A case of small bowel obstruction secondary to intussusception caused by Peutz-Jeghers syndrome

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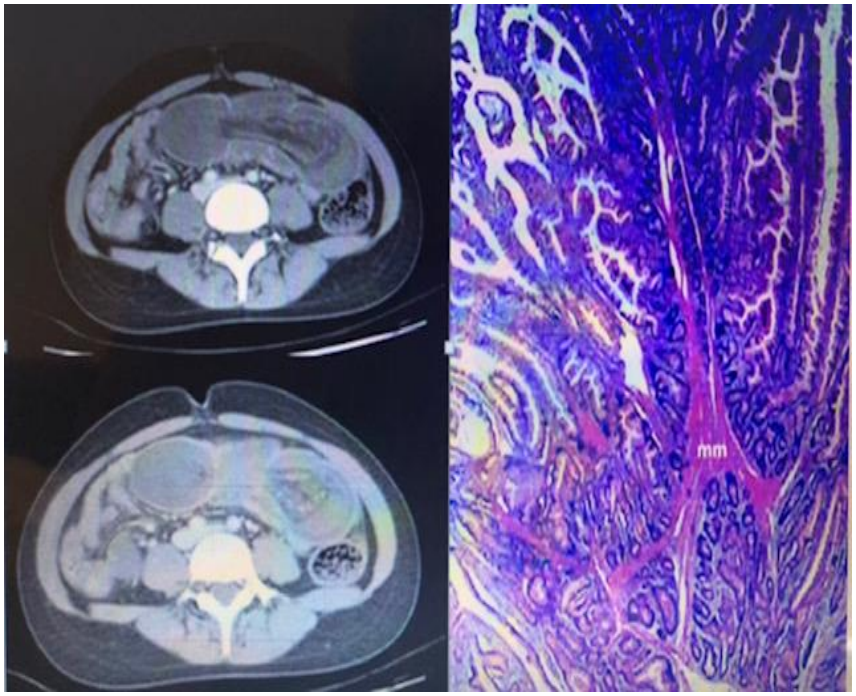

- Case: Patient JK, a 21 y/o Asian female presented to the emergency department with abdominal pain, nausea and vomiting. She states she awoke with excruciating abdominal pain which she describes as 10/10. JK states she has had similar episodes over the past few months but none that have lasted more than ½ hour. Patient JK was currently diagnosed by her PMD with microcytic anemia and prescribed iron supplements. JK started taking the supplements two days prior to this episode.
- On arrival she was alert and oriented x 3 but complained of 10/10 abdominal pain, nausea and vomiting. Patient JK describes the abdominal pain as diffuse in nature and constant. She states that the pain is mostly located in the upper quadrants. JK states that nothing alleviates it and that any movement will exacerbate the pain. JK has not been able to keep anything down and has been actively vomiting since awaking this morning. She was maintaining her airway and breathing spontaneously with a respiratory rate of 20 breaths/minute and no evidence of trauma. She had a heart rate of 65 beats/minute with a blood pressure of 144/77 mm Hg. She was afebrile. JK had pale conjunctiva along with dry mucus membranes. Abdominal exam revealed tenderness to palpation in both the left and right upper quadrants. Exam was unremarkable for any rebound, rigidity or abdominal masses. Neurological exam revealed pupils equal, round, reactive to light and accommodation at 5mm bilaterally. Extraocular muscles were intact along with cranial nerves II-XII. Strength, sensation, reflexes and coordination were equal bilaterally with no deficits noted. An electrocardiogram showed normal sinus rhythm with normal PR, QRS and QTc intervals. Initial finger stick was 133 mg/dl.
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- JK had an IV placed and was given aggressive pain management along with antiemetic medications. JK began drinking contrast for a CT scan of her abdomen and pelvis. Initial blood work showed a hemoglobin/hematocrit of 5.0/17.4 with an MCV of 60.3, JK also had a potassium level of 2.9. CT abdomen/pelvis revealed small bowel obstruction secondary to intussusception (Figure 1). Emergent surgical consult was obtained. Vital signs remained stable along with remaining blood work throughout the emergency department stay, but JK was transferred to our acute ER due to her low hemoglobin/hematocrit and CT findings. Transfusion with packed RBC's were initiated as JK was being prepared for surgery. In the operating room, an exploratory laparotomy was performed. Small bowel obstruction secondary to a tumor was found, and multiple small bowel tumors were observed. Pathology was consistent with a hamartoma and the diagnosis of PJS was initially made.
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- Discussion: Peutz-Jeghers syndrome (PJS) is an autosomal dominant inherited disorder characterized by intestinal hamartomatous polyps associated with mucocutaneous melanocytic macules[1]. Although the intestinal lesions are hamartomas, patients with this rare genetic disorder have an increased risk of developing intestinal cancer by 15 fold compared to general population[2]. This rare disorder occurs rarely with a frequency of 1 case per 300,000 people[1]. Peutz-Jeghers syndrome seems to be caused by a germline mutation of the *STK11* gene, located on band 19p13.3[1]. Phosphorylation by cAMP-dependent protein kinase A appears to regulate this protein[3].
- Morbidity in PJS primarily stems from the location of the polyps in the gastrointestinal system, such as stomach, small intestine, or colon[1]. Most common clinical presentation of this syndrome is small intestinal obstruction and intussusception (43%). Additionally, abdominal pain (23%), hematochezia (14%), and colonic polyp prolapse (7%) are possible presentation, and these typically occur in their 10's and 20's[4]. Almost half of patients with PJS develop cancer and die from it in their late 50's[5].
- In more than 95% of cases, patients with PJS presents with melanin spots and mucocutaneous pigmentation[6]. They appear as flat and small spots with brown or dark blue colors with an appearance of freckles, most commonly in the peribuccal area[6]. Cutaneous pigmentation (1- to 5-mm macules) is usually located in the perioral region, crossing the vermilion border (94%), in the perinasal and perioral areas[3]. These pigmented areas may fade after puberty and are not needed for diagnosis[3].