Gorlin Syndrome and Presentation with Intraabdominal Cysts

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Background

Gorlin-Goltz Syndrome (Focal Dermal Hypoplasia) is an autosomal dominant hereditary disorder with variable penetrance. It is characterized by cutaneous abnormalities as well as dental, osseus, ocular, and neurologic anomalies. The most concerning manifestation seen with this syndrome is basal cell carcinoma.

Etiology of Gorlin Syndrome is thought to be secondary to mutation in the PORCN gene, contributing to skin and bone development.

Very few cases of intraabdominal cysts have been described.

Case Presentation and Workup

Patient is a 41 year old female with history of Gorlin syndrome with prior basal cell nevi, odontogenic cysts, polycystic ovarian syndrome, and hypertension. Surgical history is pertinent for recent sleeve gastrectomy.

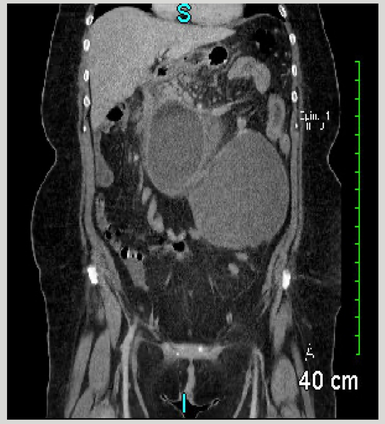
- Presented with chief complaint of epigastric abdominal pain with CT imaging demonstrating multiple large cysts adjacent to the pancreas, as well as a walled-off cystic mass within the rectus musculature
- Worsening anorexia and persistent abdominal pain prompted surgical intervention

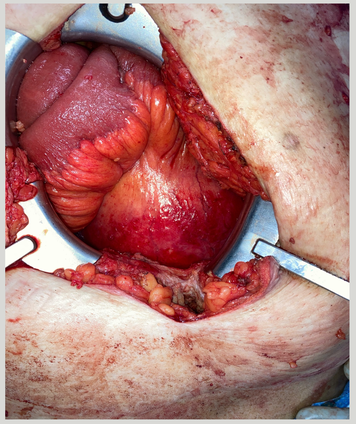
Management and Surgical Intervention

- Patient was taken for a diagnostic laparoscopy, converted to exploratory laparotomy
- Left lower quadrant cystic abdominal wall lesion contained palpable fluid collection, this was unroofed and drained – yielded large volume necrotic material
- Two large mesenteric cystic masses were identified adjacent to the pancreas with a third cystic mass located in the left lower quadrant – each sequentially aspirated and sent for fluid analysis and culture
- Portions of the wall of the cystic masses were excised and sent to pathology, working diagnosis of walled-off pancreatic necrosis at the conclusion of the procedure

Operative pathology: Benign fibroadipose tissue with chronic inflammatory infiltrate with fat necrosis for each of the intraabdominal and abdominal wall cysts

- Fluid cultures negative, *fluid analysis pertinent for elevated triglycerides* – suggestive of possible lymphomesenteric cysts
- Discharged on POD2, returned in clinic on interval follow up with improved tolerance of PO intake and repeat CT imaging demonstrating improvement in size of peripancreatic fluid collections





Left image: CT imaging demonstrating multiple central abdominal cystic lesions, well encapsulated

Right image: Intraoperative photograph with cystic mesenteric-based lesion prior to aspiration and unroofing

Discussion

Gorlin Syndrome is a hereditary disorder with variable penetrance and varying cutaneous anomalies, including most notably basal cell carcinoma.

Intraabdominal cystic lesions are uncommon upon literature review. Chylous mesenteric cysts are rare. There is only one documented case of symptomatic pancreatic pseudocysts in this patient population.

Our suspicion is given the variation of presentation of patients with Gorlin syndrome and its association with cystic pathology, that this patient had a clinically significant variable manifestation of her disease process that included multiple mesenteric cysts.

References

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