

CASE REPORT

Rapunzel Syndrome in a Patient with Autism

Otizimli Bir Hastada Rapunzel Sendromu

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ABSTRACT

Trichobezoar should be considered in the differential diagnosis in pediatric patients who present with complaints of abdominal pain, feeding problems, and vomiting as well as psychiatric problems. Rapunzel syndrome is a rare form of trichobezoar that can extend from the stomach to the small intestine and even to the colon. In this case report, a 9-year-old girl with autism accompanied by trichotillomania and trichophagia and diagnosed with Rapunzel syndrome is presented.

Keywords: Autism; Rapunzel syndrome; Trichobezoar

Öz

Trichobezoar should be considered in the differential diagnosis in pediatric patients who present with complaints of abdominal pain, feeding problems, and vomiting as well as psychiatric problems. Rapunzel syndrome is a rare form of trichobezoar that can extend from the stomach to the small intestine and even to the colon. In this case report, a 9-year-old girl with autism accompanied by trichotillomania and trichophagia and diagnosed with Rapunzel syndrome is presented.

Anahtar Kelimeler: Otizm, Rapunzel sendromu, Trikobezoar

Introduction

Rapunzel syndrome (RS) was first described by Vaughan in 1968 (1). The syndrome is named after a fairy-tale written in German by the Grimm Brothers in 1812, and the character named Princess Rapunzel had the young prince rescue her from the prison by hanging down her long golden hair from the prison tower (2).

Trichobezoars are hairballs formed as a result of chewing and swallowing hairs or other indigestible foreign materials. They are usually localized in the prepyloric region (3). A history of trichotillomania and trichophagia are common features of all such cases. Rapunzel syndrome is a rare form of trichobezoar that can extend from the stomach to the small intestine and sometimes to the colon. We aimed to present our autistic patient diagnosed with Rapunzel syndrome, since we haven't seen any case of Rapunzel syndrome in the literature associated with autism spectrum disorder (ASD).

Case

A 9-year-old, first diagnosed with ASD when he was 3 years old. Her family had earlier noticed that she was not able to speak spontaneously and seemed to avoid eye contact a lot. After her diagnosis, she was directed to special education in a center for children with ASD, but the family did not want her to receive education and did not take to the child psychiatry checkups.

Due to complaints of feeding problems, weight loss, weakness and nausea the child was admitted to the pediatric emergency. Informed consent was received from the family. Her history revealed early satiety, postprandial fullness, loss of appetite, halitosis and a habit of pulling, plucking and eating her own hair for 3 months. In her anthropometric assessment, body weight and height were 31 kg (51 p) and 132 cm (30 p), respectively. Her physical examination was normal except that a firm and non-tender mass extending from the left hypochondrium to the epigastrium was detected on abdominal palpation.

In the laboratory examinations, complete blood count, erythrocyte sedimentation rate and blood biochemistry (levels of urea, creatinine, serum electrolytes and liver enzymes) were within normal limits. Direct abdominal X-ray examination displayed enlarged small intestine loops and a sizeable soft tissue density in the stomach, with varied air-fluid levels and diffuse radiopaque foci, mostly millimetric and two in 2 cm size, suggesting beads, nails or pins (Fig. 1). Abdominal ultrasonography showed that there was some free fluid in the pelvis between the folds of the intestines.

Abdominal contrast-enhanced computed tomography (CT) showed the presence of bezoars containing metallic foreign bodies that completely filled the stomach and the third part (pars horizontalis) of duodenum and

extended to the small intestine and colon (Fig. 2).

After the surgical removal of the 72 cm bezoar extending from the stomach to the upper part of the jejunum, it was a trichobezoar and was diagnosed with Rapunzel syndrome (Fig. 3) child psychiatry outpatient check-up was recommended after discharge.



Fig. 1. Diffuse and numerous radiopaque foci in the entire abdomen with multiple air-fluid levels.

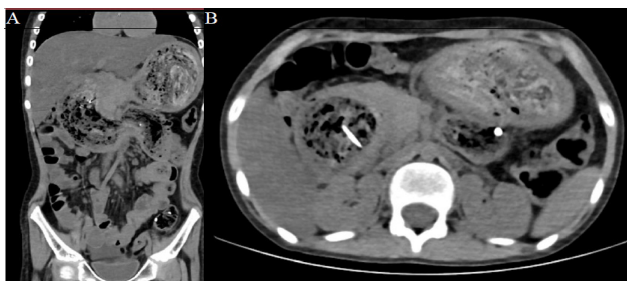


Fig. 2. (a) Coronal, (b) axial; CT scan image showing a non-homogenous non-enhancing mass with a mottled appearance within the lumen of the stomach first and second part of duodenum



Fig. 3. Trichobezoar (72 cm) containing numerous foreign bodies (beads, nails, pins and plastic parts)

Discussion

Gastric bezoars are mostly found in patients undergoing upper gastrointestinal endoscopy or imaging, because bezoars cause nonspecific symptoms in the stomach. Trichophagia constitutes only 1% of the patient group(4, 5). Phytobezoar, gastrointestinal stromal tumor, choledocholithiasis, and gastric cancer should also be considered in the differential diagnosis of trichobezoars (Table I)(4). Gastric atony may develop in patients with trichotillomania and trichophagia by large-volume trichobezoars, formed by the hair mass collected in the gastric folds which denatured by gastric acid and oxidized and then combined with food residues(3, 5).

In Rapunzel syndrome, trichobezoar is present with a tail-like extension, stretching from the pylorus to the duodenum, jejunum, ileum, or even the colon (3). A history involving hair biting is necessary for the diagnosis of Rapunzel syndrome. It mostly occurs under the age of 20 years with psychiatric disorders or autism, usually as a result of childhood neglect, abuse, or mental retardation (6, 7). Trichobezoar has been reported in different ways in patients with autism in the literature, but this case is valuable since the advanced form of trichobezoar, called Rapunzel syndrome, has never been reported.

Table 1.: Clinical and radiological differential diagnoses(4)

Differential diagnosis	Clinical Manifestations	Radiological findings
Phytobezoar	Loss of appetite, nausea, vomiting	Intraluminal mass containing air bubbles and obstruction appearance on CT examination
Gastrointestinal stromal tumor	Abdominal pain, fatigue, dyspepsia, nausea, loss of appetite, weight loss, fever and obstruction in the gastrointestinal tract	CT-confirmed viscous exophytic mass with ulceration and necrosis
Cholelithiasis	Biliary colic, jaundice, fever, nausea, vomiting, loss of appetite, pain extending to the back (pancreatitis), positive Murphy sign	USG and MRCP findings showing opacity and enlarged bile ducts
Gastric cancer	Pain, nausea, vomiting and early satiety due to obstruction	Ulcerated polypoid mass, focal thickening of the wall and mucosal irregularity on CT imaging
Trichobezoar	Nausea, pain, vomiting with or without perforation complications, pancreatitis, cholangitis	Intraluminal mottled mesh-like appearance in the epigastrium on CT imaging (can be longer in Rapunzel syndrome)

Patients with Rapunzel syndrome may be diagnosed late, as they do not show clinical symptoms until they reach a significant size, mostly due to insidious onset with atypical symptoms(5). In our case with autism too, the trichobezoar mass extending to the jejunum displayed late symptoms. Patients generally present with symptoms like abdominal pain (37%), nausea and vomiting (33.3%), abdominal mass, diarrhea or constipation, weight loss (7.4%), anorexia, early satiety, postprandial fullness, anemia and jaundice. Complications include gastrointestinal obstruction (25.9%), peritonitis and perforation (18.3%), gastrointestinal bleeding (10%), intussusception (7.4%), pancreaticobiliary obstruction, pancreatitis and malabsorption complications such as protein-losing enteropathy and iron deficiency, and megaloblastic anemia(3). Eating problems, weight loss, weakness and nausea were foremost symptoms in our case. The definitive diagnosis method is upper gastrointestinal system endoscopy, although valuable findings can be achieved for diagnosis by standing direct abdominal radiography, abdominal USG, upper gastrointestinal passage radiography and CT examinations(8). An appearance compatible with foreign bodies (bead, nail, pin, plastic parts) was found in direct abdominal radiography and CT examinations of our case, although she had no history of foreign body swallowing other than trichotillomania and trichophagia.

Ultrasonography may show an increased echogenic

band caused by hair, air, and food mixed together within the trichobezoar. USG did not provide sufficient diagnostic information in our case, although the rate of detecting trichobezoars by USG was reported as around 88%. CT shows trapped air bubbles or heterogeneous masses of homogeneous mottled appearance in the stomach or intestine region(9). The appearance of dense bezoar containing metallic foreign bodies filling the entire stomach and extending to the jejunum brought us to the diagnosis in our case. Upper gastrointestinal endoscopy is considered as the gold standard for the diagnosis of trichobezoars, but it is controversial to perform it in patients with Rapunzel syndrome, in the presence of a mass filling the stomach entirely(9). The diagnosis was concluded preoperatively, based on the patient's history, clinical examination, and radiological imaging studies.

Treatment depends on the size and location of the trichobezoars. Small trichobezoars can be removed endoscopically (10). However, Gorter et al. showed that endoscopic removal of trichobezoars was successful in only about 5% of patients (11). Laparotomy became the preferred treatment option for trichobezoars in Rapunzel syndrome, by virtue of the largest review of trichobezoar therapy, where 100 out of 108 patients (92.5%) were treated by laparotomy (4, 12). The laparoscopic approach should be considered only for eligible patients (with small to medium-sized bezoars) due to the increased operative time, the increased risk of intraabdominal spillage of trichobezoar content, and the need for longer incisions. Therefore, large (> 20 cm) symptomatic trichobezoars should be surgically removed by laparotomy, especially accompanied by Rapunzel syndrome. Enzymatic dissolution (with papain syrup, pancreatic lipase, and cellulose), Nd-YAG laser, and extracorporeal shockwave fragmentation are new approaches that need to be evaluated(13). The fact that the trichobezoar to be removed was 72 cm long in our case supports that the preferred treatment method should be laparotomy.

Although relapses of Rapunzel syndrome are rare, there is no doubt that prevention of relapses is important. For that purpose, long-term follow-up with upper gastrointestinal endoscopy or abdominal USG, psychotherapy, and psychiatric follow-up with cognitive behavioral therapy is essential following surgical removal of trichobezoars(3). Appropriate psychiatric counseling and follow-up is of great significance to achieve the main goal, which is to prevent relapses.

Conclusion

The key to achieve an accurate diagnosis is a history of psychiatric illness, a comprehensive clinical examination, and confirmation of the diagnosis by examination. Although Rapunzel syndrome is a rare disease, it should be kept in mind for differential diagnosis, especially in autistic children with a history of trichophagia or trichotillomania. Laparotomy is recognized as the most appropriate treatment option. Long-term follow-up and psychiatric control are

recommended to prevent relapses.

References

1. Vaughan Jr E, Sawyers J, Scott Jr H. The Rapunzel syndrome. An unusual complication of intestinal bezoar. *Surgery*. 1968;63(2):339-43.
2. Grimm B. Rapunzel. Translated by Godwin-Jones R. Richmond. Virginia Commonwealth University: Department of Foreign Languages. 1994:1994-9.
3. Nettikadan A, Ravi M, Shivaprasad M. Recurrent Rapunzel syndrome—A rare tale of a hairy tail. *International journal of surgery case reports*. 2018;45:83-6.
4. Vellaisamy R, Iyer S, Chandramohan SM, Harikrishnan S. Rapunzel syndrome with cholangitis and pancreatitis—A rare case report. *Open Medicine*. 2020;15(1):1137-42.
5. Gastric bezoars. Available at: <https://www.uptodate.com/contents/gastric-bezoars>. Accessed February 19.
6. Wijetilleke A, Sakran M, Kamat-Nerikar R. Vomiting in a girl with autism. *Clinical pediatrics*. 2009;48(2):224-7.
7. Grant JE, Odlaug BL. Clinical characteristics of trichotillomania with trichophagia. *Comprehensive psychiatry*. 2008;49(6):579-84.
8. Lalith S, Gopalakrishnan KL, Ilangovan G, Jayajothi A. Rapunzel syndrome. *Journal of clinical and diagnostic research: JCDR*. 2017;11(9):TD01.
9. Sidhu B, Singh G, Khanna S. Trichobezoar. *Journal of the Indian Medical Association*. 1993;91(4):100-1.
10. Chun SW, Lee K, Park YJ, Shin HD. Rapunzel Syndrome Removed with Enteroscopy in a Child. *The Korean Journal of Gastroenterology*. 2019;74(1):42-5.
11. Gorter R, Kneepkens C, Mattens E, Aronson D, Heij H. Management of trichobezoar: case report and literature review. *Pediatric surgery international*. 2010;26(5):457-63.
12. Maini A, John J. Trichobezoar requiring surgical intervention. *Journal of the American Academy of PAs*. 2018;31(9):32-4.
13. Nirasawa Y, Mori T, Ito Y, Tanaka H, Seki N, Atomi Y. Laparoscopic removal of a large gastric trichobezoar. *Journal of pediatric surgery*. 1998;33(4):663-5.