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Case Report

An unusual case of Anasarca-Rapunzel syndrome

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ABSTRACT

Rapunzel syndrome is a rare form of gastric trichobezoar extending into the intestine, often associated with malnutrition and protein-losing enteropathy. A 12-year-old female presented with epigastric abdominal pain, facial puffiness, bilateral lower limb swelling, and anorexia. She was a known case of sickle-thalassemia double heterozygous state but had never received a blood transfusion. Family history revealed parental separation and harsh paternal behavior. The patient was undernourished but otherwise developed normally. On examination, she was lethargic, exhibited generalized anasarca, severe anemia, signs of congestive cardiac failure, and patchy hair loss on the scalp. Abdominal examination revealed moderate ascites, hepatosplenomegaly, and a firm, mobile, non-tender epigastric mass (10×8 cm), suggestive of gastric bezoar. Laboratory findings showed microcytic hypochromic anemia with macro-ovalocytes in the context of sickle-thalassemia heterozygosity. The child required multiple blood and serum albumin transfusions for severe anemia and hypoproteinemia with anasarca. Hepatic and renal causes of anasarca were excluded. Cardiovascular and respiratory systems were normal. Contrast enhanced computed tomography (CECT) abdomen confirmed the gastric bezoar. After stabilization, exploratory laparotomy and trichobezoar evacuation were performed. Psychiatric counseling and diversion therapy were provided. The child was discharged in stable condition, with follow-up showing significant improvement in nutritional status, mood, and general well-being. This case highlights the unusual presentation of Rapunzel syndrome with generalized anasarca in a pediatric patient with underlying sickle-thalassemia, emphasizing the importance of multidisciplinary management and follow-up.

Keywords: Rapunzel syndrome, Generalised edema, Hypoproteinaemia, Anaemia, Epigastric mass, Trichobezoar

INTRODUCTION

Bezoars, rare concretions of indigestible material in the gastrointestinal tract, present intriguing challenges in clinical practice. Among them, Rapunzel syndrome is exceptional, featuring a trichobezoar with a tail extending into the small intestine, reminiscent of the fairy tale maiden's long tresses.[1] This syndrome, uncommon in itself, is further complicated by its association with protein-losing enteropathy (PLE), leading to anasarca. Anasarca, generalized edema due to severe protein loss through the gastrointestinal tract, is a serious but rare complication of PLE.

In Rapunzel syndrome, this complication arises from both mechanical obstructions of intestinal lymphatic drainage and mucosal inflammatory injury caused by the enlarging trichobezoar.[2] We present a unique case of anasarca associated with PLE in a young girl diagnosed with Rapunzel syndrome. This case emphasizes the importance of recognizing and understanding the diverse clinical presentations of bezoar-related complications, especially in pediatric patients. It also underscores the need for a multidisciplinary approach involving gastroenterologists, surgeons, psychiatrist and nutritionists in diagnosis and management. Through detailed description and analysis, our aim is to contribute to the literature on Rapunzel syndrome, elucidating its rare yet significant implications and stressing the importance of early recognition and intervention to prevent severe complications such as anasarca.

CASE REPORT

A 12-year-old female child, XYZ, presented with complaints of epigastric abdominal pain, facial puffiness, bilateral lower limb swelling, reduced urine output, anorexia, and easy fatigability for the past 15 days. There was no history of fever, diarrhoea, vomiting, or yellowish discoloration of the skin. Past medical history revealed a recent diagnosis of sickle-thalassemia double heterozygous state following a bout of viral hepatitis A infection one month prior to admission. However, there was no history of previous blood transfusions. The child also had a history of pica, specifically craving for mud and chalk. Family history indicated parental disputes and separation for 3 years, along with reports of harsh behaviour from the father towards the child. The mother had never observed the child pulling her hair and consuming it, and the patient herself was unaware of such behaviour. Despite these challenges, the child exhibited normal developmental milestones and average scholastic performance. The mother was found to be a carrier of the sickle cell trait. The patient's body weight and height were below the third percentile for her age and gender, suggestive of undernutrition, with a body mass index (BMI) of 12.5 kg/m^2. She was in stage 2 Tanner sexual maturity rating. Her diet displayed a prolonged daily deficit of 55% calories and 56% protein. On general examination, the child appeared lethargic and apathetic but was well-oriented to time, place, and person. Vital signs revealed tachycardia (130 beats/min) and tachypnea (34 breaths/min). Peripheral perfusion was adequate with warm extremities and bounding peripheral pulses. Physical examination revealed severe pallor and xerosis of the skin, patchy areas of hair loss on the scalp, puffy eyelids, and bilateral soft pitting edema (grade 3) of both lower limbs extending up to the thighs. Abdominal examination revealed a distended abdomen with a smiling umbilicus, horseshoe dullness indicating moderate ascites, a soft 5 cm liver with mild tenderness, and a firm 6 cm spleen (Hacket grade 3). Notably, there was a well-demarcated epigastric mass (10×8 cm) that was firm, mobile, non-tender, and dull on percussion.

Management and outcome

Laboratory investigations revealed a hemoglobin level of 6 gm/dl, a total leukocyte count of 33000/cu mm, and a platelet count of 2.24 lakh. Retic count was 1%. C-reactive protein (CRP) was elevated at 100. Peripheral smear examination indicated moderate anisopoikilocytosis with mildly microcytic and hypochromic red blood cells, along with a few macro-ovalocytes and sickle cells. Serum B12 level was 186 pg/ml. Other biochemical parameters including blood urea, serum creatinine, electrolytes, bilirubin, liver enzymes, and coagulation profile were within normal limits. Urine examination was unremarkable. Total protein was 4.5 gm/dl and serum albumin were 2.5 gm/dl. There was no evidence of hyperlipidemia. A normal 2D echocardiogram ruled out cardiac involvement. Ultrasonography of the abdomen revealed a large echogenic mass lesion with strong posterior acoustic shadowing within the stomach, accompanied by moderate ascites. Contrast-enhanced computed tomography (CECT) of the abdomen and pelvis confirmed the presence of a large, well-defined hypodense lesion with interspersed mottled air foci in the stomach, along with edematous bowel wall thickening involving the jejunal loops. Gastrointestinal endoscopy was planned for the examination and attempted evacuation of the bezoar contents, but due to the child's fragile condition and the large size of the bezoar, the procedure was deferred.

Treatment involved serum albumin transfusions for hypoproteinemia and packed red blood cell transfusions for severe anemia. Despite a transient resolution of ascites and facial puffiness, anasarca reappeared. Eventually, the patient underwent exploratory laparotomy with the successful evacuation of a large trichobezoar measuring 60 cm in length. Psychiatric consultation was sought, and counselling, along with diversion therapy, was initiated for the caregivers. The patient was discharged upon resolution of edema and improvement in the overall clinical condition. Follow-up over 3 months showed weight gain and continued psychological counselling for both the patient and the parents.

Figure 1: Patchy hair loss over scalp.

Figure 2: Evacuated mass of trichobezoar with its long tail (total length-60 cm).

DISCUSSION

Trichotillomania, a psychiatric condition marked by recurrent hair pulling leading to hair loss and functional impairment, presents unique challenges in diagnosis and management.[3] While it predominantly affects females in adulthood, the sex distribution in childhood is equal. Associated psychosocial dysfunction, low self-esteem, and social anxiety often result from an inability to control hair pulling and subsequent alopecia.[4,5] Trichotillomania frequently coexists with other psychiatric disorders such as major depressive disorder, anxiety disorders, and substance use disorders.

Hair pulling in trichotillomania can be either focused, as a coping mechanism for tension or distress, or automatic, occurring without conscious awareness during passive activities. The diagnosis of trichobezoar, a collection of indigestible materials often composed of hair, should be considered in individuals who exhibit symptoms such as early satiety, nausea/vomiting, abdominal pain, or unexplained weight loss, particularly if they have a history of hair ingestion.[6]

Diagnostic evaluation typically includes abdominal examination, blood tests for anemia, and abdominal CT scan, which is diagnostic in the majority of cases.[3] Initial management may involve enzymatic debridement or chemical dissolution, although trichobezoars are typically resistant to chemical dissolution. Endoscopic fragmentation or surgical removal may be necessary if conservative measures fail.[6,7]

CONCLUSION

In summary, trichotillomania and trichobezoar represent distinct yet interconnected clinical entities that require a comprehensive approach to diagnosis and management. Understanding the psychosocial implications of trichotillomania and recognizing its potential complications, including trichobezoar formation, are essential for providing optimal care to affected individuals.

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REFERENCES

1. Stinco M, Montemaggi A, Noccioli B, Resti M, Grosso S, Trapani S. An Unusual Case of Hypoproteinemia in Childhood: Keep in Mind Trichobezoar. Front Pediatr. 2020;8.
2. Nour I, Abd Alatef M, Megahed A, Yahia S, Wahba Y, Shabaan AE. Rapunzel syndrome (gastric trichobezoar), a rare presentation with generalised oedema: case report and review of the literature. Paediatr Int Child Health. 2019;39(1):76-8.
3. King RA, Zohar AH, Ratzoni G, Binder M, Kron S, Dycian A, et al. An Epidemiological Study of Trichotillomania in Israeli Adolescents. J Am Acad Child Adolesc Psychiatry. 1995;34(9):1212-5.
4. Diefenbach GJ, Tolin DF, Hannan S, Crocetto J, Worhunsky P. Trichotillomania: Impact on psychosocial functioning and quality of life. Behav Res Ther. 2005;43(7):869-84.
5. Swedo SE LH. Tricotillomania: an obsessive compulsive spectrum disorder? Psychiatry Clin North Am. 1992;1461795.
6. Mahvi DA, Mhavi DM. Sabiston Textbook of General Surgery: Biological basis of Surgery. 21st Edition. Townsend M, Daniel Beauchamp R, Mark Evers B, Mettox KL, Editors. Canada: Elsevier. 2021;1236-7.
7. Grant JE, Dougherty DD, Chamberlain SR. Prevalence, gender correlates, and co-morbidity of trichotillomania. Psychiatry Res. 2020;288:112948.

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