Portal vein thrombosis in a patient with hollow visceral myopathy

Introduction

Hollow visceral myopathy is a rare inherited disorder of smooth muscle that leads to impaired peristalsis and affects the function of the gastrointestinal tract. Patients with hollow visceral myopathy most commonly present with abdominal pain and vomiting, as a result of intestinal pseudo-obstruction. This article describes a woman with known hollow visceral myopathy who presented with these symptoms but in whom the cause was acute portal vein thrombosis. This has not previously been described in hollow visceral myopathy and raises the possibility that hollow visceral myopathy may predispose to venous thrombosis.

Discussion

Hollow visceral myopathy is a heterogeneous heritable disorder that can affect any part of the gastrointestinal tract. Both autosomal dominant and autosomal recessive variants have been described, but little is known about the underlying genetic defects. However, studies have identified an association with abnormal smooth muscle actin (Lehtonen et al 2012).

The proximal small bowel is most commonly affected, leading to chronic abnormal dilatation (Mitros et al, 1982), although any part of the gastrointestinal tract can be involved (Knafelz et al, 1996). The underlying pathological process is degeneration of smooth muscle cells within the muscularis propria, often with associated fibrosis (Smith et al, 1982; Sipponen et al, 2009). Some patients remain asymptomatic over long periods while others suffer repeated episodes of abdominal pain mimicking mechanical bowel obstruction.

The literature on hollow visceral myopathy is sparse but there is one other case recorded where a man with hollow visceral myopathy died from what was described as 'generalized thrombosis' (Mann et al, 1997). This earlier case, together with the unusual location of the thrombosis in the current patient, raises the possibility of a pro-thrombotic tendency during acute episodes of intestinal pseudo-obstruction in hollow visceral myopathy. An alternative explanation in the current case would be that volume depletion and localized compression of the portal vein led to thrombosis. Nevertheless, these two cases do suggest that prophylactic low molecular weight heparin is an essential facet of management when these patients present with acute gastrointestinal symptoms. BJHM

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Case Report

A 52-year-old woman presented with a 4-day history of diffuse non-colicky abdominal pain associated with vomiting. Preceding this she had 3 days of watery brown diarrhoea followed by constipation.

She had been diagnosed with hollow visceral myopathy as a child 40 years previously following investigation of persistent abdominal pain. Since then her symptoms had been limited to intermittent alternating diarrhoea and constipation often associated with abdominal pain. Her mother and two maternal cousins were also known to have hollow visceral myopathy; her mother had suffered only from abdominal pain and changing bowel habit but one cousin required surgical bowel resection and the other developed intestinal failure requiring long-term parenteral nutrition. Both had died in their forties but the causes of death were not known.

On admission, she was afebrile and looked well. Her abdomen was soft but diffusely tender, with normal bowel sounds. Cardiorespiratory examination was normal and urinalysis was negative. Her blood tests revealed a white blood cell count of 14 x 10^9/litre with a C-reactive protein level of 231 mg/dl. She had normal liver and renal function tests, clotting studies and amylase.

Ultrasonography of the abdomen suggested portal vein thrombosis. This was confirmed with a computed tomography scan which demonstrated extensive thrombosis of the portal venous system together with thrombosis of the superior mesenteric vein and some of its tributaries (*Figure 1*). There was also marked dilatation of the second and third parts of the duodenum, which had a maximum diameter of 13 cm. There was no radiological evidence of mechanical obstruction of the duodenum. Upper gastrointestinal endocscopy performed during the same admission showed only mild gastritis.

An intravenous heparin infusion was commenced and subsequently converted to low molecular weight heparin. Over the course of 7 days her symptoms improved and the C-reactive protein level steadily diminished. She was discharged on low molecular weight heparin which continues. For several weeks she had quite marked ascites but this gradually resolved and 6 months later she remained well with no gastrointestinal symptoms and normal inflammatory markers.

During the initial admission she had marked thrombocytosis (peaking at 937 x 10^9/litre). This was investigated with a JAK2 mutation test that proved positive. However, her platelet count normalized as inflammatory markers dropped and has remained normal over the subsequent 12 months. As a result, the JAK2 finding is thought to be incidental; it is known that the JAK2 mutation can be detected in healthy individuals (Sidon et al, 2006). The initial thrombocytosis was therefore considered to be reactive rather than a result of an underlying myeloproliferative disorder.

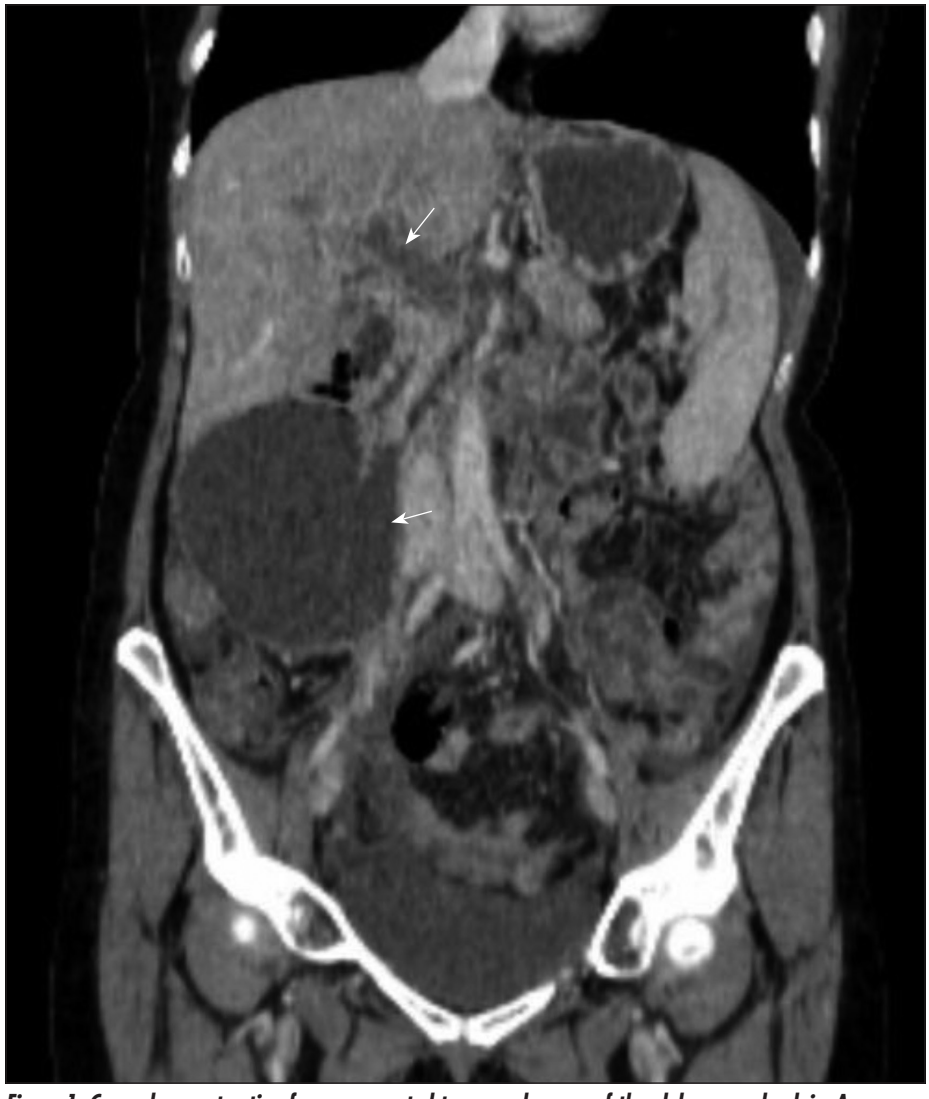

Figure 1. Coronal reconstruction from a computed tomography scan of the abdomen and pelvis. Arrows indicate the portal vein thrombosis and dilated duodenum.

LEARNING POINTS

- Where patients with hollow visceral myopathy present with abdominal pain, the likely cause is intestinal pseudo-obstruction.
- Abdominal imaging of patients with hollow visceral myopathy demonstrates marked dilatation of segments of the gastrointestinal tract, most commonly the proximal duodenum, without any evidence of mechanical obstruction. This may be a chronic finding.
- Portal vein thrombosis should be included in the differential diagnosis of patients presenting with acute abdominal pain. Ascites caused by portal vein occlusion may only appear later and resolve as collaterals form.
- Adequate thromboprophylaxis is important in patients with hollow visceral myopathy who present with acute symptoms as they may be at particular thrombotic risk.

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