**LETTER TO THE EDITOR**

**Intestinal volvulus and its increased incidence in patients with Cornelia de Lange syndrome**

**Dear Sir,**

I read with great interest the recent article by Papiamonis et al. One gastrointestinal complication that is seen with an increased frequency in patients with Cornelia de Lange syndrome and that is often under-reported is intestinal volvulus.

Volvulus in Cornelia de Lange syndrome is most common in the cecal portion of the intestine. Non-fixation of the caecum is the most common attributable cause of the volvulus. Concurrent non-fixation of the ascending colon is seen at the same time. López et al. have recently reported volvulus of the sigmoid colon too in patients with the Cornelia de Lange syndrome. Interestingly, patients with more severe mental retardation are more likely to develop a cecal volvulus. Overall, malrotation is seen in 2.3% of patients with the Cornelia de Lange syndrome.

Patients usually present with severe abdominal pain. Simultaneous bilious vomiting is usually present. Physical examination usually reveals abdominal distension with concurrent abdominal tenderness especially in the right lower quadrant. Rebound and guarding may develop rapidly. Volvulus may appear at any age. For instance, Husain et al. have reported cecal volvulus in patients as young as four years of age. Patients have the typical features of Cornelia de Lange syndrome such as mental retardation and marked failure of growth. Facial examination usually reveals a crescent shaped oral opening with low set ears and micro-brachycephaly. Other gastrointestinal anomalies such as colonic duplication and pyloric stenosis may be present at the same time.

X-ray examination usually reveals the "half coffee bean" sign. Marked colonic dilatation usually accompanies the above sign. Contrast enhanced studies typically reveal the "bird's beak" sign. This is secondary to obstruction of the twisted intestinal portion. Emergent laparoscopy or laparotomy is required. This involves volvulus release. Delayed surgical intervention may result in cecal necrosis as well as necrosis of the ascending colon and the ileum. Volvulus release is usually followed by fixation of the cecum to the right peritoneum. Rarely, a cecal volvulus may occur secondary to GI procedures.

For instance, Frischman et al. recently reported the case of a patient with the Cornelia de Lange syndrome who developed a volvulus following gastro-duodenoscopy. As is obvious from the above discussion, an increased incidence of volvulus is seen in patients with the Cornelia de Lange syndrome. This complication should be watched out for in patients with the Cornelia de Lange syndrome presenting with acute abdominal pain.

**Conflict of interest**

The author has no conflicts of interest to declare.

**References**


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29 October 2013