DIAGNOSIS OF MALIGNANT GASTROINTESTINAL NEUROECTODERMAL TUMOR (GNET). WHAT IS IT?

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Background

- Malignant Gastrointestinal Neuroectodermal Tumor (GNET) is an extremely rare primary mesenchymal tumor in the gastrointestinal tract that remains largely unknown due to its rarity and misdiagnosis
- Previously known as Clear Cell Sarcoma-like Tumor of the GI Tract (CCSLTGT), it was only recently termed GNET, a separate entity, in 2012
- Only 111 cases were reported in the literature as of December 2021, making diagnosis and treatment plans extremely challenging
- Literature describes the tumor as aggressive and associated with a poor prognosis

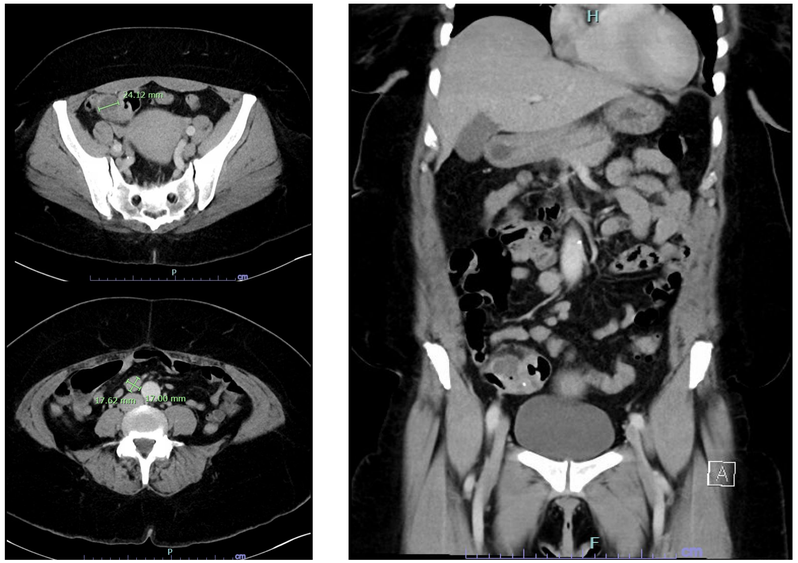


Image 1. CT images of severe irregular thickening in a loop of small bowel in RLQ and enlarged 1.7x1.8cm enlarged mesenteric lymph node

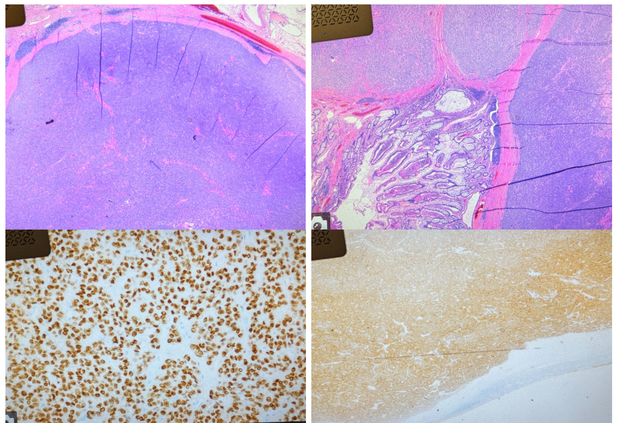
Case

- 56-year-old African American woman with a history of hypertension and uterine fibroids presented with a one-year history of diffuse abdominal pain that acutely worsened, accompanied by nausea and vomiting
- Physical exam demonstrated diffuse abdominal tenderness, most pronounced in the right abdomen. She was able to tolerate liquids but not solid foods due to postprandial cramping
- CT scan of the abdomen revealed severe, irregular thickening of the small bowel in the right lower quadrant, a 2.4-cm fluid collection adjacent to the bowel, and a 1.7 × 1.8-cm right mesenteric lymph node. These findings were new compared with a CT scan performed one year earlier
- Underwent diagnostic laparoscopy, during which the involved segment of small bowel was resected with primary anastomosis, and an excisional biopsy of the mesenteric lymph node was performed
- Pathologic evaluation demonstrated epithelioid and spindle cell transformation of the tumor. Immunohistochemical staining was positive for S-100 and SOX10. Fluorescence in situ hybridization (FISH) revealed an EWSR1 gene rearrangement, confirming the diagnosis of GNET
- Uncomplicated postoperative course and was discharged on postoperative day 4 with full return of bowel function, tolerance of a general diet, and adequate pain control

Image 2. (Top Left) Mesenteric Lymph Node replaced by tumor with strong epithelioid expression
(Top Right) Small Bowel mucosa with tumor invading in the wall
(Bottom Left) Strong Positive SOX100 Expression
(Bottom Right) Diffuse S100 Expression

Conclusion

- Diagnosis and management of GNET is clinically challenging. In this case, time to diagnosis including complete pathologic and genetic evaluation was 16 days
- Tumor was initially misdiagnosed as metastatic melanoma, with the diagnosis later corrected after FISH analysis confirmed EWSR1 rearrangement
- Due to the absence of established clinical guidelines for GNET, the recommendations offered at discharge included a referral to medical oncology and gastroenterology
- This case underscores three clinical and diagnostic highlights regarding GNET tumors
1. The aggressive nature of GNET, demonstrated by rapid progression within one year
2. the extensive and time-consuming workup required to achieve an accurate diagnosis
3. the lack of standardized strategies to guide clinicians managing patients with this rare malignancy.



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