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Rapunzel syndrome's silent cry: addressing neglect and psychiatric factors in pediatric cases

Arshia Warsi[1], Rabeea Mushtaq[2,3*], Syed Waqas Ali[4] and Farah Anwer[5]

Abstract

Introduction Rapunzel syndrome is a very rare form of trichobezoar. It is strongly linked to psychiatric conditions; trichotillomania and trichophagia. These conditions in the pediatric population point to the presence of childhood stressors like child neglect, abuse, etc. This case report highlights that child neglect may be an underlying contributing factor in selected cases of Rapunzel syndrome. Hence, a multidisciplinary approach should be sought in all cases of Rapunzel syndrome, catering to the emotional and psychological needs of the pediatric population.

Case presentation A 7-year-old girl from a low socioeconomic background presented with periorbital and facial swelling, abdominal distension, and pallor. On physical examination, she showed bilateral pitting edema and a distended abdomen with poor overall hygienic condition. Her family history was positive for Wilson's disease. Lab investigations highlighted iron deficiency anemia (Hb 8.2 g/dL), with normal liver function. Ultrasound demonstrated ascites, liver changes, and pleural effusion. The work-up for Wilson's disease was unremarkable. During the hospital course, her abdominal distention lessened; a non-tender epigastric mass (4 × 5 cm) was thus discovered. An abdominal X-ray revealed an entangled mass outlining the gastric shadow. Her parents also revealed a history of trichophagia. Suspecting a large trichobezoar, a laparotomy was performed. After the midline incision, the stomach was opened anteriorly between stay sutures along the curvature. It delivered a large trichobezoar, 65 cm in length extending up to the proximal jejunum. Rapunzel syndrome diagnosis was finally established.

Conclusion This unique presentation emphasizes the need to consider this rare etiology, even with unusual clinical histories especially with a history of child neglect, while collaborative surgical interventions facilitate successful outcomes.

Keywords Rapunzel syndrome, Trichobezoar, Wilson's disease, Child neglect

Background

Trichobezoar refers to the presence of hairballs within the gastrointestinal tract mostly in the stomach. Rapunzel syndrome is a very rare form of trichobezoar characterized by the presence of a large hairball in the stomach that extends into the small intestine reaching as far as the ileum, or colon in some cases. Trichobezoars are more common in young females and are strongly linked to psychiatric conditions, trichotillomania, and trichophagia [1]. According to DSM-V, trichotillomania is a repetitive and compulsive urge to pull one's hair and is classified as obsessive–compulsive and related disorders. Twenty to 30% of people with trichotillomania will end up suffering from trichophagia [2]. Few pieces of evidence suggest the

*Correspondence:
Rabeea Mushtaq
rabeeamushtaq77@gmail.com
[1] Anatomy Department, Dow University of Health Sciences, DIMC Ojha Campus, Karachi, Pakistan
[2] Karachi, Pakistan
[3] Forensic Department, Dow University of Health Sciences, DIMC Ojha Campus, Karachi, Pakistan
[4] Department of Paediatric Surgery, Dr. Ruth KM Pfau Civil Hospital, Dow University of Health Sciences, Karachi, Pakistan
[5] Dow University of Health Sciences, Karachi, Pakistan

association between trichotillomania and child neglect [3]. Pulling hair serves as a coping mechanism for Childhood stressors like neglect, abuse, etc. Child neglect, characterized by the failure to meet a child's basic physical and emotional needs, may be an underlying contributing factor in selected cases of Rapunzel syndrome [4]. By delving into the interplay between this extraordinary syndrome and the possibility of child neglect, we highlight the importance of a multidisciplinary approach to care, aiming to safeguard the well-being of the pediatric population. This case also provides insights into this rare syndrome's rare presentation, highlighting that it is often underdiagnosed due to its rarity and non-classical presentation.

Case presentation

A 7-year-old girl was admitted to the pediatrics department of Civil Hospital, Karachi via OPD with periorbital and facial swelling; and abdominal distension. No ongoing or previous history of fever was reported. There were no neurological or psychiatric complaints in the history. Parents belong to low socioeconomic backgrounds with five more kids. The eldest sibling was diagnosed case of Wilson's disease. On physical examination, there were no signs of malnutrition except pallor. The hygienic condition of the child was poor. Pulse and blood pressure were 100 bpm and 102/60 respectively. There was marked peri-orbital edema with bilateral pitting edema of lower limbs extending up to shins. The abdomen was grossly distended with an abdominal girth of 64.5 cm, no visceromegaly or abdominal mass was appreciated. The rest of the systemic examination was unremarkable.

Investigations

On lab investigations, hemoglobin was 8.2 g/dl, white blood cells 7.4×10^9/L, and platelets 294×10^9/L. Peripheral film and iron profile were consistent with iron deficiency anemia. Serum albumin was 1.7 g/dl. The basic metabolic profile, liver function test, lipid profile, bleeding time, and coagulation profile were normal. The urine analysis (microscopy and protein) report was normal. Ultrasound abdomen showed gross ascites, bilateral minimal pleural effusion, and coarse echotexture of the liver with normal liver span. Keeping in view the family history of Wilson's disease and ultrasound findings, serum ceruloplasmin level and 24-h urinary copper with challenge test were done and were unremarkable. The patient was on injection Lasix for symptomatic relief. On the fourth day of admission as edema lessened, a firm, mobile, non-tender, 4×5 cm mass was appreciated in the epigastric region on deep palpation. Abdominal X-ray showed a paucity of left-sided bowel gas shadow with entangled mass outlining the gastric shadow (see Fig. 1).

Upon inquiry, the parents revealed a prolonged history of trichophagia. Given the clinical history, all the above findings favored a very large trichobezoar as the possible culprit. Pediatric surgery was taken on board and decided to proceed with laparotomy without further investigation. Endoscopy was not attempted due to high failure rates associated with large trichobezoar [5]. After

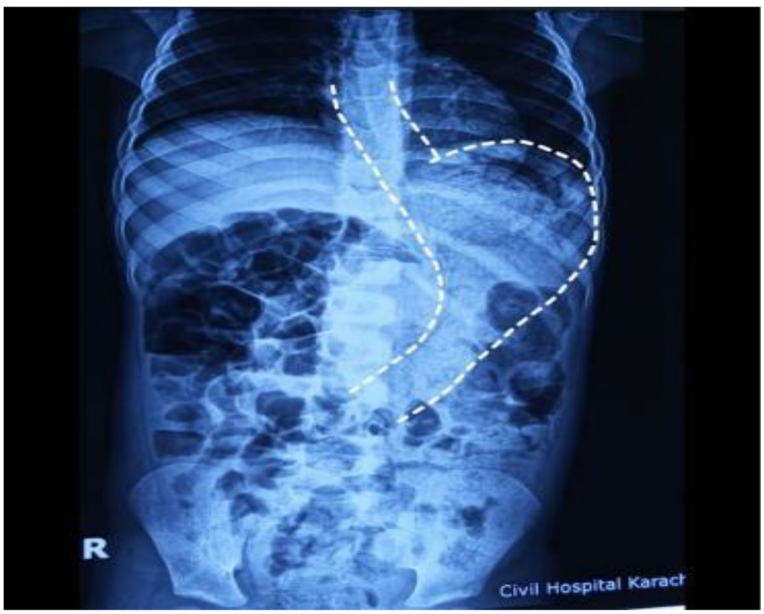

Fig. 1 Abdominal X-ray showing a paucity of left-sided bowel gas shadow with entangled mass outlining the gastric shadow

optimization with a transfusion of albumin and blood, the patient was taken for surgery. A midline laparotomy incision was made and the abdomen entered. An attempt was made to deliver the stomach in the wound but due to the huge size of the trichobezoar, it was not possible (see Fig. 2). The stomach was packed with iodine-soaked gauze on the sides and opened anteriorly between stay sutures along the curvature. The gastric bezoar was delivered followed by gentle distal manipulation and pulling of the tail which extended up to the proximal jejunum with a length of approximately 65 cm. The whole mass was removed intact (see Fig. 3).

The stomach was closed in two layers followed by layered closure of the abdomen. Postoperative recovery was uneventful. An in-hospital psychiatric review was taken. She was discharged on the 6th post-op day on full oral

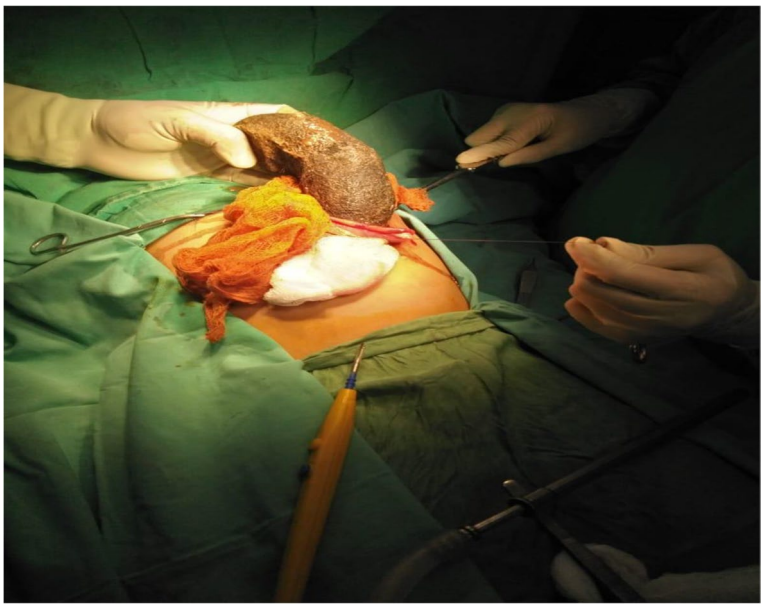

Fig. 2 Removal of trichobezoar during surgery

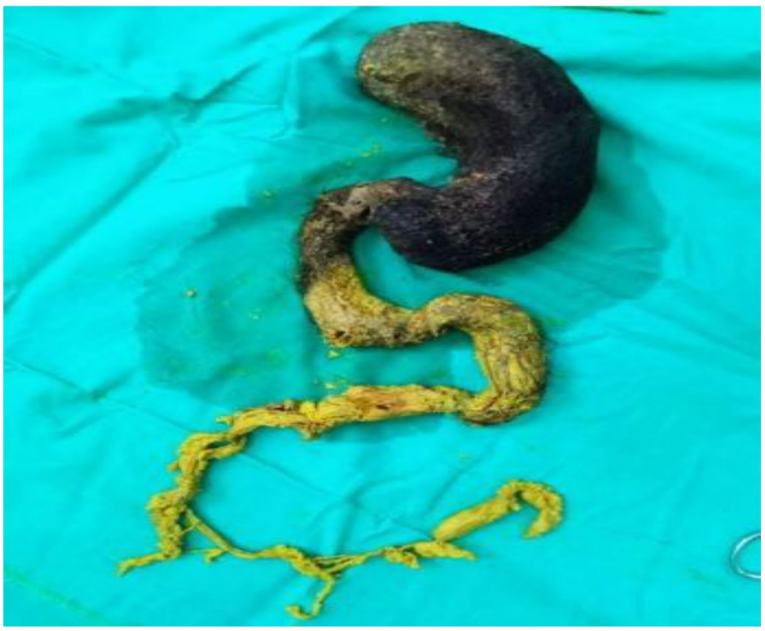

Fig. 3 65 cm Trichobezoar extracted

feed. She was advised of proper psychiatric follow-up. The last post-op follow-up at 1 month was uneventful.

Discussion

Most cases of trichobezoar present with non-specific symptoms like vomiting, constipation, abdominal pain, weight loss, and loss of appetite. Hence, they are often underdiagnosed and grow considerably to cause complications that include gastric outlet obstruction, hematemesis, gastric ulceration and perforation, intussusception, and protein-losing enteropathy [6]. If large, they can extend into the small intestine causing cholangitis, obstructive jaundice, and pancreatitis [7]. In two case reports, Rapunzel syndrome presented solely with iron deficiency anemia [8, 9]. Generalized edema in the pediatric population is also one of the rare presentations of this syndrome [10].

In our case, generalized edema was the initial presentation with no supporting clue to the diagnosis in the history and examination except alopecia patches. The child's parents were not inquired specifically about the history of trichophagia and trichotillomania initially. Although child neglect was suspected pertaining to the overall health of the child. Considering the history of Wilson's disease in the family, initial workup was directed towards it leading to a delay in the diagnosis, a true example of confirmation bias in medical diagnosis [11].

Rapunzel syndrome mostly arises in the context of psychiatric conditions—trichotillomania and trichophagia. In the pediatric population, hair-pulling behavior is mostly autonomic type, so children are often unaware of their act. Parents also do not pay much attention to this behavior. Psychiatric consultation for trichotillomania in the pediatric population is thus, rare, resulting in the development of secondary complaints.

In our case, no stressful life event or child abuse was identified, although child neglect was suspected due to the overall condition of the child and the presence of risk factors like poor socioeconomic status, chronically sick child in the family, and a large number of family members.

To confirm the diagnosis, imaging modalities like ultrasound and CT scan can be used. X-rays can also provide clues to the diagnosis. CT scan is the most commonly used imaging tool for the diagnosis with a higher detection rate than other imaging modalities. Nevertheless, upper gastrointestinal endoscopy is considered the gold standard for the diagnosis of trichobezoar [12]; however, we used X-rays along with clinical findings to reach a diagnosis.

While phytobezoars can be successfully treated medically using Coca-Cola or cellulase, no such treatment benefits trichobezoar. Endoscopic removal is another option. Various case reports have shown successful removal of trichobezoar in the pediatric population but still success rate of 5% has been reported [13]. It is associated with a higher complication rate causing ulceration, esophagitis, and esophageal perforation. Laparoscopic removal is an attractive option because it is less invasive and associated with shorter hospital stays. To date, many cases have been successfully treated [14]; however, it is not a widespread approach due to its complexity, increased risk of spillage of contents, and difficulty in looking for satellite lesions in the gastrointestinal tract. Laparotomy is considered the gold standard approach with a 100% success rate [15]. In Rapunzel syndrome, laparotomy is the safest and most commonly sought treatment approach. However, in the literature, the combined endoscopy-laparoscopy-gastrostomy approach in Rapunzel syndrome is also documented [16].

Conclusion

In conclusion, this case report provides insights into the rare presentation of Rapunzel syndrome and highlights the potential association between this syndrome and child neglect. A multidisciplinary approach to care, involving healthcare professionals from various specialties, is crucial for addressing the physical and psychological aspects of the patient's well-being. Early recognition and intervention can help ensure the proper management of Rapunzel syndrome. Further research and awareness are needed to enhance our understanding of the complex relationship between Rapunzel syndrome and child neglect, ultimately improving patient outcomes and promoting child welfare.

Abbreviations

CT Computed tomography
OPD Out patient department

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Authors' contributions

AW drafted the idea and concept, collected information, drafted the manuscript, approved the final version, and is responsible for her contributions. RM did the reviewing and editing, is the corresponding author as well, has approved the final version, and is responsible for her contributions. SWA provided with necessary patient information, did the editing, has approved the final version, and is responsible for his contributions. FA did the reviewing and editing, has approved the final version, and is responsible for her contributions. All authors read and approved the final manuscript.

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Declarations

Ethics approval and consent to participate
Patient information or clinical picture was not shared, hence, ethical approval is not needed.

Consent for publication
This manuscript has not been published in any journal and the authors are not willing to publish it in any other journal. The study did not violate any national or international laws on human, animal, and environmental rights.

Competing interests
The authors declare that they have no competing interests.

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References

1. Snorrason I, Ricketts EJ, Stein AT, Thamrin H, Lee SJJ, Goldberg H et al (2022) Sex differences in age at onset and presentation of trichotillomania and trichobezoar: a 120-year systematic review of cases. Child Psychiatry Hum Dev 53(1):165–171
2. Grant JE, Odlaug BL (2008) Clinical characteristics of trichotillomania with trichophagia. Compr Psychiatry 49(6):579–584
3. Yasir W, Sherin M, Farhana Kazmi S (2021) The role of childhood trauma in developing nail biting and trichotillomania among adolescents. Int J Innov, Creat and Change 15. Available from: www.ijicc.net
4. Ventura DE, MardirosHerbella FA, Schettini ST, Delmonte C (2005) Rapunzel syndrome with a fatal outcome in a neglected child. J Pediatr Surg 40(10):1665–1667
5. Gorter RR, Kneepkens CMF, Mattens ECJL, Aronson DC, Heij HA (2010) Management of trichobezoar: case report and literature review. Pediatr Surg Int. 26:457–463
6. Molinares-Pérez DA, Arraut-Gámez RE, Gómez-Barrios JD, Gómez-Cadavid N, Milian-Berrio MM (2022) Rapunzel syndrome associated with bowel obstruction and upper GI bleeding: case report and literature review. Cir Cir 90(6):848–852
7. Vellaisamy R, Iyer S, Chandramohan SM, Harikrishnan S (2020) Rapunzel syndrome with cholangitis and pancreatitis–a rare case report. Open Medicine 15(1):1137–1142
8. Ozkan AE, Danis N, Altundal M, Sevencan NO, Karatas F, Kayhan B (2020) An unusual cause of anaemia: Rapunzel syndrome, a case report. J Pak Med Assoc 70(7):1269–1271
9. Ancona G, Crea N, Scanu P, Batog I, Scurati G, Halim BA (2021) Acta scientific clinical case reports giant trichobezoar in young patient with severe anemia review of literature and psychological considerations case report
10. Nour I, AbdAlatef M, Megahed A, Yahia S, Wahba Y, Shabaan AE (2019) Rapunzel syndrome (gastric trichobezoar), a rare presentation with generalised oedema: case report and review of the literature. Paediatr Int Child Health 39(1):76–78
11. Elston DM (2020) Confirmation bias in medical decision-making. J Am Acad Dermatol. 82:572
12. Wang Z, Cao F, Liu D, Fang Y, Li F (2016) The diagnosis and treatment of Rapunzel syndrome. Acta Radiol Open 5(11):205846011562766
13. Iwamuro M, Okada H, Matsueda K, Inaba T, Kusumoto C, Imagawa A et al (2015) Review of the diagnosis and management of gastrointestinal bezoars. World J Gastrointest Endosc 7(4):336
14. Cintolo J, Telem DA, Divino CM, Chin EH, Midulla P (2009) Laparoscopic removal of a large gastric trichobezoar in a 4-year-old girl. J Society Laparoendosc Surg 13(4):608–611
15. Darwish AA, Abdelgawad AEM, Platt E (2019) Garrett-Cox R (2019) Formation of a temporary gastrostomy to aid delivery of gastric trichobezoar and decrease incidence of wound infection. BMJ Case Rep 12(6):e228660. https://doi.org/10.1136/bcr-2018-228660
16. Poerwosusanta H, Halim PG, Sitompul A, Wibowo AA (2020) Combined endoscopy-laparoscopy-gastrostomy extraction for Rapunzel syndrome. J Pediatr Surg Case Rep 1:61

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