



## Rubinstein–Taybi Syndrome with Cecum Volvulum: Case Report

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### Abstract

Rubinstein-Taybi syndrome is considered a pathology of malformative origin secondary to a chromosomal microdeletion; presenting a prevalence of 1-9/100,000 live births with characteristic features such as microcephaly, distinctive facial features, short stature, intellectual disability, and conduct disorders. In this case report, the presence of a cecum volvulus with involvement of the ascending colon is presented in a patient with Rubinstein-Taybi syndrome, who at the time of admission and on a plain radiograph was identified with the typical kidney image, compatible with cecum volvulus, for which he underwent emergency surgical intervention. Subsequently, the patient was discharged within a month without complications.

### Introduction

Rubinstein-Taybi syndrome was first described in 1963 by Jack Rubinstein and Hooshang Taybi and corresponds to a microdeletion of chromosome 16p13.3 with an autosomal dominant inheritance pattern, characterized by congenital abnormalities such as microcephaly, dysmorphic facial features, short stature, intellectual disability, conduct disorders, genitourinary malformations and wide thumbs and first toes, which also gives its other name as shovel toe syndrome [1]. The diagnosis can be made clinically, finding the pathognomonic anomaly of the thumbs or through hybridization techniques.

An association between the syndrome and a probable increased incidence of intestinal malrotation was reported in patients with Rubinstein Taybi in 2015, an association between these two entities is suggested in the case series presented by Stevens, however further studies and larger populations are required to confirm this association [2]. In patients with malrotation, cases of volvulus and intestinal obstruction that require surgical management appear more frequently. The volvulus in patients with malrotations usually has atypical presentations, with the affected side being the right colon in most cases.

### Case Presentation

We present the case of a 26-year-old male patient with a confirmed diagnosis of Rubinstein-Taybi and a history of Hodgkin lymphoma, diaphragmatic repair and bladder plasty who went to the emergency department in February 2020 for presenting a 36-h history of anorexia and gastrobiliary vomits on numerous occasions. On abdominal examination, the patient was found with marked

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**Figure 1:** Anteroposterior simple radiography where classic kidney signs in left hypocondrium is appreciated; this is the dilated and volvulated cecum. It must not be confused with the neumoperitoneum of an intestinal perforation.



**Figure 2:** 2A Necrotic and ischemic right colon. 2B Necrotic fragment of ileon and right ascending colon. 2C Mesenteric anticlockwise rotation with necrotic changes in cecum (green arrow).

distension, absence of bowel sounds, cutaneous hyperalgesia and tympanic percussion with loss of liver dullness and pain on superficial and deep palpation, associated with involuntary muscular resistance. As part of the diagnostic approach, a plain abdominal radiograph was performed (Figure 1) where the characteristic sign of the kidney in the left hypochondrium corresponding to the cecum volvulus was found, which is why he was urgently operated with the following findings: Cecum volvulus that involved a large part of the ascending colon and approximately 10 cm from the terminal ileum (Figures 2A-2C). Right hemicolectomy was performed with ileo-transverse end-to-end anastomosis. Upon revision of the cavity, redundant sigmoid colon with a long mesentery and free fluid of inflammatory characteristics were found in approximately 300 cc. The patient after the surgical event showed progressive improvement, being discharged without complications 1 month after admission.

## Discussion

Volvulus can occur anywhere in the gastrointestinal tract, being the most common presentation the colon [3]. The most common presenting volvulus are sigmoid (43% to 71%) and cecum (10% to 52%) [4]. The cecum volvulus is when a rotation or torsion of the mobile cecum, ascending colon and terminal ileum occurs around its mesentery, being responsible for 1% to 3% of mechanical colonic origin obstructions [5], in close association with its embryological origin, the cecum is free due to a lack of fixation to retroperitoneal structures or a larger mesentery. This anatomical variant presents in up to 25% of adult population [6].

There are 3 types of cecum volvulus classified as type 1: The cecal volvulus generates a torsion on its own axis clockwise, remaining in the right hypochondrium, type 2: The cecum volvulus is generated by a terminal ileum torsion in the cecum resulting in a new localization of this structure (usually in the left hypochondrium) having a characteristic torsion of the mesentery (Figure 2C) that is generated in an anti-clockwise direction and type 3: Which is the "cecal bascule". In which the cecum folds over the ascending colon without generating torsion of the mesentery, found in 10% of the cecum volvulus [7,8].

Another history described in the genesis of the cecal volvulus in patients with malrotation is the history of abdominal surgery, since it is reported in some series that up to 53% of patients have this associated factor, related to the adhesions that form and generate stitches of fixation from which the hemicolon rotates [9].

The preoperative diagnosis of cecum volvulus based merely on clinical findings is very difficult to obtain due to its unspecific

presentation, most of the time patients with clinical symptoms of intestinal obstruction are admitted and it is not until abdominal radiographs are requested that the volvulus is identified. In some series it has been reported that up to 56% of cecal volvulus can be diagnosed with an abdominal radiograph [10]. The characteristic sign in "kidney form" described in the bibliography is located most of the time in the upper left quadrant, other imaging findings that we can find is a dilated cecum (>10 cm) with the presence of air-fluid levels, dilation of loops of small intestine located lateral to the cecum and absence of gas in the distal colon [6].

Computed tomography is currently the preferred imaging method for the diagnosis of cecal volvulus. Among the radiological signs that we can find, this is the swirl sign, which is defined as a turn of the cecum's around the mesenteric vessels [6,7]. The association of a distended ectopic cecum together with the swirl sign is already accepted to diagnose cecum volvulus, while thickening of the intestinal wall, intestinal pneumatosis, increased density of mesenteric fat and pneumoperitoneum tells us about a complicated disease [11]. Because cecum volvulus type 1 and 2 generate a rotation over their mesentery, this can induce vascular strangulation, making them more likely to complicate than the cecal bascule type [11].

The treatment for cecal volvulus will always be surgical, among the therapeutic options we have manual detorsion, cecopexy, cecostomy and right hemicolectomy either by laparoscopic or open approach. Manual detorsion is only recommended when no compromise of the colon is found; resection is mandatory when there are findings of perforation or gangrene. This is seen in about 50% of patients with cecum volvulus. Cecostomy has been associated with low recurrence rates, but high morbidity and mortality rates compared to cecopexy, in several articles it is mentioned that the use of cecostomy should only be performed in hemodynamically unstable or high-risk patients, this due to the fact that cecostomy is associated with necrosis of the cecum and fistula formation. A primary anastomosis or ileostomy with a mucous fistula after hemicolectomy is recommended depending on the patient's condition, surgical time, and bowel condition for a correct anastomosis, this being the most widely accepted treatment for cecum volvulus [6,12].

## Conclusion

Rubinstein-Taybi syndrome is a low prevalence chromosomal disease with anomalies and complications widely described, not being volvulation or gastrointestinal disorders one of the main ones. All patients with an intellectual disability, who present an objective physical examination with data on peritoneal irritation, with

subjective questioning, should be evaluated with an imaging study to rule out any complication, even when these complications are not described in the literature as common in this specific pathology. The presence of intestinal obstruction of mechanical origin secondary to a cecum volvulus is a very rare etiology, but it requires timely diagnosis and treatment because the associated complications present a high morbidity and mortality.

## References

1. Rana V, Srivastava N, Kaushik N, Mohan M, Chhabra R. From diagnosis to management: Rubinstein-Taybi syndrome. *IJSS Case Rep Rev.* 2017;3(11):4-7.
2. Stevens CA. Intestinal malrotation in Rubinstein-Taybi syndrome. *Am J Med Gen.* 2005;167(10):2399-401.
3. Peterson CM, Anderson JS, Hara AK, Jeffery WC, Menias CO. Volvulus of the gastrointestinal tract: Appearances at multimodality imaging. *Radiographics.* 2009;29:1281.
4. Ballantyne GH, Brandner MD, Beart RW Jr, Ilstrup DM. Volvulus of the colon. Incidence and mortality. *Ann Surg.* 1985;202(1):83.
5. Lee SY, Bhaduri M. Cecal volvulus. *CMAJ.* 2013;185:684.
6. Hasbahceci M, Basak F, Alimoglu O. Cecal Volvulus. *Indian J Surg.* 2012;74(6):476-9.
7. Madiba TE, Thomson SR. The management of cecal volvulus. *Dis Colon Rectum.* 2002;45:264-7.
8. Delabrousse E, Sarliève P, Saille N, Aubry S, Kastler BA. Cecal volvulus: CT findings and correlation with pathophysiology. *Emerg Radiol.* 2007;14:411.
9. Consorti ET, Liu TH. Diagnosis and treatment of caecal volvulus. *Postgrad Med J.* 2005;81:772-6.
10. Ballantyne GH, Brandner MD, Beart RW Jr, Ilstrup DM. Volvulus of the colon. Incidence and mortality. *Ann Surg.* 1985;202(1):83-92.
11. Delabrousse E, Sarliève P, Saille N, Aubry S, Kastler BA. Cecal volvulus: CT findings and correlation with pathophysiology. *Emerg Radiol.* 2007;14:411-5.
12. Rabinovici R, Simansky DA, Kaplan O, Mavor E, Manny J. Cecal volvulus. *Dis Colon Rectum.* 1990;33:765-9.