

Rapunzel Syndrome that Mimic Celiac Disease in a Three Year Old Girl

Virtut Velmishi^{1*}, Ermira Dervishi¹, Saimir Heta², Spiro Sila², Hysen Heta² and Paskal Cullufi¹

¹Service of pediatric gastroenterology, University Hospital Center “Mother Teresa” Tirana-Albania

²Service of pediatric surgery, University Hospital Center “Mother Teresa” Tirana-Albania

Abstract

Background: Rapunzel syndrome is an extremely rare intestinal condition in humans resulting from eating hair (trichophagia). Rapunzel syndrome presents a rare type of trichobezoar with an extension of the hair into small bowel

Case presentation: We present a three year old girl with Rapunzel syndrome, where the trichobezoar was not suspected at all. The history of disease was similar with celiac disease even though serology was negative.

Conclusion: In children with a long history of gastrointestinal symptoms, upper digestive endoscopy is a cornerstone of Rapunzel syndrome as a diagnostic tool (in our case for another objective). Open surgery remains the mainstay of large trichobezoar removal

Keywords: Bezoar; Trichotilomania; Trichotilofagia; Trichobezoar; Rapunzel Syndrome

Case Presentation

A three year old girl is presented in our clinic with anorexia, vomiting, irritability, abdominal pain and constipation. She is the first child of an Albanian couple. The pregnancy and delivery were uneventful. She is vaccinated according to Albanian schedule. She has no history of previous diseases or psychiatric problems. According to her parents she has lost 3 kg in the last 2 months presenting symptoms as above mentioned. Initially she was treated in a local hospital but without any improvement (Figure 1).

On physical examination this girl presents signs of malnutrition. Her weight was 11 kg (-3DS) and her height was 92 cm (-2DS). Cardiac sounds were normal. Auscultation of lungs was uneventful. Abdomen was distended but without hepatosplenomegaly. Her behavior was normal for age and neurological evaluation was normal. The laboratory analyses shows: WBC=12 x 10³/mm³; RBC=5.08 x 10⁶/mm³; HGB=7.7g/dl; PLT=358; x 10³/mm³. Urinalysis and biochemical findings were normal. IgA + IgG antitransglutaminase was normal. Abdominal ultrasonography and X-ray does not reveal anything

Corresponding Author: Virtut Velmishi, Service of pediatric gastroenterology, University Hospital Center “Mother Teresa” Tirana-Albania, E-mail: tutimodh@yahoo.com



Consulting the surgeons we decided to perform a barium enema to exclude Hirschprung disease. We had some diagnostic hypothesis such as: Coeliac disease, food allergy, Hirschprung disease, metabolic disease etc. The most possible diagnosis for us was Celiac disease considering weight loss, abdominal pain and chronic constipation even though serological tests were normal. We decided to perform an upper digestive endoscopy for intestinal biopsy. During the examination, surprisingly we have noticed a large mass of hair in the stomach of girl. We tried to remove this trichobezoar but our efforts were not successful. We passed the endoscope into pylorus and we noticed that a cluster of hair was extended in duodenum. Knowing the difficulty of removing endoscopically this trichobezoar we decided to perform gastrotomy. The surgeons removed this large trichobezoar which has a tail as it is shown in the picture. The diagnosis was clear now: Rapunzel syndrome. We have retaken the history from the parents for strange behavior of their daughter in the past (thricotilomania). They confirmed that two or three months ago her nanny has seen this girl eating her hair several times. Despite this, the parents denied again to have seen their daughter to do this action in their presence, underestimating the observation of her nanny. However this girl was referred to our psychiatrist for further evaluation (Figure 2).

Discussion

Rapunzel Syndrome is a rare form of trichobezoar. It is named after a tale written in 1812 by the Brothers Grimm about a young maiden, Rapunzel, with long hair who lowered her hair to the ground from a castle, which was a prison tower to permit her young prince to climb up to her window and rescue her[1]. This syndrome was originally described by Vaughan et al. in 1968 [2]. The commonly accepted definition is that of a gastric trichobezoar with a tail extending to the jejunum, ileum or the ileocecal junction

Figure 1 Images of 3 year old girl which show signs of malnutrition

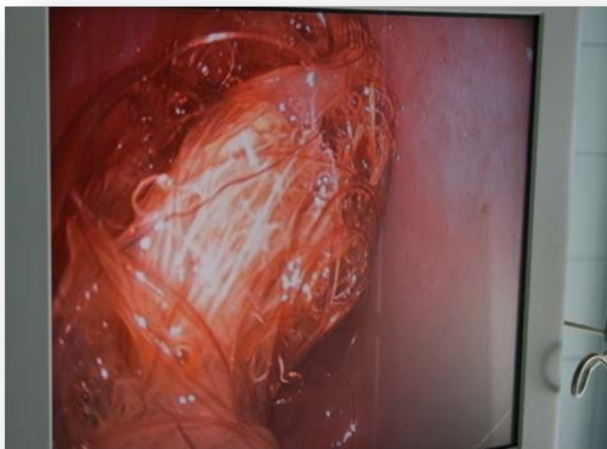


Figure 2: Endoscopic view of trichobezoar

The common presentation of trichobezoar is in young females usually with an underlying psychiatric disorder. In our case the presentation is in a very young age with hair extending down to the small bowel, causing symptoms, which could mimic gastrointestinal disease as enteropathy from gluten.

In the literature review performed in 2007 are identified 27 cases of Rapunzel syndrome described between 1968 and 2006 [3]. A few other reports are published in the medical literature after this date [4-6].

Conuguntla and Joshi reported in 2009 the youngest documented case of Rapunzel syndrome in the United States: a 5 year old girl with mental retardation with abdominal pain, vomiting, and a nontender abdominal mass [7].

The complications of Rapunzel syndrome range from attacks of incomplete pyloric obstruction to complete obstruction of the bowel to perforation to peritonitis and mortality [5-7]. Patient with Trichotillomania (a psychological condition that involves strong urges to pull hair), around 30% will engage in trichophagia, and of these, only 1% will go on to eat their hair to the extent requiring surgical removal [8]. Duncan et al recommended bezoar extraction by multiple enterotomies in cases of Rapunzel syndrome [9].

There has been few cases of recurrence following successful surgery [10]. Esophagogastroduodenoscopy is a method of choice for diagnosing trichobezoars. The typical colour of trichobezoar at endoscopy is black. It allows the clinician to distinguish between phytobezoars and trichobezoars. This is very important because treatment depends on the nature of a bezoar [11]. In the early stages endoscopic removal is not without risk of bowel perforation and should be reserved for small Trichobezoars only [12] (Figures 3).

Laparotomy is a therapy method of choice, with respect to its success, possibility of careful examination of whole gastrointestinal tract (stomach and intestines), as well as low level of complications [13]. Other methods including the use of laser ignited mini-explosive technique were used successfully [14]. Laparoscopy has been also used in recent years.

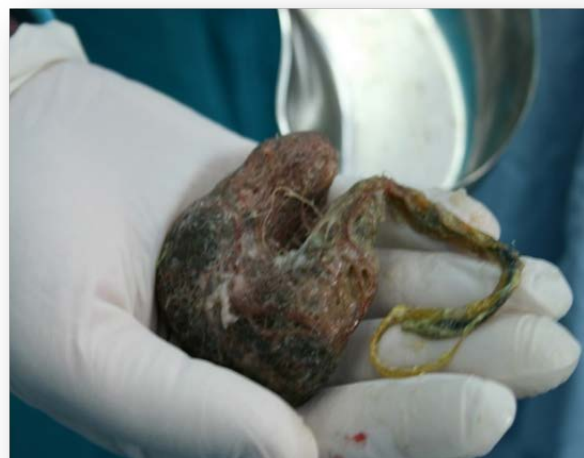


Figure 3: Images during surgery and after removal of trichobezoar.



Conclusion

In children with a long history of gastrointestinal symptoms, upper digestive endoscopy is a cornerstone of Rapunzel syndrome as a diagnostic tool (in our case for another objective). It is important to consider such diagnosis in face of suggestive symptoms, even signs of tricotillomania are not present. Open surgery still remains the main stay of large Trichobezoar removal especially if it has an extension into the bowel, which might be missed with other methods of treatment.

References

1. [Phillips MR, Zaheer S, Drugas GT \(1998\) Gastric trichobezoar: case report and literature review. Mayo Clin Proc 73: 653-656.](#)
2. [Vaughan ED Jr, Sawyers JL, Scott HW Jr \(1968\) The Rapunzel syndrome. An unusual complication of intestinal bezoar. Surgery 63: 339-343.](#)
3. [Gupta V, Naik S, Chaudhary AK, Jain P, Sharma AK \(2007\) Rapunzel Syndrome Reviewed and Redefined. Dig Surg 24: 157-161.](#)

4. [Mohite PN, Gohil AB, Wala HB, Vaza MA \(2008\) Rapunzel Syndrome Complicated with Gastric Perforation Diagnosed on Operation Table. J Gastrointest Surg.](#)
5. [Pérez E, Sántana JR, García G, Mesa J, Hernández JR, et al. \(2005\) Gastric perforation due to trichobezoar in an adult \(Rapunzel Syndrome\). Cir Esp 78: 268-270.](#)
6. [Ventura DE, Herbella FA, Schettini ST, Delmonte C \(2005\) Rapunzel syndrome with a fatal outcome in a neglected child. J Pediatr Surg 40: 1665-1667.](#)
7. [Conugunta V, Joshi DD \(2009\) Rapunzel syndrome:a comprehensive review of an unusual case of thrichobezoar. Clin Med Res 7: 99-102](#)
8. [Frey AS, McKee M, King RA, Martin A \(2005\) Hair Apparent: Rapunzel Syndrome. Am J Psychiatry 162: 242-248.](#)
9. [Duncan ND, Aitiken R, Venugopal S, West W, Carpenter R \(1994\) The Rapunzel syndrome.Report of a case and review of literature. West Indian Med J 43: 63-65](#)
10. [Memon SA, Mandhan P, Qureshi JN, Shairani AJ \(2003\) Recurrent Rapunzel syndrome – a case report. Med Sci Monit 9: 92-94.](#)
11. [De Backer A, Van Nooten V, Vanderplas Y \(1999\) Huge gastric trichobezoar in a 10 year old girl:case report with emphasison endoscopy in diagnosis therapy. J Pediatr Gastroenterol Nutr 28: 513-515.](#)
12. [Dumonceaux A, Michaud L, Bonnevalle M, Debeugny P, Gottard F, et al. \(1998\) Trichobezoar in children and adolescent. Arch Paediatr 5: 996-999.](#)
13. [Gorter RR, Knepekens CM, Mattens EC, Aronson DC, Heij HA \(2010\) Management of thrichobezoar: case report and literature review. Pediatr Surg Int 26: 457-463.](#)
14. [Huang YC, Liu QS, Guo ZH \(1994\) The use of laser ignited mini-explosive technique in treating 100 cases of gastric bezoars. Zhonghua Nei Ke Za Zhi 33: 172-174.](#)