Peutz–Jeghers syndrome (PJS) is inherited as an autosomal dominant disorder presenting as hamartomatous polyps in the small bowel, mucocutaneous pigmentation and with a predisposition to develop cancer. We report a case of PJS, with an adenomatous giant gastric polyp. The purpose is to highlight that adenomatous giant gastric polyp may be an extremely rare presentation of PJS. Awareness of this possibility will help us in not missing out these atypical cases of PJS.

**Keywords:** Adenomatous polyp, giant gastric polyp, hamartomatous polyp, Peutz–Jeghers syndrome

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**INTRODUCTION**

Peutz–Jeghers syndrome (PJS) is a condition inherited in an autosomal dominant manner with features of gastrointestinal polyps, mucocutaneous pigmentation, and predisposition to malignancy.[1] The polyps found in PJS are predominantly hamartomatous in nature but may also be adenomatous or hyperplastic, as also they are most commonly found in the small intestine but rarely, they may also be located in the stomach, duodenum, colon, or even other extra-intestinal locations such as the renal pelvis and gall bladder.[2] We hereby report a case of PJS, highlighting the atypical presentation of PJS in the form of a giant gastric adenomatous polyp.

**CASE REPORT**

Our case was a 2-year-old boy who was incidentally diagnosed with anemia during the evaluation of an upper respiratory tract infection. There was no history of hematemesis, melena, or abdominal pain. The child was evaluated by the pediatric gastroenterologist with an upper gastrointestinal endoscopy and was found to have a large sessile gastric polyp that was not amenable to endoscopic resection. Physical examination was insignificant except for a few melanotic spots over the lower lip [Figure 1]. Radiological imaging revealed a large hyper-enhancing mass lesion in the lumen of the stomach, measuring about 8 cm in greatest dimension along with multiple small intestinal polyps.

Laparotomy revealed a large sessile polyp involving the greater curvature and posterior wall of the stomach measuring about 10 cm × 8 cm along with multiple small sessile polyps involving the corpus of the stomach sparing the antrum and pylorus, requiring partial gastrectomy [Figure 2]. There were two more polyps, one each in the duodenum and distal jejunum in their antimesenteric border and they could be excised by enterotomy. The child had an uneventful postoperative course. The histopathology revealed the features of tubulovillous adenoma in all the resected polyps. At 36 months of follow-up, the child is asymptomatic and disease-free.

**DISCUSSION**

The sine qua non of PJS diagnosis is the hamartomatous polyp, which is characterized on histopathology by distinctive interdigitating smooth muscle bundles in a typical arborizing pattern throughout the lamina propria. Rarely, the polyps may also be adenomatous which may, therefore, lead us to the diagnosis of familial adenomatous polyposis syndrome, as this happens mostly with polyps arising in the colon with PJS.[3] In such cases, if there are no family history or external sign, the diagnosis of PJS may be difficult to establish.

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Melanotic spots in our case were unmissable, and hence, it was PJS despite the atypical histology.

The inheritance pattern of this syndrome is autosomal dominant with the involvement of a variant of STK11. However, approximately 45% of affected individuals have no family history of PJS, and in them, it is a de novo mutation that is responsible for this phenotype. In our case, it seems prudent to speculate that it was a de novo mutation since a positive family history was lacking. A formal analysis of the STK11 gene has not been done in our case.

The most common types of gastric polyps in children are fundic gland polyps, hyperplastic polyps, and adenomas. The prevalence of gastric polyps is even less in children as compared to adults (0.7% vs. 6.3%). Giant gastric polyps are very rare, more so in PJS. Few case reports of giant gastric polyps do exist, but mostly they are in adults. The largest gastric Peutz–Jeghers-type polyp was reported by Lunca et al., measuring 150 mm × 70 mm × 50 mm, and the second-largest polyp of this category was reported by Zou et al. that measured 110 mm × 80 mm × 4 mm in 2017. Both the cases were adults, and they were reported in a 53-year-old male and a 43-year-old female, respectively. It is worth noting that a giant gastric polyp in PJS is extremely rare, and to our belief, this is the first case being reported from the pediatric age group.

Gastric polyps are amenable to endoscopic polypectomy. However, the large size of the gastric polyp posed a unique challenge in the management. Endoscopic polypectomy or mucosal resection was technically challenging due to the large size and sessile nature of the polyp. Open surgical excision was therefore performed. A normal cuff of the stomach was available for gastroplasty. We could also palpate the additional lesions in the duodenum and jejunum that were subsequently removed by small enterotomies. Due to rarity, no guidelines are available, but both surgical resection and endoscopic mucosal resection are reported. Lunca et al. described surgical resection as in our case, but due to extensive involvement total gastrectomy and esojunal anastomosis with Roux-en-Y loop was resorted upon. In contrast, Zou et al. were the first to report endoscopic submucosal resection for a giant solitary gastric polyp in PJS.

Thus, PJS can have a very straightforward presentation or may have an uncharacteristic presentation with atypical histology, as described in the index case. Family history and the presence of mucocutaneous pigmentation may help to clinch the diagnosis in the absence of STK11 mutational analysis. The need for an increased index of suspicion for the disease, genetic counseling of family members as well as long-term surveillance for the early detection of malignancies cannot be overemphasized.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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