CASE REPORT

PEUTZ-JEGHERS SYNDROME ANAEMIA
AS ONLY PRESENTATION

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Abstract

Peutz-Jeghers syndrome (PJS) is a rare autosomal dominant genetic disease manifesting as mucocutaneous pigmentation and multiple polyps in gastrointestinal tract. We report this case of PJS as it is a rare entity with our patient having a unique presentation of severe anaemia only without any gastrointestinal complaints.

Keywords: Peutz-Jeghers Syndrome, Anaemia, Intestinal Polyposis

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Introduction

Peutz-Jeghers syndrome (PJS) is a rare inherited disease that is characterized by gastrointestinal polyps in association with pigmentation affecting skin and mucous membranes. PJS has autosomal dominant inheritance. Patients with PJS have an increased risk of developing cancers compared with the general population in addition to the various problems which pertain to the intestinal polyposis.

Case Presentation

A twelve-year-old female presented with progressive shortness of breath, paleness of the body and easy fatigability for last three months and swelling of the feet developing over last one week. Her dietary intake was adequate. On examination she was well preserved with normal vitals. She had severe pallor, pedal oedema extending up to the legs and hyper pigmented macules over the face and lips (Figure 1).
There was no lymphadenopathy, icterus, clubbing, cyanosis or superficial and deep bleeds with a normal jugular venous pressure. Cardiovascular system examination had a short systolic murmur with normal heart sounds. On investigations her haemoglobin was 4.1gm/dL. Peripheral blood film showed a microcytic, hypochromic picture with corrected reticulocyte count of 1.1% and no abnormal/immature cells or parasites. Red cell distribution width was 15.8%. The total leukocyte, differential leukocyte, platelet counts and erythrocyte sedimentation rate were within normal limits. Antinuclear antibodies and direct Coombs’ test were negative. Her renal and hepatic function tests, ultrasonographic, doppler and echocardiography studies were normal. Serum iron studies were reported as iron 23ug/dL, transferrin saturation 5%, total iron binding capacity 433ug/dL suggesting iron deficiency. Stool examination was negative for parasites but positive for occult blood. Endoscopy and colonoscopy were done in view of occult blood in stools which showed three polyps in antral region, scalloped folds in the second part of duodenum and a large simple polyp at the hepatic flexure (Figure 2).
Figure 2. Intestinal polyposis

Histopathological examination of the polyps showed polypoidal structure lined by normal colonic mucosa with variably sized gland with smooth muscle bundles having racemose appearance (Figure 3).
Few glands were cystically dilated having intraluminal eosinophilic secretions. Final diagnosis of severe anaemia due to gastrointestinal blood loss as a consequence of intestinal polypsis part of Peutz-Jeghers syndrome was kept. Polypectomy was done and patient was discharged on iron. On follow-up after 3 months her haematocrit increased towards age appropriate levels with serum iron studies being reported as normal. Repeat stool testing showed no gross or occult blood. She is doing well with a normal haematocrit and no blood in stools for last six months. Family screening could not be performed due to the financial constraints of the family.

Discussion

Peutz-Jeghers syndrome named after Augustine’s Peutz and Harold Joseph Jeghers is also known as hereditary intestinal polyposis syndrome. It is an autosomal dominant genetic disease manifesting as benign hamartomatous polyps in the gastrointestinal tract and melanotic macules on the skin, especially on the lips and oral mucosa [1]. Its incidence is approximately 1 in 25,000 to 300,000 births [2].
The main criteria for clinical diagnosis of PJS are:

1. Family history of Peutz-Jeghers syndrome.

2. Mucocutaneous manifestations include appearance of melanocytic macules (pigmented spots). They are tan, dark brown or bluish black flat patches of 1 to 5 mm in size present in 95% of the patients. These lesions usually appear before 5 years of age. They may fade after puberty and are seen over the mouth, lips, gums, inner lining of the mouth, eyes, hands and feet, fingers and toes, anus and genital areas. Localization in the oral mucosa is typical of patients with PJS and does not happen with other types of dermatologic pigmented lesions.

3. Gastrointestinal manifestations which include benign hamartomatous polyps found in the gastrointestinal tract having a 15-fold increased risk of developing intestinal cancer compared with the general population. They manifest as anemia, rectal bleeding, abdominal pain, obstruction, and/or intussusception. A rectal polyp may be found during a rectal examination or may prolapse.

Clinical diagnosis is established when 2 out of the above 3 features are present and 90-100% of these patients have a mutation in the tumour suppressor gene STK11/LKB1 located on chromosome 19 for which molecular genetic testing is available [3]. A cumulative risk for all cancers by age 30 years is 5% and rises up to 85% by the age of 70 years [4].

In investigative workup, complete blood cell (CBC) count, iron studies and faecal occult blood are directed to detect any faecal blood loss and its consequences. Various imaging studies and endoscopic studies have a role in diagnosis and treatment. Multiple polyps and a large mass at hepatic flexure on contrast barium and endoscopic studies are seen in PJS. Histological examination of polyps shows extensive smooth muscle arborization throughout the polyp giving the appearance of pseudoinvasion, because some of the epithelial cells are surrounded by the smooth muscle. Various tumour markers like cancer antigen (CA)-19-9 and CA-125 testing is indicated every year starting at age 18 years, and CA 19-9 is indicated every 1-2 years starting at age 25 years [5].

**Treatment**

Management includes resection of symptomatic and large polyps and surveillance for cancers. Resection of the polyps is required only if serious bleeding or intussusceptions occurs. Various modalities for resection are enterotomy for removing large and single nodules, resection of short lengths of heavily involved intestinal segments and snaring the polyps during colonoscopy if they are within reach.

Pharmacological agents like cyclo-oxygenase (COX) inhibitors, mammalian target of rapamycin (mTOR inhibitors; sirolimus) and RAD001 (everolimus) has been under trial for PJS.

Surveillance guidelines for early detection of cancer are [6]:

- Small bowel radiography, esophagastroduodenoscopy and colonoscopy every 2 years
- Computed tomography or magnetic resonance imaging of the pancreas, ultrasound of the pelvis (women) and testes (men) yearly
- Mammography (women) from age 25 and papanicolaou (Pap) test every year
Conclusion

PJS should be suspected in any patient with mucocutaneous pigmentation and gastrointestinal features. It can rarely present as anaemia without any gastrointestinal symptoms as in our patient. It needs more attention because of the complications related to polyps and its association with the cancers.

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References


