



A classical case of Peutz–Jeghers syndrome with brief review of literature



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Abstract PJS is an autosomal dominant genetic disease associated with melanin pigment spots on the oral mucosa, lips, nasal alae, palm and soles, as well as hamartomatous polyps in the alimentary canal. Polyps are often a cause of intussusception in the affected patients. Cancers of gastrointestinal system, uterus and breast are common in patients with PJS. Long-term follow-up is required to prevent intussusception in children and cancer in adults. We report a classical case of Peutz–Jeghers syndrome presenting with jejunoileal intussusception in a 9 year old child.

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1. Introduction

Peutz–Jeghers syndrome (PJS) is a rare disorder characterized by typical pigmented perioral macules, pigmented spots in the buccal mucosa which are present in 90% of patients [1,2], and multiple hamartomatous polyps predominantly in the gastrointestinal (GI) tract, rarely more than 20 [3]. Polyps may occasionally be absent. Polyps vary in size from a few millimeters to 7 cm. Most patients have a characteristic clinical course of recurrent episodes of polyp induced bowel obstruction and bleeding. The disease affects males and females equally. In addition to polyposis, the risk of gastrointestinal and extra-gastrointestinal malignancies is significantly increased in PJS patients [4]. The relative risk of

dying from a gastrointestinal cancer is 13 times higher and risk of any other malignancy (especially cancer of the reproductive organs, breast, pancreas and lung) is 9 times higher than in the general population [5].

2. Case report

A 9 year old girl was apparently alright before, presented with slight intermittent episodes of abdominal pain without localization and vomiting. On examination the child was toxic, restless with rapid pulse. Marked tenderness was present in the upper abdomen and a palpable, mobile firm to hard mass of the size of a fist was felt. Abdominal ultrasonography revealed intussusception. An emergency laparotomy was performed. Jejunoileal intussusception showing approximately 15 cm of jejunum invaginating in to the ileum was found (Fig. 1). The intussusception could not be released, hence it was resected and sent for histopathology.

Gross specimen of small intestine measured 15 × 4 cm in length with grayish dull appearing serosa. There was no

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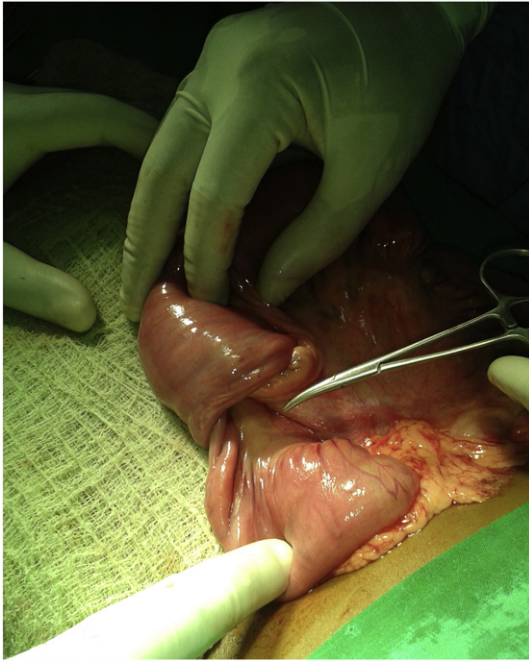


Fig. 1 Intraoperative demonstration of jejunoileal intussusception.

gangrene. On cut opening lumen showed multiple pedunculated polyps ranging from 1 to 3 cm in diameter. Two large pedunculated polyps were found as the leading point for the intussusceptum. The polyps were reddish gray in color, smooth surface and cut section was congested without any focal hemorrhage and necrosis (Fig. 2). Histopathology revealed complex arborizing cores with smooth muscle bundles supporting aggregates of benign cystically dilated glands suggestive of hamartomatous polyp. Atypia or malignancy is not observed in the neoplastic glands (Fig. 3A and B). Lymph nodes examined showed reactive hyperplasia.

Family history did not reveal history of PJS/any other polyposis syndrome in the family members. On careful re-examination of the child multiple pigmented patches were seen over buccal mucosa (Fig. 4). The girl was discharged well

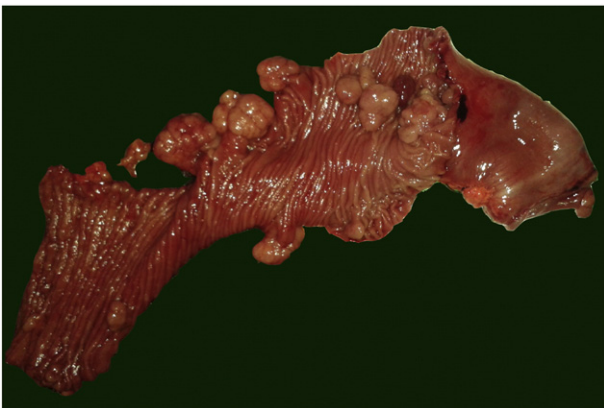


Fig. 2 Two large polyp as the leading point of the intussusceptum.

on the 8th post-operative day with a diagnosis of Peutz-Jeghers syndrome with an advice for regular follow ups. On follow up she was found to be absolutely alright without any complications for last 8 months till the manuscript was prepared.

3. Discussion

Peutz in 1921 published the first case with GI familial polyposis with pigmentations and later in 1949 documented that these are associated with increased risk of malignancy [1,6]. PJS is a rare disease with an incidence of 1 in 30,000 to 120,000 live births [7]. It is an inherited GI hamartomatous polyposis syndrome with mucocutaneous pigmentation. The most distinctive clinical feature are melanin pigmentations (black–brown spots) in the lips and buccal mucosa. Pigmentations can also be seen in other parts of the body, such as fingers, toes, hands, feet and the mucosa of the nose, conjunctiva and rectum. Some patients do not present with the full spectrum of the disease. Multiple hamartomatous polyps in the gastrointestinal tract are the hallmark of PJS. Mostly gastrointestinal polyps are found in the small intestine. They can also be found in the stomach and large intestine [7].

Giardello et al [8] proposed diagnostic criteria for PJS which requires histopathological confirmation of hamartomatous gastrointestinal polyps and two of the following features: small bowel polyposis, positive family history and pigmented skin or mucosal brown macules.

PJS shows an autosomal dominant pattern of inheritance with both familial (80%) and sporadic (25%) transmissions. Two independent groups of investigators identified the mutated gene responsible for PJS [9,10]. The gene was localized to chromosome 19p34-p36 and is known as STK 11, a serine-threonine kinase involved in growth control regulation. Not all patients with PJS have a mutation in this gene. Mutations of chromosomes 6q and 19q have been suggested in a few families [11]. In the present case the diagnosis was confirmed because of the hamartomatous small intestinal polyps and mucocutaneous hyperpigmentations. No family history of PJS was detected in the present case suggesting a sporadic new mutation.

Complications induced by polyps include colicky abdominal pain, bleeding, and bowel obstruction due to intussusception. The time for commencement of abdominal symptoms vary from as early as the first year of life to the age of 40 years [12]. By the age of 10 years, 30% of patients with PJS already require a laparotomy [13].

These patients are prone to many extra-intestinal tumors like testicular sertoli cell tumors, ovarian tumors like sex cord tumors with annular tubules, granulosa theca cell tumors, cystadenomas, breast tumors like carcinoma breast, papilloma with squamous metaplasia, cholangioma, pancreatic adenocarcinoma, adenoma malignum, bronchial carcinoids, papillomas in bladder and pelvis [14].

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