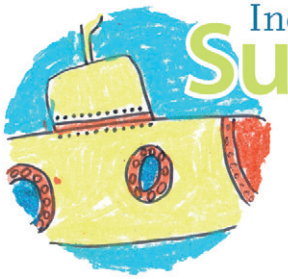

Index of Suspicion

3 Abdominal Pain, Nausea, and Vomiting in a 12-year-old Girl

Izaskun Melania Iglesias, MD, MBA,* Alexander Fernandez,*
Eduardo Smith-Singares, MD, FACS,* Kenneth Soyemi, MD, MPH, MBA,*
Rosibell Arcia, MD*

John H Stroger Hospital of Cook County, Chicago, IL.

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PRESENTATION

A 12-year-old girl presents to the emergency department with a 5-hour history of abdominal pain, nausea, and vomiting. The pain is periumbilical, radiating to the left lower quadrant, colicky, and rated 5/10 at the onset. In the emergency department, she rates the pain as 8/10 and has two nonbloody, nonbilious episodes of emesis. She denies any fever, constipation, or diarrhea. She is not sexually active, had menarche 2 months ago, and had her last menstrual period end 3 days ago. She denies dysmenorrhea. She has no allergies, hospitalizations, or previous surgeries.

On physical examination, the patient is in severe distress, afebrile, and pale. Mucous membranes are moist, and the girl has hyperpigmented perioral and intraoral mucosal lesions, freckles on her face, and macules on the tips of the fingers. Her abdomen is soft, nondistended, and tender to palpation on bilateral lower quadrants with rebound tenderness. No masses are palpable and the Rovsing sign is positive. The Murphy sign, psoas sign, and costovertebral angle tenderness are all negative

Pertinent laboratory finding include: white blood cell count of 22,800/μL (228 $\times$ 10^9/L, (71% neutrophils, 18% lymphocytes, 10% monocytes, 1% eosinophils), hemoglobin of 10.5 g/dL (105 g/L), hematocrit of 31.9% (0.32), mean corpuscular volume of 78.2 μm^3 (78.2 fL), red cell distribution width of 14.1%, and platelet count of 477 $\times$ 10^3/μL (477 $\times$ 10^9/L).

DISCUSSION

The girl was hospitalized for sequential abdominal examinations and pain management. She received two boluses of normal saline, 30 mg ketorolac and 2 mg morphine for pain control, and 4 mg ondansetron for nausea. Pediatric surgery was consulted.

Initial ultrasonography was inconclusive but revealed a dilated tubular blind structure in the right lower quadrant containing echogenic material that was concerning for acute appendicitis. Repeat abdominal ultrasonography performed the following morning showed an edematous loop of bowel with surrounding moderate amount of fluid in the right upper and lower quadrants and pseudokidney appearance suggestive of intussusception. The patient underwent emergent exploratory

laparotomy. Upon entering the peritoneal cavity, surgeons identified a large segment of ischemic bowel. After evisceration, they noted a 360° clockwise torsion of the jejunum around the intussusception, which they de-torsioned counterclockwise. After attempted reduction, they resected the intussuscepted portion of jejunum. A loop of distal jejunum was sufficiently damaged during these maneuvers to warrant resection as well. Both segments of small bowel were sent for pathology, and two small bowel anastomoses were created to restore continuity. Upon palpation of the rest of the small bowel (including the intervening segment), surgeons detected two large intraluminal masses. One was located at the ligament of Treitz and the other within the intervening segment of small bowel. Both were removed through small overlying enterotomies that were subsequently repaired in two layers. Finally, an incidental appendectomy was performed.

The pathology report described the two segments of small bowel. A proximal jejunal section showed hemorrhagic necrotic bowel with infarcted hamartomatous (Peutz Jeghers) polyps, and a distal jejunal segment had transmural ischemic changes and hemorrhage but no polyps. In addition, the two polypectomy specimens were described as hamartomatous polyps.

The Condition

Peutz Jeghers syndrome (PJS) is a rare autosomal dominant genetic disease that is associated with hamartomatous polyps in the gastrointestinal tract and hyperpigmented macules on the lips and oral mucosa, with possible complications of intussusception, obstruction, and malignancy. The association of intussusception and volvulus in a patient with PJS is very rare.

Even though this patient was not diagnosed before the admission, the physical findings of mucocutaneous lesions should have prompted clinicians to obtain more history, including family history. Also, physical examination of family members could potentially have helped in making the diagnosis. Subsequent examination revealed that the girl's mother also had mucocutaneous lesions.

Once a patient is suspected of having PJS, further genetic evaluation should be undertaken. Genetic testing is not definitive and mutations of serine/threonine kinase 11 (*STK11*) tumor suppressor gene can only be detected in 50% to 60% of patients. Gene mutations affect the function of the protein STK11, resulting in uncontrolled cell growth and the formation of noncancerous polyps and cancerous tumors in patients with PJS. A geneticist confirmed the diagnosis of PJS in this patient, but the family refused genetic testing.

Hamartomatous polyps in PJS are precancerous. They show low-grade dysplasia that eventually leads to a high-grade dysplasia and finally carcinoma. The polyps are usually found in the small intestine, and current guidelines are to remove polyps larger than 1.0 to 1.5 cm in adults with PJS. In children, the management depends on disease severity. To prevent serious complications, screening of PJS patients with upper and lower endoscopy has been recommended as early as age 10 years.

The Complication

When patients who have PJS present with abdominal pain, intussusception should be considered in the differential diagnosis. Intussusception in healthy children is usually diagnosed between ages 2 months and 2 years. Older patients should always be suspected to have a cause (leading point) for the intussusception. The proportion of intussusceptions due to an underlying cause increases with age. Typically a lesion can be identified as the lead point in patients older than age 5 years. Some of the associated lesions are Meckel diverticulum, lymphoma, and hamartomatous polyp due to PJS. Other diseases associated with intussusception are Henoch-Schönlein purpura and cystic fibrosis.

Lessons for the Clinician

- Clinicians should have a high index of suspicion for intussusception when a patient who has Peutz Jeghers syndrome (PJS) presents with abdominal pain.
- Patients who have PJS should be educated on the associated complications of polyps in the gastrointestinal tract.

Suggested Readings for this article are at http://pedsinreview.aappublication.org/content/37/2/83.

ANSWER KEY FOR FEBRUARY 2016 PEDIATRICS IN REVIEW:

Trends in the Diagnosis and Management of Pediatric Appendicitis: 1. B; 2. B; 3. A; 4. C; 5. E.

Protozoan Parasites: 1. B; 2. C; 3. C; 4. C; 5. A.

Knee Pain, Part II: Limb- and Life-threatening Conditions, Hip Pathology, and Effusion: 1. D; 2. A; 3. A; 4. A; 5. B.