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Case Report

Rapunzel syndrome and gastric Peutz Jeghers (hamartomatous) polyps: case report

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ABSTRACT

Rapunzel syndrome is an extremely rare complication of a trichobezoar. These females have a history of trichophagia and trichotillomania. Peutz Jeghers syndrome (PJS) is an uncommon autosomal dominant syndrome with a variable to high penetrance that leads to the development of the polyps within the gastrointestinal mucosa. This case report of a 25-year-old deaf and dumb female presented with pain and lump in upper abdomen, vomiting, nausea, loss of appetite, loss of weight. An exploratory laparotomy with anterior gastrotomy was performed and a giant trichobezoar with tail extending into the duodenum was removed. There were multiple polyps in the lower stomach excised. Histopathology reports showed Peutz Jeghers (hamartomatous polyps). She recovered well and was discharged on the 10th day. Patient was advised regular follow up.

Keywords: Rapunzel syndrome, Giant trichobezoar, Trichotillomania, Trichophagia, Peutz Jeghers (hamartomatous) polyps, Anterior gastrotomy

INTRODUCTION

Gastric trichobezoar is a mass or collection of undigested and accumulated own hair into the stomach.[1] Trichobezoar are usually the result of compulsive pulling out (Trichotillomania) hair and eat one's own hair (trichophagia). Trichobezoar occurs commonly in adolescent females with underlying psychiatric disorders.[2-4] The name Rapunzel syndrome comes from the Grimm Brothers' fairytale of a 12-year-old princess who was shut into a tower with neither stairs nor doors by an enchantress who climbed up the tower's walls with the help of Rapunzel's long tresses.[5]

Peutz Jeghers syndrome is a rare autosomal dominant syndrome only few numbers of cases have been reported in literature. This syndrome consists of the association of gastrointestinal polyps, mucocutaneous pigmentation and a familial incidence. In PJS the GIT develops hamartomatous polyps, most commonly in the small bowel, colon and rectum (> 90% of cases) and less commonly in the stomach. Here, we report a very rare case of a female suffering from Rapunzel syndrome with Peutz Jeghers (hamartomatous) polyps.

CASE REPORT

A 25-year-old deaf and dumb woman who is mentally subnormal having 1-year-old child admitted in general surgery ward with complaints of pain in abdomen with lump in upper abdomen, nausea, vomiting, anorexia, weight loss 10 days before admission. Her parents give history that, from 5 years of age she has been pulling her hair and ingesting it (trichophagia). Clinical examination revealed a malnourished lady having well defined mass occupying the epigastric region upto the umbilicus. Up and down mobile, side to side restricted mobility. Mass firm, tenderness present. Laboratory test results showed- Hb –

11 gm/dl, hypoproteinaemia (total 4 gm/dl and albumin – 2.35 gm/dl) with normal liver function test.

Ultrasonography (USG) abdomen – E/O big brightly Echoic lesion with acoustic shadowing in stomach region s/o foreign body in stomach. The diagnosis was made at upper GI scopy. Ball of hair occupying larger portion of stomach. Thickened gastric folds, reduced distensibility, pylorus- not visualised. Impression –Trichobezoar (Figure 1).

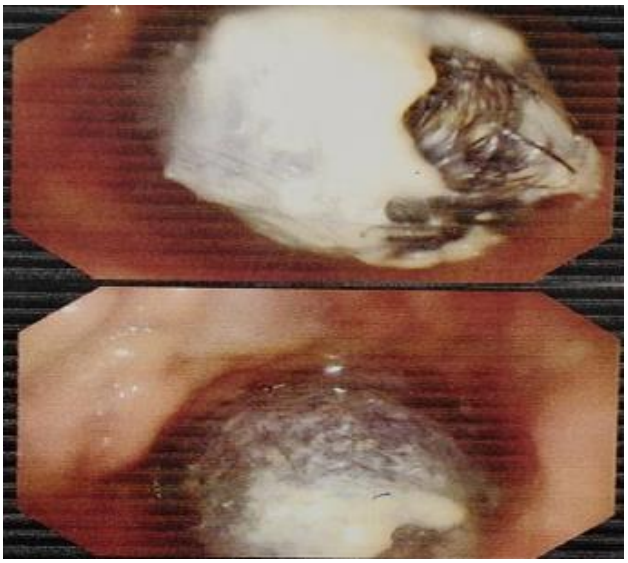

Figure 1: OGD scopy showing Trichobezoar.

Exploratory laparotomy with anterior gastrotomy done. There was big trichobezoar occupying near about whole stomach and entering in the duodenum (Figure 2).

Figure 2: Trichobezoar specimen.

There were multiple gastric polyps in the lower part of stomach, excised (Figures 3 and 4).

By these features, the diagnosis was clear of Rapunzel syndrome with gastric polyps and polyps were send for histopathology (Figures 5 and 6). The gastrotomy was closed with continuous 3-0 prolene (Figure 7). The patient had an uneventful postoperative period and discharged on 10th day.

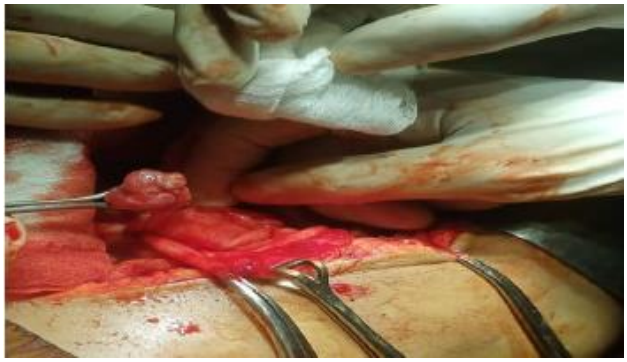

Figure 3: Gastric polyp.

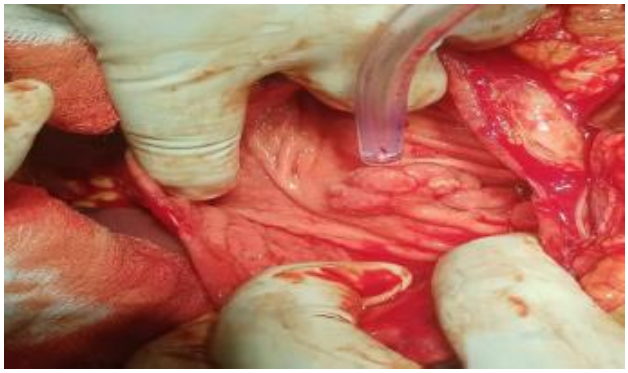

Figure 4: Gastric polyp.

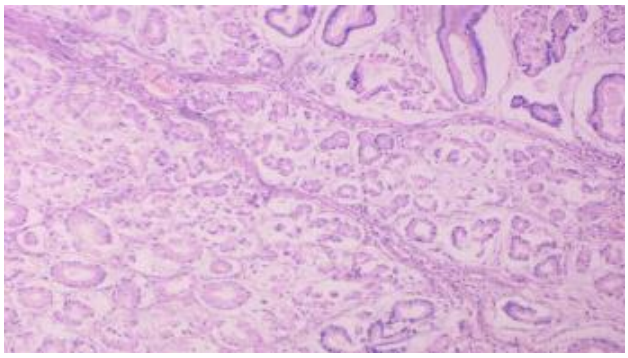

Figure 5: HPE 10 X showing christmas tree appearance. Peutz Jeghers (hamartamatous polyp).

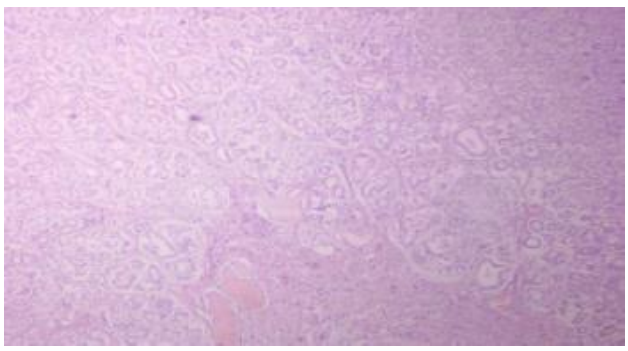

Figure 6: HPE 4X showing projection of mucosa with muscularis mucosa at base. Peutz Jeghers (hamartamatous polyp).

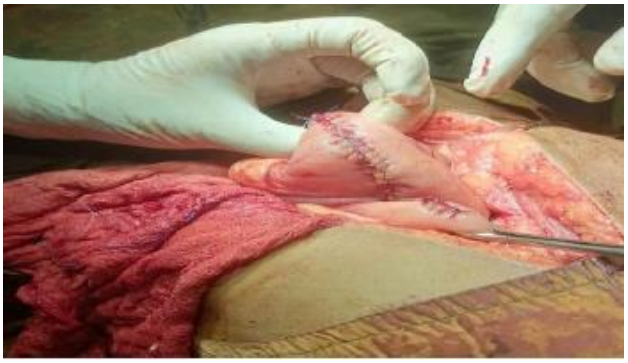

Figure 7: Anterior gastrotomy was closed with prolene 3-0 sutures.

DISCUSSION

Bezoars are concretions of foreign material in the gastrointestinal tract. They are classified into Trichobezoars (by hair), phytobezoars (vegetables), lactobezoars (dairy products), pharmacobezoars (medicine), according to substance ingested.[1] Trichobezoar is a ball of swallowed hair that collects in the stomach and fails to pass through the intestine. They are confined to the stomach in most of the cases. However, rarely there is continuous extension of the trichobezoar beyond pylorus, such condition is termed as Rapunzel syndrome. Near about hundred cases of Rapunzel syndrome have been reported in the literature. Trichobezoars commonly occur in females, often with underlying psychiatric disorder.[2]The presentation may vary, and these patients may not give history due to social stigma. Trichobezoars are usually the result of compulsive pulling out and eating one's hair. The disorder is predominantly observed in adolescents, more frequently in girls than boys.[3,4] Trichobezoar ineffectively moved by peristalsis due to its smooth surface and poorly digested keratinaceous substance. As a result, the hair becomes matted into a ball and is retained in the stomach. The ball can reach sizes sufficient to cause distension of stomach and malnutrition.[5]

Most common presenting symptom include abdominal pain, nausea, vomiting, occurs in 35% of patients [5] on clinical examination, epigastric masses are seen in 85% of patients. Anemia and hypoalbuminaemia have been described.[6] Less commonly trichobezoar may cause small bowel obstruction, pancreaticobiliary obstruction.[7,8] Most of the patients deny the history of trichotillomania and trichophagia even when specifically asked. The alopecia may be noted. In this case, the parents of the lady admitted her having habit of eating her hair, since the age of five years. The patient is deaf and dumb so can't give history.

Endoscopy is the choice of examination in the diagnosis of gastric trichobezoar as it allows visualizing the hair strands. Computed tomography (CT)- scan delineate a well-defined, oval intra luminal mass with air bubbles retained within the intestine or homogeneous mat appearance in the region of the stomach or intestine.[9]

In this case, the patient was mentally subnormal, deaf, so she was neglected, and no attention given. She got married, but her husband also left her with a child. Due to continuous vomiting, she was brought to the hospital. Peutz jegher syndrome (PJS) is an unusual hamartomatous polyposis of gastrointestinal tract with pigmentation around lips and macules on the buccal mucosa.PJS is a rare autosomal dominant syndrome and few cases have been reported in literature.[10] PJS affects around 1:83000 to 200,000 births.[11] PJS was first reported by Peutz in 1921 and subsequently detailed case PJS by Jeghers McKusik and Katz in 1949.[12-14] In PJS, the GI tract develops hamartamatous polyps, which occur most commonly in the small bowel, colon and rectum. (> 90% of cases) and less commonly in the stomach or urinary tract.[15] Hamartomas can range from 5 to 50 mm in diameter. Hamartomas are associated with intussuception, bleeding, anaemia, and obstruction.[16] Here, we are presenting a rare case of big trichobezoar with Rapunzel syndrome and having multiple polyps (hamartomatous) (Figures 3-6).

In this case, only gastric hamartomatous polyps, no mucocutaneous pigmentation, no familial history of PJS. All polyps more than 1 cm are excised. Most publications recommended polypectomy for polyps in the stomach or colon greater than 1 cm in size. We didn't find Rapunzel syndrome with hamartamatous polyps published in literature.

We report this case of giant gastric trichobezoar and Rapunzel syndrome with gastric Peutz Jegher (hamartamatous) polyps which is very rare and maybe the first case.

CONCLUSION

Rapunzel syndrome is an extremely rare condition. Trichobezoar should be considered as a differential diagnosis in a young female with epigastric mass. As in our case upper GI endoscopy diagnosed trichobezoar but not polyps. In giant trichobezoar treatment of choice is exploratory laparotomy with anterior gastrotomy when gastric polyps are present excise all polyps more than 1 cm. The patient should be called regularly for follow-up, and as in our case as patient is deaf and dumb with mentally subnormal, parents should be counselled to take care of the patient and further follow-up.

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