptoms in patients, however the randomness of organ location can confuse clinicians when infections or inflammations occur. Depending upon the type of anomalies present on imaging, further work-up may be warranted. We present a case report of a young Caucasian male patient with the polysplenic form of heterotaxy syndrome, whom presented with an atypical pain location concerning for appendicitis.

2. Case report

A 17-year-old Caucasian male presented to the emergency department with a complaint of mid-hypogastric pain that began earlier in the day; the initial pain was described as dull and non-radiating. At the time of presentation, the patient endorsed his pain level at a 6 out of 10 on the visual analogue scale and was sharp in character. He endorsed nausea and vomiting, but denied hematemesis, melena, hematochezia, fever, or chills. The patient was a relatively healthy young male, with only concussions and bipolar disorder in his past medical history. He denied any current medications, but endorsed previous lithium use 2 years ago. Social history was negative for any substance, alcohol, tobacco use, and adoption. His family history was non-contributory to this case and no history of psychiatric conditions were noted on either side. A CBC analysis showed significant leukocytosis at 19.7 × 10^3/μL (reference range: 3.7–10.5 × 10^3/μL) with a left shift. The basic

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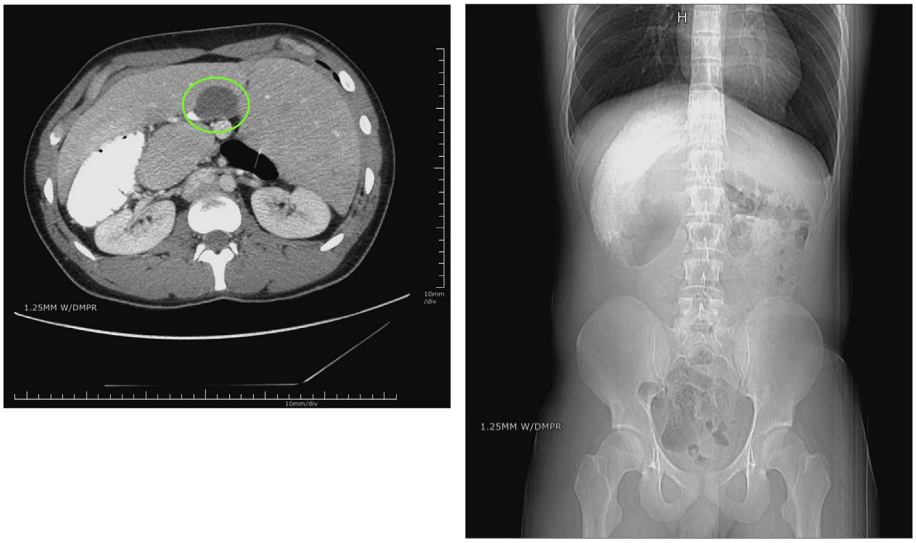

Fig. 1. 1.1. Abdominal CT, axial view, showing a midline liver and gallbladder (green circle). 1.2. Abdominal and pelvic x-ray showing the midline liver.

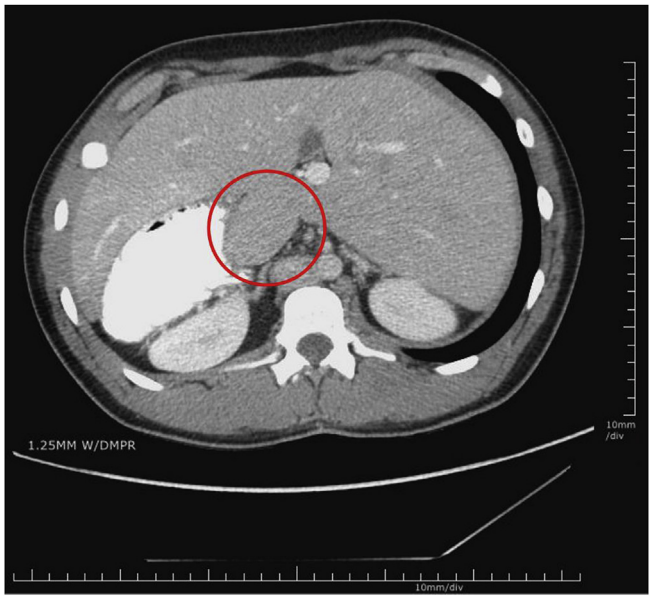

Fig. 2. Abdominal CT, axial view, showing polysplenia with a spleen encircled in red.

metabolic panel showed some slight electrolyte abnormalities, including hypokalemia at a value of 3.0 mmol/L (reference range: 3.3–5.1 mmol/L) and borderline hyponatremia at 132 mmol/L (reference range: 133–145 mmol/L). The patient was slightly hypocapnic at 19 mmol/L (reference range: 22–32 mmol/L). He had an elevation of his indirect bilirubin at 1 mg/dL causing an elevation in his total bilirubin at 1.2 mg/dL (reference range: 0.0–1.0 mg/dL); also of note, his direct bilirubin was within normal range at a value of 0.2 mg/dL (reference range: 0.0 0.3 mg/dL). Urine analysis showed an elevated specific gravity at 1.030 (reference range 1.010–1.025) and was positive for ketonuria at 80 mg/dl. A CT of the abdomen and pelvis with contrast was obtained and demonstrated a number of anomalies. In the lower thorax, continuation of the azygos vein into the inferior vena cava or absence of the hepatic segment of the inferior vena cava with azygos continuation was noted. The liver was found more toward midline (Fig. 1.1-1.2) and polysplenia was noted (Fig. 2).

The stomach was located on the right side (Fig. 3.1-3.3), with the cecum in the right pelvis.

The appendix was located in the mid-pelvis and was found to be diffusely enlarged at 12 mm in diameter; multiple appendicoliths were also seen on imaging, consistent with appendicitis (Fig. 4.1-4.2).

The patient was then made NPO (*nil per os*) pre-operatively and placed on subcutaneous heparin for deep vein thrombosis prophylaxis. Ertapenem was given in the emergency department for antibiotic coverage and a laparoscopic appendectomy for acute appendicitis was planned for later in the day. Intraoperatively, the liver appeared to be mostly on the left side of the abdomen, the bowel was noted to be in abnormal position, and multiple spleens were seen. With some difficulty, the appendix was identified on the cecum, retracted upward, and the mesoappendix was taken down with a harmonic until the base of the appendix was identified clearly. The appendix was removed and sent to pathology with a final diagnosis of acute appendicitis. The tubular appendix measured from 6.9 cm up to 1 cm in diameter. The wall of the appendix was diffusely thickened with diffuse edema and congestion with no evidence of perforation. Post-laparoscopic appendectomy, the patient was stable, voiding, and ambulating without difficulty.

3. Discussion

Heterotaxy arises from an abnormality in the left-right axis resulting in an anomalous arrangement of cardiac chambers, lungs, vessels, and abdominal organs. Sequela that are usually seen include loss (e.g. asplenia) of structures, gain of structures (e.g. polysplenia), failure to regress certain symmetric embryonic structures (e.g. persistent left superior vena cava), or lateralization (e.g. left atrial isomerism) [7]. Resembling how our patient presented, heterotaxy syndrome can occur secondary to gene mutations in an isolated individual or can occur as an inherited form. Regarding inherited forms of heterotaxy, multiple modes of inheritance have been described, from autosomal recessive to x-linked recessive to autosomal dominant with incomplete penetrance [7,8]. The cause of most cases of heterotaxy defects remains vastly unknown and are being identified as not having a monogenic etiology; however recent data shows possible maternal environmental influences in addition to genetic predisposition and chromosomal mutations [9,10]. The prevalence of heterotaxy, including both right and left atrial isomerism forms, has been roughly estimated at 0.99 per 10,000 births, citing a study conducted over 27 years and including 201,084 patients [9]. Another study estimated the prevalence of heterotaxy syndrome at 0.8 per 10,000 individuals, citing data collected over 11 years and including a patient population (n) of 4,664,529[10]. Heterotaxy syndrome