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Intestinal malrotation in an adult with Sakati-Nyhan-Tisdale syndrome: A case report

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Abstract
Intestinal malrotation results from failure of embryological counter clockwise rotation of the small bowel about the superior mesenteric artery, resulting in abnormal anatomic positioning and fixation of the bowel within the abdomen. Intestinal malrotation is rare (0.5-1% of total population) and is usually diagnosed within the first month of life with recurrent volvulus. The Ladd's procedure is the standard of care surgical intervention for intestinal malrotation to improve anatomic positioning of the bowel and reduce the risk of future volvulus. Sakati-Nyhan-Tisdale Syndrome refers to a distinct syndrome of malformations, including craniofacial defects, limb abnormalities, and congenital heart defects. There is no direct evidence to suggest a common association between congenital intestinal malrotation and Sakati-Nyhan-Tisdale syndrome.

We report the case of a 35-year-old female with a history of Sakati-Nyhan-Tisdale Syndrome and intestinal malrotation, presenting with recurrent postprandial epigastric and right upper quadrant pain. Imaging confirmed congenital malrotation without obstruction. She underwent a laparoscopic Ladd's procedure, which was converted to open laparotomy due to limited space and complex adhesions. Intra-operative findings confirmed malrotation and revealed midgut volvulus. The patient had an uneventful postoperative recovery and was discharged home after four days.

Congenital intestinal malrotation predominantly pediatric disease. In adults, its rarity and chronic, nonspecific gastrointestinal symptomology frequently leads to delayed diagnosis and increases risk for severe complications like volvulus if not identified. Its embryological origin involves abnormal rotation and fixation of the intestines during fetal development, and it is frequently associated with other congenital anomalies.

Increased clinical suspicion of congenital intestinal malrotation and early imaging are crucial for timely diagnosis and intervention in adults, preventing serious complications. Recognizing its possible association with rare syndromes may further enhance patient evaluation and care. Despite being extremely rare, future patients diagnosed with Sakati-Nyhan-Tisdale syndrome may benefit from additional workup of intestinal malrotation.

Keywords: Congenital intestinal malrotation, Sakati-Nyhan-Tisdale syndrome, Ladd's procedure, intestinal volvulus, surgical case report

Introduction

Intestinal malrotation is a rare congenital anomaly, affecting 0.5- 1% of the total population, resulting from failure of the small bowel to rotate 270° counterclockwise around the superior mesenteric artery during the first ten to twelve weeks of embryological development [1-2]. The resulting abnormal rotation and fixation of the bowel within the abdominal wall causes a narrow mesenteric base and Ladd's band (ceco-duodenal adhesion attached to the right lateral abdominal wall) to form. These patients are predisposed to obstruction and midgut volvulus, and most frequently present with severe episodic abdominal pain and bilious emesis within the first month of life in 50-80% of cases [3-4]. Failure to recognize the condition, resulting in delayed diagnosis, is largely attributed to non-specific symptoms, such as non-bilious vomiting, abdominal pain, or distension, which intermittently present throughout childhood and into adulthood [5].

The gold standard for diagnosis of malrotation is through an upper GI contrast study. Classic findings include a duodeno-jejunal flexure that does not pass to the left of midline, and a "corkscrew appearance" if a volvulus is present at the time of imaging [4]. If a malrotation is diagnosed, a Ladd's procedure is indicated, even if the patient is asymptomatic, to minimize the risk of volvulus or to accomplish de-rotation of present volvulus.

Sakati-Nyhan-Tisdale Syndrome, also known as Acrocephalopolysyndactyly Type III, is a rare (<1:1,000,000) acrocephalosyndactylia first described in 1971 that characterized by

abnormalities in the bones of the legs, congenital heart defects, craniofacial defects, and craniosynostosis [6]. Previous case reports of those born with the condition remark inguinal hernias as a common occurrence, however, no previous cases have reported intestinal malrotation as an associated abnormality [6].

This report describes a case of a 35-year-old female with Sakati-Nyhan-Tisdale Syndrome presenting with symptoms of chronic, intermittent bowel obstruction secondary to intestinal malrotation who was successfully treated with a laparoscopic converted to open Ladd's procedure.

Case Report

A 35-year-old female presented for a review of recurrent postprandial epigastric and right upper quadrant abdominal pain over 6 months. Her medical history includes Sakati-Nyhan-Tisdale Syndrome for which she has undergone numerous surgical operations to correct craniofacial defects and limb abnormalities, although she had no previous abdominal surgeries. Her abdominal pain was presumed to be related to biliary dyskinesia based on a prior hepatobiliary iminodiacetic acid (HIDA) scan. Given her unique medical history and presentation, there was increased suspicion of a possible bowel abnormality as the underlying cause of her pain. Clinical examination revealed mild tenderness to palpation of the epigastrium, and biochemical panels were unremarkable. Upper gastrointestinal (GI) series confirmed congenital intestinal malrotation without evidence of active obstruction (Fig 1).

After obtaining informed consent, the patient underwent an elective laparoscopic Ladd's procedure. Upon entering the abdomen, significant abnormal anatomy was observed, most closely resembling congenital intestinal malrotation. There was evidence of volvulus of the midgut, which likely caused a chronic partial obstruction, and there was evidence of severe venous congestion over much of the mesentery (Fig 2A). After releasing adhesions overlying the duodenum, running from the inferior edge of the liver and gallbladder to its attachments on the mesentery of the jejunum (Fig 2B), the underlying presence of severe adhesive disease between the duodenum, porta hepatis, gallbladder, and colon was revealed (Fig 2C). Volvulus of the small bowel was noted (Fig 2D). The entirety of the duodenum was released, allowing it to lie flat rather than its original corkscrew orientation, and was found to be entirely intraperitoneal. There were retroduodenal adhesions to the inferior vena cava, which was found to be significantly dilated. As an aside, the patient's ovaries were found to be streaking into the lower abdomen bilaterally which explains inability for previous ultrasounds to localize the ovaries.

The lysis of adhesions and freeing of the bowel was significantly impaired by both the extent of adhesions and the small stature of the patient. It was therefore decided to convert to open laparotomy to finish the case and ensure adequate detorsion of the small bowel. After opening the abdomen, the attachments from the right colon (on the left side of the abdomen) to the duodenum were lysed, followed by the retroduodenal attachments to the inferior vena cava. The small bowel was then run from the duodenum to the terminal ileum and was found to be volvulized in a clockwise orientation as expected. It was de-volvulized by rotating it counterclockwise, which slightly improved the previously noted venous congestion of the mesentery (Fig 3). The stomach and pancreas were visualized and appeared to be largely normal and in the correct anatomic position. The colon was then run from the terminal ileum to the rectum, and it was found to be largely free with minor left abdominal sidewall attachments which resembled normal anatomy, and did not require significant lysis of adhesions (Figure 3). An appendectomy was then performed. The bowel was then run in its entirety yet again from the stomach to the rectum, and all of the bowel was found to be free and without signs of volvulus.

All of the small bowel was placed on the patient's right side, and all of the colon was placed on the patient's left side to prevent recurrence of volvulus. Finally, the laparotomy incision was closed in layers, concluding the procedure. The patient's postoperative recovery was unremarkable, and they were discharged home 4 days postoperatively.

Figure format

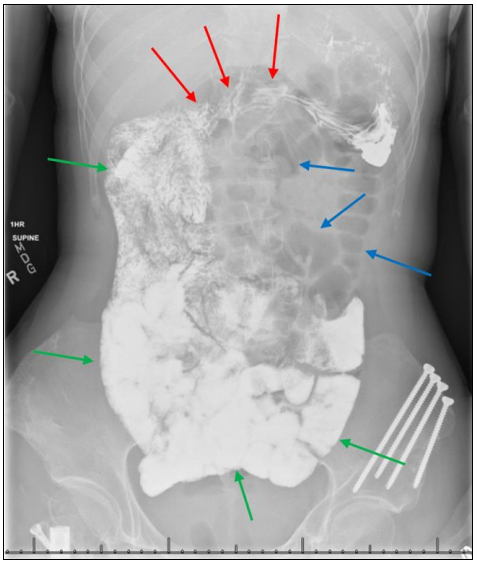

Fig 1: Radiograph from the patient's upper gastrointestinal contrast study showing a duodenum (red arrows) that remains confined to the right hemiabdomen and does not cross the midline as it transitions to the jejunum. Nearly all of the small bowel (green arrows) is situated within the right hemiabdomen, while the entire colon (blue arrow) is located within the left hemiabdomen.

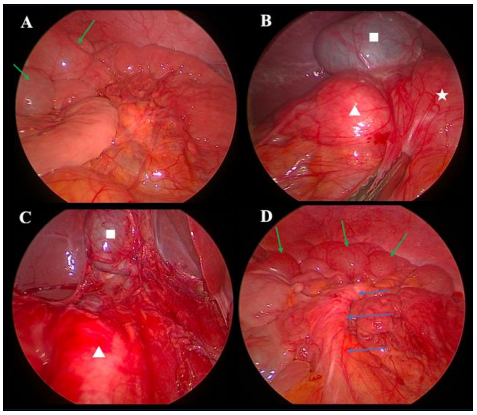

Fig 2: Laparoscopic images depicting (2A) venous congestion of the mesentery and dilated jejunal loops (green arrows), (2B) Band adhesions between the serosa of the gallbladder (■) and the mesentery overlying the duodenum (★) and colon (▲), (2C) exposure of the colon (▲) following lysis of band adhesions, and (2D) Volvulus of the small bowel resulting in twisting of the mesentery (blue arrows) and significant dilation of jejunal loops (green arrows)

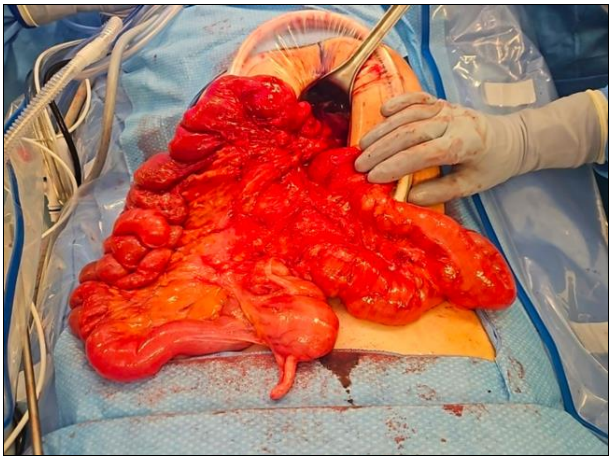

Fig 3: Intraoperative image showing the terminal ileum after detorsion, before appendectomy, with improvement of previously noted venous congestion. The cecum, ascending colon, and the proximal transverse colon are visible, mobilized, and appear free of adhesions.

Discussion

Congenital intestinal malrotation is most frequently diagnosed in infancy, with 90% of cases identified within the first year of life[7]. Rarely, malrotation presents in adulthood, accounting for 0.2-0.5% of all cases [8]. In adults, malrotation often manifests as chronic nonspecific gastrointestinal symptoms, such as abdominal pain, nausea/vomiting, constipation, bloating, or GERD [9-10], with some studies estimating that 32-80% of adults experience symptoms for years prior to diagnosis [9-11]. Since it is often considered a condition of infancy, delayed diagnosis in adults may be due to its rarity in this population, leading to misdiagnosis for other common adult conditions like peptic ulcer, irritable bowel syndrome, GERD, or psychiatric diseases. Adults typically experience vague, more chronic abdominal symptoms compared to the pediatric population, so diagnosis and intervention may be delayed until a severe complication occurs or unless it is found incidentally on imaging. In the pediatric population, 70-80% of infants are diagnosed after an emergent presentation related to volvulus or intestinal ischemia, whereas 27-33% of adults present with volvulus [12-14]. In adults with chronic, nonspecific gastrointestinal symptoms, clinicians should maintain a broad differential diagnosis that includes congenital intestinal malrotation to prevent delayed diagnosis, which can contribute to years of life-disrupting symptoms or the development of an acute abdomen necessitating emergency surgery or bowel resection.

Contaminant congenital anomalies are not uncommon in cases of intestinal malrotation, and in some instances, anomalies are related to the timing of embryological development of other organs occurring simultaneously with the 270° rotation of the intestines between the 5th-12th week of gestation. Some studies estimate that up to 60% of patients have co-occurring anomalies, including intestinal atresias, annular pancreas, congenital cardiovascular defect, anorectal malformation, Trisomy 21, Meckel diverticulum, and duodenal web [15-16].

Sakati – Nyhan - Tisdale Syndrome (Acrocephalopolysyndactyly Type III) is a disorder characterized by severe craniosynostosis, acrocephaly, limb abnormalities such as hypoplastic tibias, bowed femora, polydactyly, and syndactyly, as well as congenital heart disease. It exhibits autosomal dominant inheritance and features craniofacial anomalies, limb deformities, and developmental delays, with some patients also showing scalp atrophy and abnormal skin findings [6, 17]. There is no direct evidence associating Acrocephalopolysyndactyly Type III and intestinal malrotation. The only patient described with this condition in the literature was found to have an inguinal hernia and no other GI defects [6]. Notably, the most common of the Acrocephalopolysyndactyly disorders, Type II (Carpenter Syndrome), has been associated with ventral and umbilical hernia, and in one case, situs inversus of the stomach and liver and intestinal malrotation [18-19]. Despite its rarity, patients diagnosed with Sakati-Nyhan-Tisdale syndrome may benefit from additional imaging and diagnostic workup of intestinal malrotation to prevent long-term, undiagnosed symptoms and a potentially emergent abdominal presentation.

Conclusion

Congenital intestinal malrotation is most commonly diagnosed in infancy, but its presentation in adults is rare and often manifests as nonspecific GI symptoms, leading to frequent misdiagnosis or delayed diagnosis. Increased awareness among clinicians is essential to consider malrotation as a part of a differential diagnosis, especially in adult patients with chronic, unexplained abdominal complaints. Early imaging and prompt surgical intervention can prevent severe complications such as volvulus and bowel ischemia. Although rare, recognizing the potential coexistence with syndromes like Sakati-Nyhan-Tisdale can improve comprehensive patient assessment and management.

Conflict of Interest

Not available.

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Not available.

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