Journal of Medical Diagnostic Methods
ISSN: 2168-9784

Blommaert, et al., J Med Diagn Meth 2015, 4:1
DOI: 10.4172/2168-9784.1000166

Case Report Open Access

A Rare Case of Polysplenia Syndrome and Congenital Cloaca with Symptomatic Intestinal Non-rotation in an Adult Patient

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Received Date: Jul 01, 2014, **Accepted Date:** Jan 30, 2015, **Published Date:** Feb 3, 2015

Abstract

A 52-year-old women presented to our emergency department with nausea, vomiting and acute, progressive abdominal pain in the entire abdomen radiating to the back. Her history reported a congenital cloaca, for which she underwent reconstructive surgery twice during childhood. On physical examination normal bowel sounds were heard and no signs of peritoneal irritation were present. A contrast-enhanced computed tomography showed dilated small intestinal loops in the left upper quadrant and a bowel obstruction due to a non-rotation of the midgut. Furthermore, interruption of the vena cava inferior with azyguous continuation, polysplenia, short pancreas, bicornuate uterus and a small diaphragmatic hernia were seen. In this case report, these congenital anomalies are presented in the context of an unusual case of polysplenia syndrome.

Introduction

Situs ambiguous, or heterotaxy, is a rare congenital abnormal position of organs and vessels. This phenomenon is an intermediate form between situs solitus, which is the usual arrangement, and situs inversus, in which all organs and vessels are mirrored. Since there are many ways in which organs may be arranged, the clinical presentation of a heterotaxy syndrome can vary between patients. Heterotaxy can be further subdivided in an asplenia and polysplenia syndrome [1-3].

Polysplenia syndrome, a condition in which two or more spleens are present, is also called bilateral left-sidedness or left-isomerism. Left-sidedness results in bilobed lungs with hyparterial bronchi on each side, multiple spleens and a centrally located liver [4]. Autopsies of 146 polysplenia patients showed left-sidedness in only 55% of the patients [5]. Besides left-sidedness, abdominal heterotaxy was present in 57% of the polysplenia patients. In some polysplenia patients, the presence of multiple spleens is the only anomaly [5,6]. Examples of abdominal heterotaxy, besides polysplenia, are an interruption of the inferior vena cava (IVC) with azygous continuation, short pancreas, intestinal malrotation and a preduodenal portal vein [1]. Although the etiology of this syndrome is poorly understood, several genes associated with left-right embryological development. (e.g.CFC1, SHROOM3, GALNT11) have recently been identified [7-9]. We present an interesting case of an adult patient with polysplenia syndrome with multiple abdominal abnormalities, including a congenital cloaca. This is the first case report describing this unique combination of abnormalities in a patient with polysplenia syndrome.

Case Report

A 52-year-old woman from Turkish origin presented to our emergency department with acute, progressive abdominal pain since 12 hours, persistent nausea and vomiting. She suffered from progressive pain in the entire abdomen radiating to the back, with occasionally an increase in severity of complaints. Last defecation had taken place the previous day with unremarkable stools. Medical history reported reconstructive surgery for a congenital cloaca during childhood, angina pectoris (with percutaneous coronary intervention performed twice), diabetes mellitus type 2, hypothyroidism and an unexplained anemia.

On physical examination, a moderately ill patient was seen. Her vital signs showed no abnormalities, with a heart rate of 86 beats per minute, blood pressure of 116/53 mmHg, body temperature of 36.7°C and an oxygen saturation of 96% without additional oxygen supply. Inspection of the abdomen showed scars from earlier reconstructive surgical procedures by laparotomy, without any abdominal distention. Normal bowel sounds were heard and palpation was painful in all quadrants, without signs of peritoneal irritation. Laboratory serum analysis showed a white-blood cell count of 13,400 mm^3, with normal distribution. Values of hemoglobin, transaminases, creatinin and C-reactive protein were within the reference range. However, during the next two days, CRP rose to 264 mg/L.

Abdominal radiography showed some intraluminal fluid levels in the small bowel and constipation, in combination with mildly dilated proximal intestinal loops. No abnormalities were seen on chest X-ray. The diagnosis bowel obstruction (probably due to constipation or intra-abdominal adhesions) was made and the patient was admitted and treated conservatively with enemas and gastric suction. Since no clinical improvement was observed over the next day, a contrast-enhanced computed tomography (CECT) was performed, which showed dilated small intestinal loops due to a bowel obstruction in the left upper quadrant (Figure 1). The cecum and the entire large intestine were positioned on the left side of the abdomen, which suggests non-rotation of the bowel, a specific type of intestinal malrotation. Furthermore, interruption of the inferior vena cava with azyguous continuation, polysplenia, bicornuate uterus, right-sided stomach, small diaphragmatic hernia, short pancreas and a hydronephrosis of the right kidney were described.

The patient underwent an explorative laparotomy and peroperatively a bowel obstruction in the ileocecal angle in the left

upper quadrant of the abdomen was seen, possibly caused by adhesions or Ladd's bands. No gangrene was present. Ladd's procedure was performed; the obstruction was dissolved, Ladd's bands were cut and afterwards the appendix (located in the left upper quadrant) was removed. Postoperative recovery was complicated due to an aspiration pneumonia, which was treated with intravenous antibiotics.

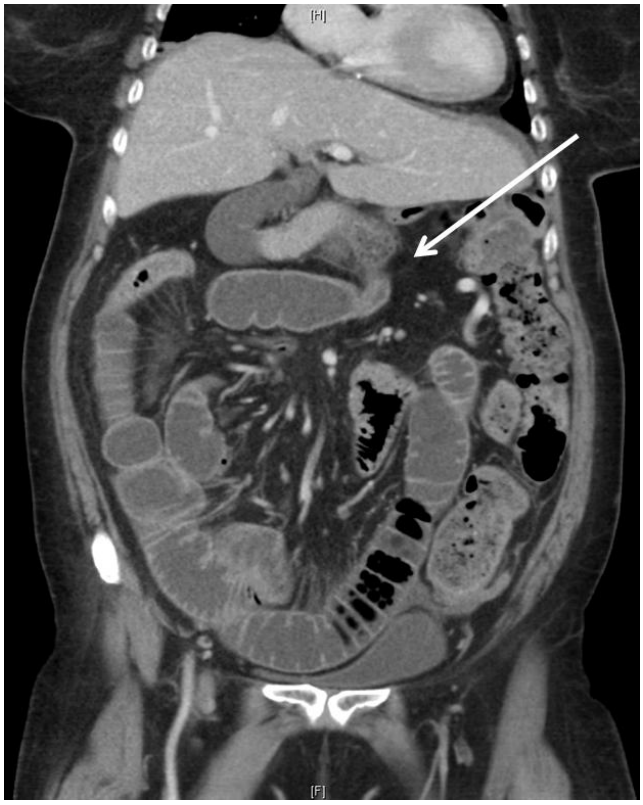

Figure 1: Coronal image of a CT showing the entire colon on the left side of the abdomen and dilated small intestinal loops due to an obstruction in the left upper quadrant (arrow). The obstruction is located where the terminal ileum transits into the cecum.

Discussion

Malrotation of the bowel is found in the majority of patients with heterotaxy syndrome [4,5-10]. It is caused by an abnormal embryological development of the midgut. Until the fifth week of pregnancy, the gastrointestinal tract is a straight tube of endoderm. During the fifth week (stage I), the midgut begins to rotate 90° counter-clockwise extra-embryologically. As a result, the small intestine is located on the right and the colorectum on the left. Starting in the tenth week (stage II), the midgut returns into the abdominal cavity and rotates another 180° counter-clockwise. At this point, the rotation is completed (270°) and the cecum is positioned in the right lower quadrant from where it descends into the right illiac fossa. At the same time the ascending and descending colon attach to the posterior abdominal wall (stage III) [11,12].

When the midgut fails to rotate normally, a malrotation develops. The non-rotation of the aforementioned patient occurred in stage II, since the midgut did not rotate the final 180° and, consequently, the entire colorectum remained on the left side of the abdomen. The pathological fibrous bands which attach the cecum to the posterior abdominal wall are called Ladd's bands and have the tendency to compress the duodenum. Furthermore, the cecum is very prone to volvulus, because of its relatively mobile position in the abdomen [11,12].

A symptomatic malrotation in adults is rare, since approximately 80% of patients presents within one month after birth, mostly with bilious vomiting. By the age of one year, approximately 90% of all patients has been diagnosed with a (symptomatic) malrotation and has already undergone surgery [11]. The remaining 10% of patients will develop symptoms in older age or will remain asymptomatic. It is reported that some asymptomatic patients suffer from irritable bowel syndrome [12]. When a patient develops symptoms of bowel obstruction, mostly caused by volvulus, Ladd's procedure should be performed. This procedure includes a counter-clockwise derotation of the volvulus, cutting Ladd's bands to mobilize the duodenum, widening of the small intestine's mesentery, and (if necessary) resection of non-vital gut. It also includes performing an appendectomy prophylactically to avoid misdiagnosing an acute appendicitis in the future [2,13].In asymptomatic patients (i.e. when malrotation is found accidentally), no consensus exists about performing a Ladd's procedure, since it is unclear whether the risks of this procedure outweigh the slight chance of a midgut volvulus in the older age [2].

Our patient showed multiple anomalies on CT, most of which are associated with polysplenia. The combination of IVC interruption with azyguous continuation, right-sided stomach and short pancreas has often been described in literature [1,5,6]. The presence of a bicornuated uterus and diaphragmatic hernia in patients with polysplenia syndrome is rare and only a few cases have been described [2,11-14]. However, the presence of a congenital cloaca in combination with an intestinal malrotation is extremely rare. In the literature, only one case has been reported, describing an infant with (among other anomalies) the combination of a congenital cloaca and intestinal malrotation. This infant died shortly after birth due to multiple other congenital defects (e.g. Potter sequence, VACTERL association, single umbilical artery, imperforate anus, micropenis with empty scrotum and cystic renal dysplasia). These other anomalies did not seem to be caused by the polysplenia syndrome [15].

Conclusion

Symptomatic malrotation in the context of a polysplenia syndrome is a rare anomaly, but is frequently diagnosed in childhood. Multiple other anomalies have been associated with the polysplenia syndrome, however, we present the first adult patient with polysplenia syndrome in combination with a congenital cloaca.

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