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Association of Pediatric Rapunzel Syndrome with Protein-Losing Enteropathy

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Abstract

Rapunzel Syndrome describes the rare presentation of a trichobezoar with extension of ingested hairs through the pylorus into the small intestine. In this report, we describe the case of a 4-year-old girl presenting with anasarca found to have hypoalbuminemia, severe malnutrition and Protein-Losing Enteropathy (PLE). Further questioning revealed a history of trichotillomania and trichophagia. An upper GI series demonstrated findings of a trichobezoar extending into the proximal duodenum, consistent with Rapunzel Syndrome. An exploratory laparotomy with anterior gastrotomy removed an extensive trichobezoar. The patient subsequently tolerated advancement of diet with improvement in total serum protein levels. This signifies a rare presentation of PLE in the setting of Rapunzel Syndrome, as well as the youngest reported case of Rapunzel Syndrome to occur in the United States.

Introduction

First described by Vaughan et al. in 1968, the aptly named "Rapunzel Syndrome" describes the presentation of a gastric trichobezoar with extension of the ingested hairs through the pylorus into the small intestine [1]. This long tail of hair is reminiscent of that which Rapunzel used to lift up her prince in the traditional fairytale by the Grimm brothers [2]. Since Vaughan's initial reporting, there have been fewer than 40 pediatric cases of Rapunzel Syndrome cited in the literature with typical presenting features including abdominal pain, nausea/vomiting, obstruction, peritonitis, weight loss and a mean age of 10.8 years [3-5]. In this case report we discuss an unusual presentation of Rapunzel Syndrome in a 4 year old female with anasarca, hypoalbuminemia, severe malnutrition and protein-losing Enteropathy. This represents the youngest case of Rapunzel Syndrome in the United States [6].

Case Presentation

A 4-year-old African American girl presented from outpatient clinic with 3 days of periorbital and lower extremity swelling in the setting of 1 week of watery, malodorous stool and non-bloody, non-bilious emesis. On examination she appeared generally malnourished and withdrawn with grade 1 pitting pedal edema, a tender, distended abdomen and thin extremities. Initial laboratory workup revealed hypoalbuminemia of 1.0 gm/dL (3.6 to 5.2), hypoproteinemia of 3.3 gm/dL (6.0 to 7.8), and a normal urinalysis. Infectious gastroenteritis workup was negative. Abdominal ultrasound showed dilated fluid filled loops of bowel with associated colonic wall thickening.

Dietary history revealed a poor and restrictive diet over the preceding 2 years typically consisting of bread, tea, and 2 cups of 2% milk per day. Further work-up demonstrated iron deficiency anemia (hemoglobin 9.8 gm/dL,10.2 to 12.7) with a serum iron of 14 mcg/dL, low pre-albumin (5.7 mg/dL, 16.4 to 32.0), vitamin D deficiency (25-Hydroxy Vitamin D <4 ng/mL, 30 to 100), zinc deficiency (44 mcg/dL, 48 to 119), and a normal folate level.

Patient was admitted directly from clinic to the general pediatric service for treatment of presumptive acute gastroenteritis in the setting of poor chronic protein dietary intake. Due to the presence of edema in association with hypoalbuminemia, a 25% albumin infusion of 0.5 g/kg with furosemide was given with subsequent improvement in edema, though albumin levels only transiently improved. Emesis and diarrhea resolved within 5 days of admission without

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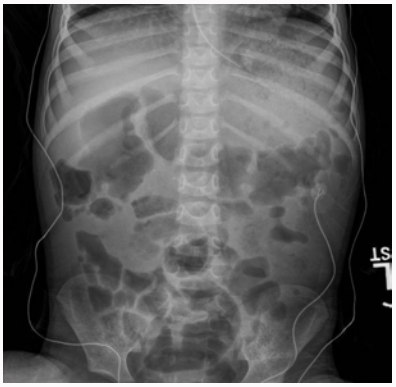

Figure 1: A portable 1 view abdominal radiograph obtained secondary to difficulty with NG tube placement revealed enteric tube terminating early in the region of the gastro-esophageal junction. Haziness over the gastric cavity was later noted.

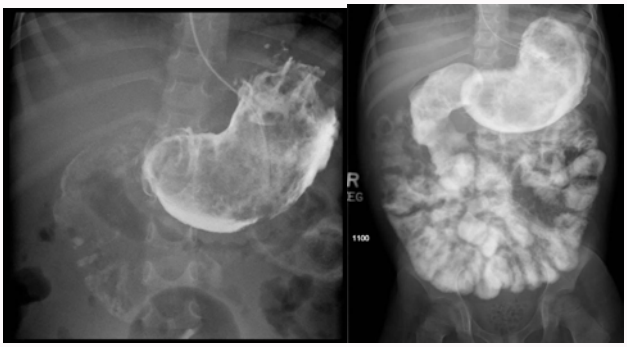

Figure 2: An upper GI fluoroscopy with serial images was completed demonstrating a large amount of intraluminal material occupying the stomach (A) with apparent extension into the distal duodenum and proximal jejunum (B) compatible with a bezoar.

intervention. However, patient continued to have abdominal distention with limited oral intake. Persistent feeding refusal despite resolution of gastroenteritis symptoms, in addition to a concern for chronic malnutrition, led to subsequent placement of a Nasogastric (NG) tube and initiation of supplemental feeds. Further investigation into the patient's dietary history demonstrated a likely adequate protein intake (given milk consumption) of approximately 1.14 g/kg. Thus, poor protein intake was unlikely the sole etiology of hypoalbuminemia and hypoproteinemia. Supplementary workup resulted with elevated stool alpha-1 antitrypsin (>1.13 mg/gm, 0 to 0.50) confirming suspicion of Protein-Losing Enteropathy (PLE) though the underlying etiology remained unclear. Fecal calprotectin was also elevated at 376.2 mcg/g (normal range: <162.9) suggesting an inflammatory process, presumably related to an acute infectious etiology. Differential diagnosis for PLE at that time included enteric infections, hypertrophic gastritis (Ménétrier disease), inflammatory bowel disease, Celiac disease, and intestinal lymphangiectasis. An abdominal radiograph revealed a coiled NG tube at the gastroesophageal junction with a hazy opacification corresponding to the gastric cavity (Figure 1). Coinciding abdominal exam was concerning for increasing abdominal distension and a palpable epigastric mass.

At this juncture, additional questioning elicited a history of trichotillomania and trichophagia, leading to concern for a trichobezoar. An upper GI series demonstrated large amounts of

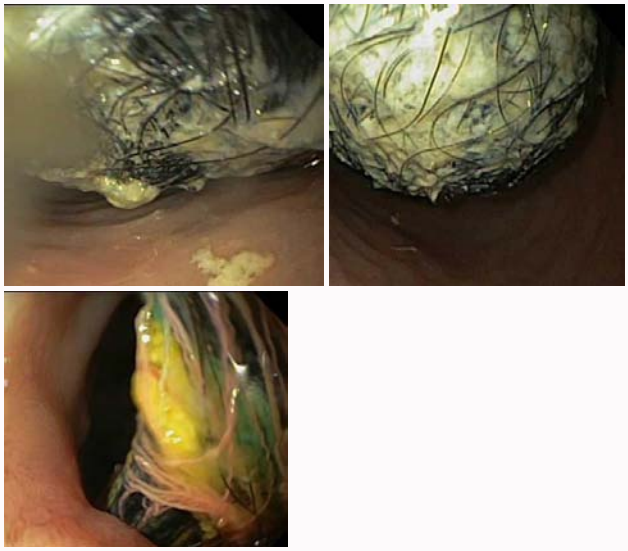

Figure 3: Esophagogastroduodenoscopy revealed a large trichobezoar occupying the entire stomach (A,B) and extending through the pylorus into the duodenum (C).

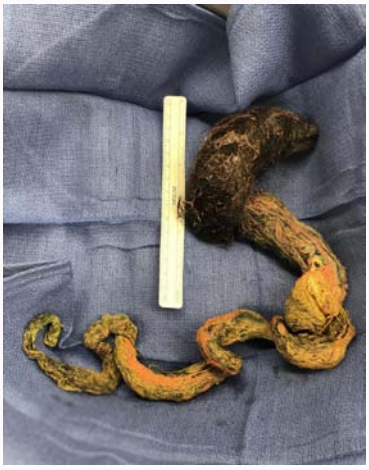

Figure 4: A trichobezoar measuring 14.7 cm × 12.1 cm × 5.1 cm was removed via exploratory laparotomy. It was composed of dark brown hair and colored fabric-like sutures.

intraluminal material consistent with a bezoar (Figure 2A, 2B). Findings were confirmed with an esophagogastroduodenoscopy revealing a large trichobezoar occupying the stomach and extending through the pylorus into the proximal duodenum, consistent with Rapunzel Syndrome (Figure 3A-3C). Duodenal biopsies demonstrated distortion of the glandular architecture, villous blunting, decreased number of goblet cells and no Paneth cells. Gastric mucosa biopsies showed chronic, focally active gastritis and immunostaining detected rare *Helicobacter pylori* organisms. An interdisciplinary General Surgery, Gastroenterology, and Hospitalist team elected to provide Total Parental Nutrition (TPN) for 2 weeks prior to surgical intervention to optimize patient's nutritional status and subsequent post-surgical outcomes.

An exploratory laparotomy with anterior gastrotomy removed an extensive trichobezoar measuring 14.7 cm × 12.1 cm × 5.1 cm and composed of severely matted, dark brown, morcellated hair with multiple colored, fabric-like sutures (Figure 4).

Post-operatively, the patient was able to tolerate slow advancement of enteral nutrition that allowed TPN to be weaned and corresponded to improved total protein (5.8 gm/dL) and albumin (2.1 gm/dL) levels. She was discharged home on post op day #12 with oral vitamin D, ferrous sulfate and zinc supplements with plans for follow-up for feeding therapy evaluation and ongoing psychiatric therapy given her severe trichophagia.

Discussion

We describe an unusual case of a 4 year old female presenting with PLE secondary to an extensive gastroduodenal trichobezoar, characteristic of Rapunzel Syndrome. The youngest previous case of Rapunzel Syndrome in the United States occurred in a 5-year-old girl with developmental delay presenting with abdominal pain and emesis [6]. The complexity and atypicality of our case is several fold. Our patient's initial presentation of vomiting and diarrhea, likely an acute gastroenteritis, was inconsistent with the degree of hypoalbuminemia and anasarca present which led to further work-up and eventual diagnosis of PLE. PLE is rarely the presenting symptom of a trichobezoar, as oppose to abdominal pain, obstruction, peritonitis, or acute weight loss. Additionally, her extremely young age, neurotypical development, and lack of prior psychiatric diagnoses differs from other cases of Rapunzel Syndrome described in the literature.

PLE is a collection of diseases characterized by excessive loss of protein into the gut with subsequent development of hypoproteinemia. Typical presentation is edema with complications including as cites and pleural/pericardial effusions. Diagnosis of PLE is through identification of increased fecal concentrations of alpha-1-antitrypsin, a reliable endogenous marker of enteric protein loss given its stability in the gastrointestinal tract and similar size to albumin [7]. In our patient, the early finding of elevated stool alpha-1-antitrypsin lead to the diagnosis of PLE though the underlying cause of PLE initially remained elusive.

Causes of PLE are varied but can be divided into 3 categories: Increased interstitial pressure, erosive gastrointestinal disease and non-erosive gastrointestinal disease [8]. Increased interstitial pressure can be seen in cardiac conditions for example post-fontan procedure or in relation to lymphatics such as in intestinal lymphangiectasia. Erosive GI diseases include inflammatory bowel diseases with protein loss resulting in PLE secondary to leakage of proteins through the eroded epithelium. Non-erosive GI diseases include both infectious gastroenteritis and Menetrier's disease, which lead to PLE through excessive protein loss due to malabsorption or surface epithelial cell loss. Menetrier's disease is a protein-losing hypertrophic gastropathy characterized by gyriform enlargement of gastric mucosal folds which leads to hypoproteinemia through excess protein-rich mucous secretion [9].

Menetrier's disease with a coinciding trichobezoar has been previously reported in 4 cases [9-11]. In each of these cases, the finding of a trichobezoar was associated with hypertrophic gastropathy and PLE. However, our patient lacked the typical endoscopic findings of Menetrier's disease. This suggests an alternative relation between trichobezoars and PLE independent of hypertrophic gastroenteropathy.

The etiology of our patient's PLE was likely multifactorial. First, a presumed infectious gastroenteritis was supported by the acuity of diarrhea, emesis, and an elevated stool calprotectin. Another infectious etiology pertinent to our case is *H pylori* gastritis though noted to be an inconspicuous pathologic feature. A more plausible explanation is that the large trichobezoar itself contributed to the PLE primarily by direct mucosal trauma and irritation. The trichobezoar can also cause a component of gastrointestinal lymphatic obstruction similarly seen in congenital lymphangiectasia, which then leads to increased protein leakage. Previously, Hossenbocus et al. [12] hypothesized that the presence of a gastric trichobezoar induced gastric polyp formation with subsequent irritation and PLE. Our patient's duodenal pathology was especially remarkable for villous blunting, raising an interesting differential diagnosis to include acute and chronic enteropathies. We believe these findings represent acute (infection) on chronic effects of an obstructing mass, malabsorption and malnutrition. The diagnosis of autoimmune enteropathy was suggested by our pathologist due to loss of Paneth cells, and decreased goblet cells, but overall lacked typical clinical features of intractable diarrhea with TPN dependence.

Conclusion

Rapunzel syndrome is an uncommon diagnosis with fewer than 40 reported pediatric cases worldwide. We discuss a novel presentation of Rapunzel syndrome in an atypically young child with protein-losing enteropathy, which led to an instructive and thought provocative process of evaluation and multidisciplinary management. PLE has previously been described in association with trichobezoars; however, this was always in the setting of hypertrophic gastroenteropathy or polyposis. In this case, we hypothesize alternative etiologies of PLE including foreign body induced lymphatic obstruction and local mucosal irritation.

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