NYU Long Island School of Medicine

DEPARTMENT OF PATHOLOGY

Short Report in Pathology

Organ system: Gastrointestinal Tract

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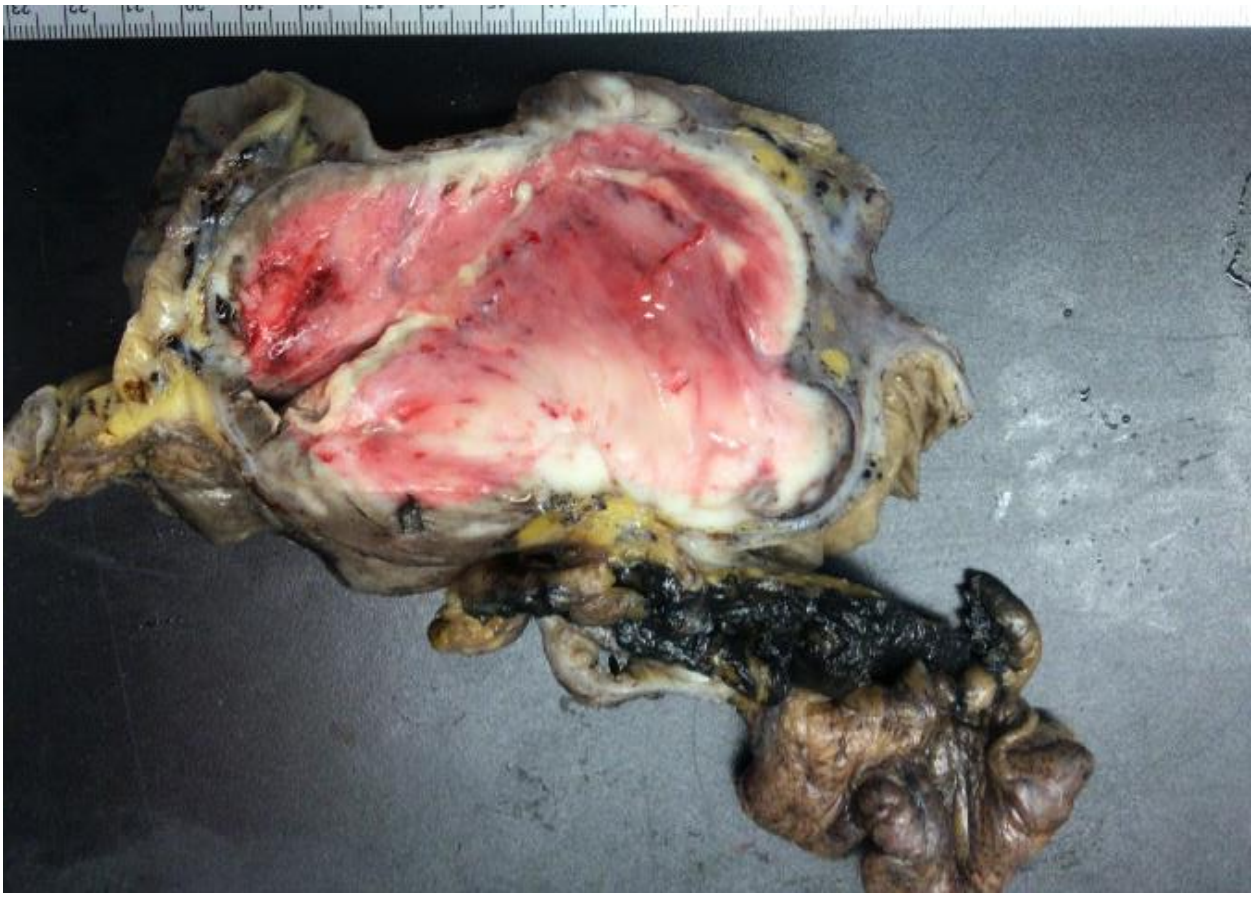
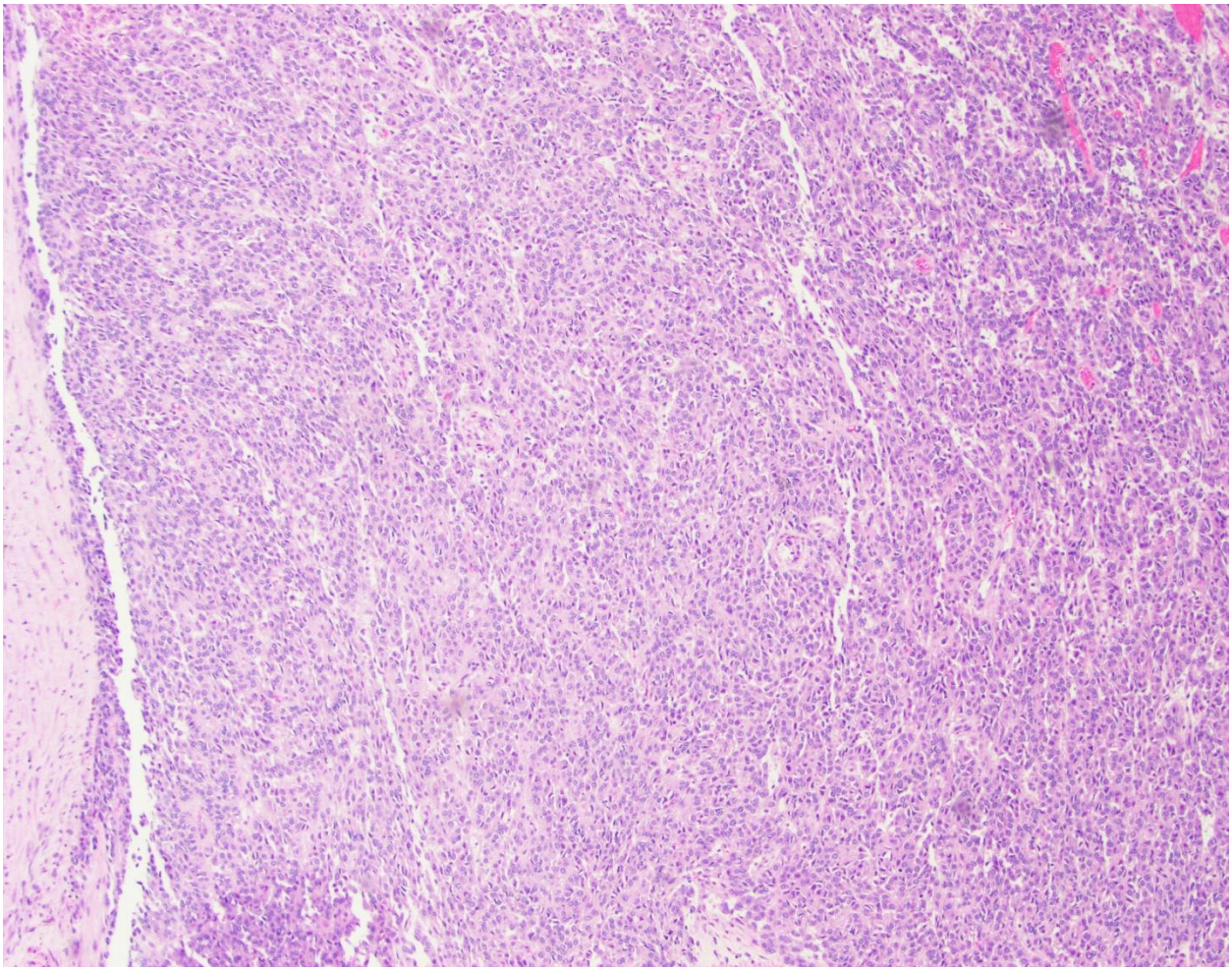
HISTORY

29-year-old male with no significant past medical history presented with cough and abdominal cramping/swelling with fever. He also had associated drenching night sweats, soaking through his sheets and changes of bowel habit. During this period, he also lost 30lb of weight, prompting him to seek medical attention. His lab values showed high ESR, leukocytosis and thrombocytopenia. His HIV test came back as negative. His hemoglobin dropped to 5.2 prompting referral to the ED. CT of the abdomen revealed large abdominal mass (8.4 cm) in the central abdomen.

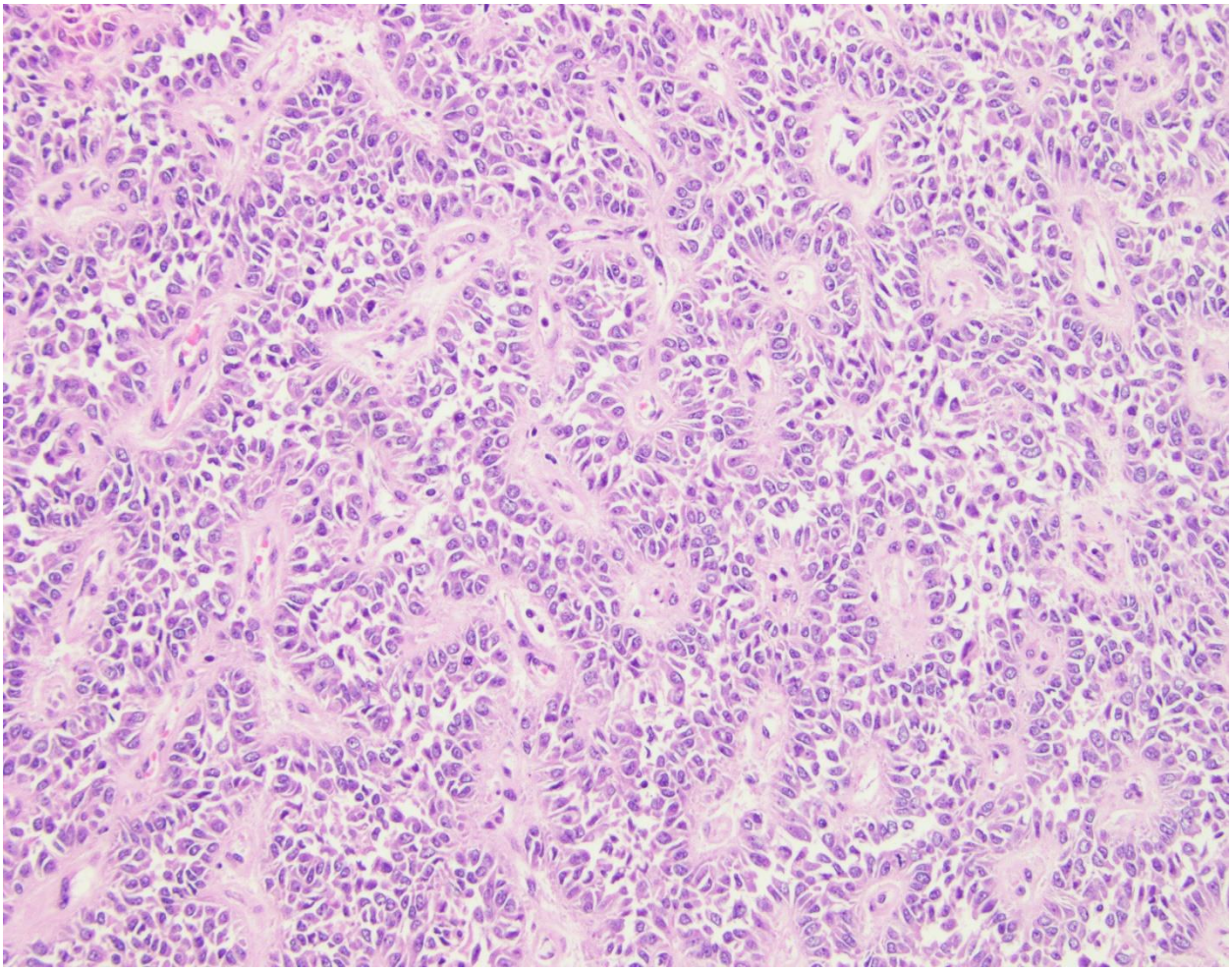
Gross and microscopic Images



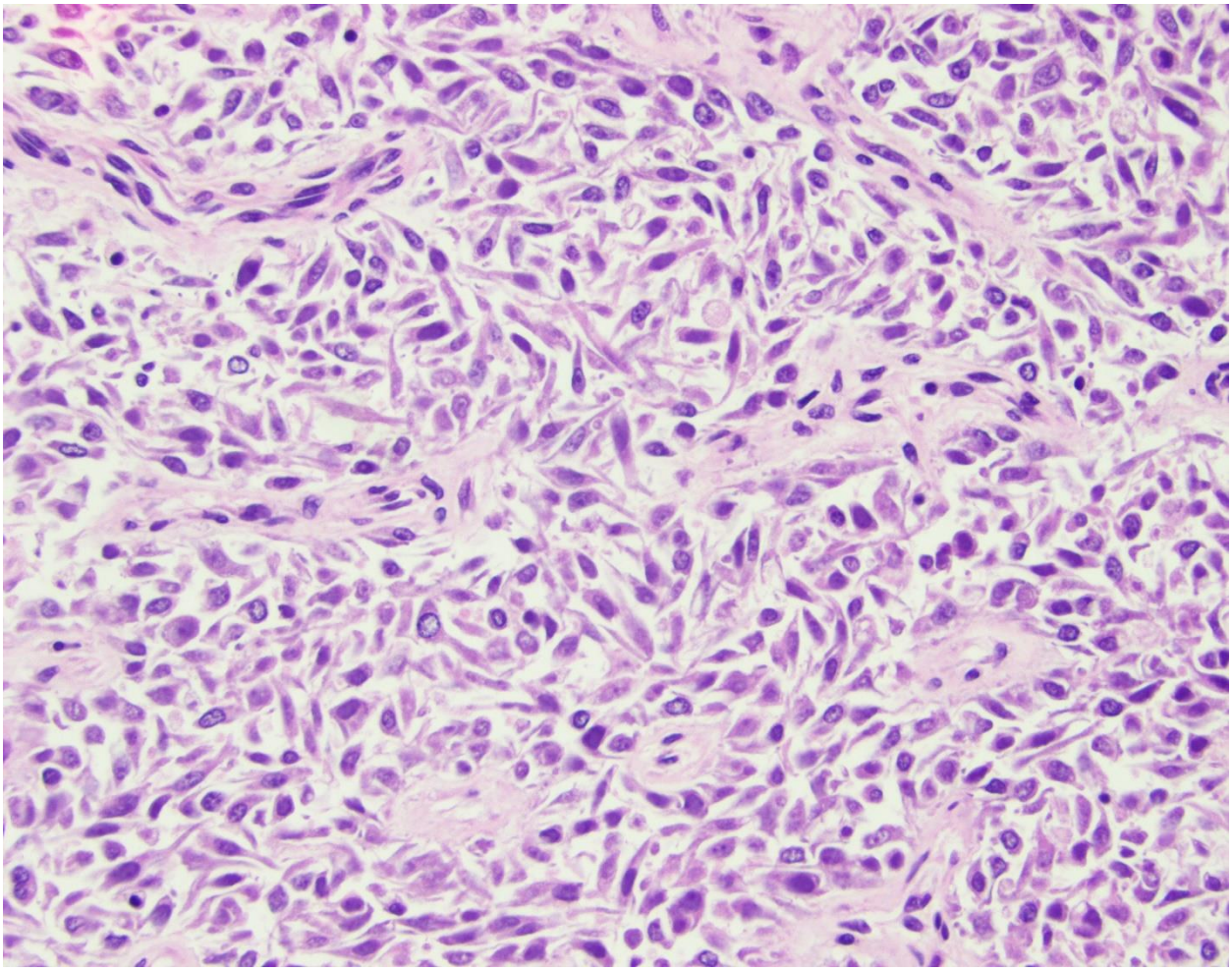
Macroscopic image: 8.4 cm mass with a solid tan fleshy cut surface. The tumor is centered within the wall of the bowel and with secondary involvement of mucosa and submucosa



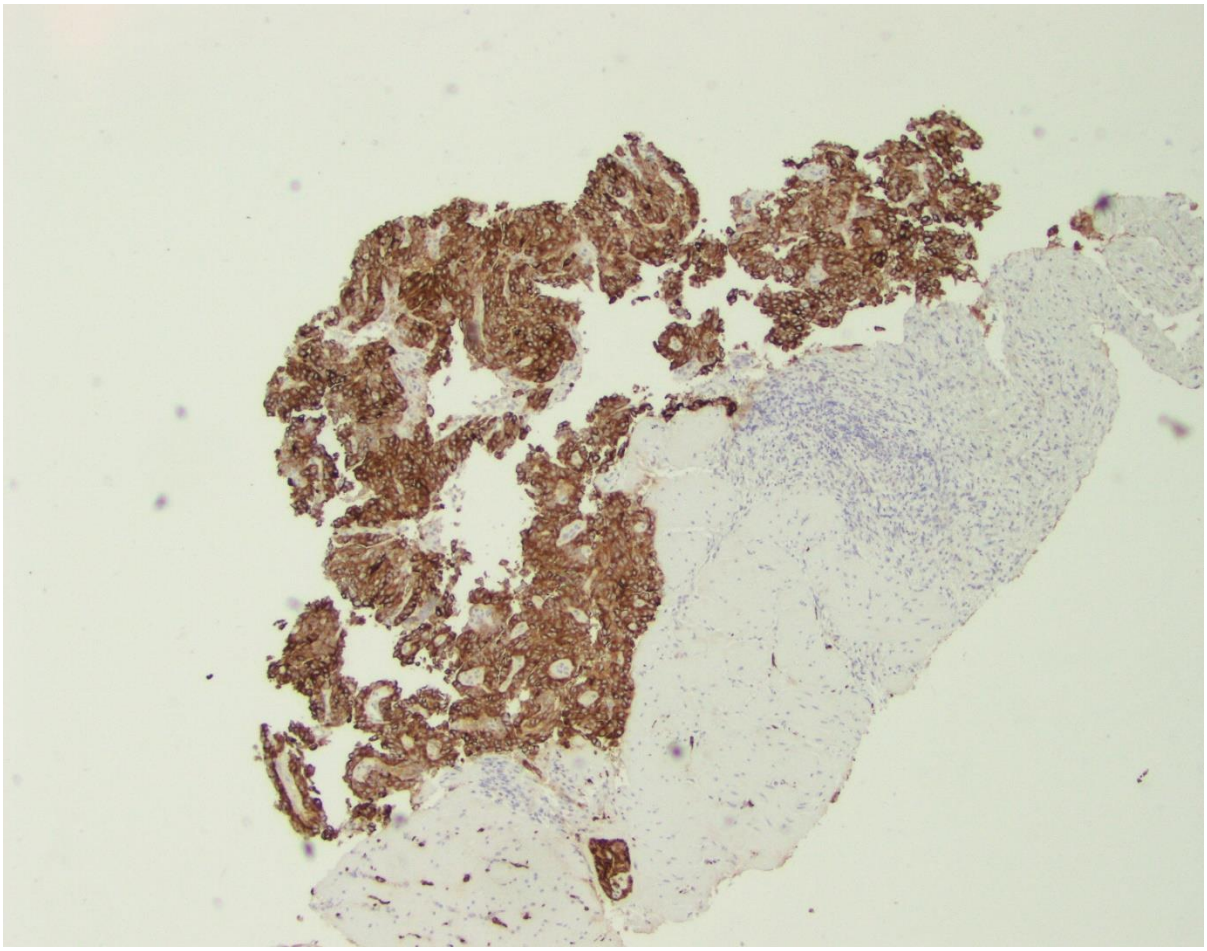
Tumor cells are round to oval primitive looking, relatively uniform (H&E, 100X)



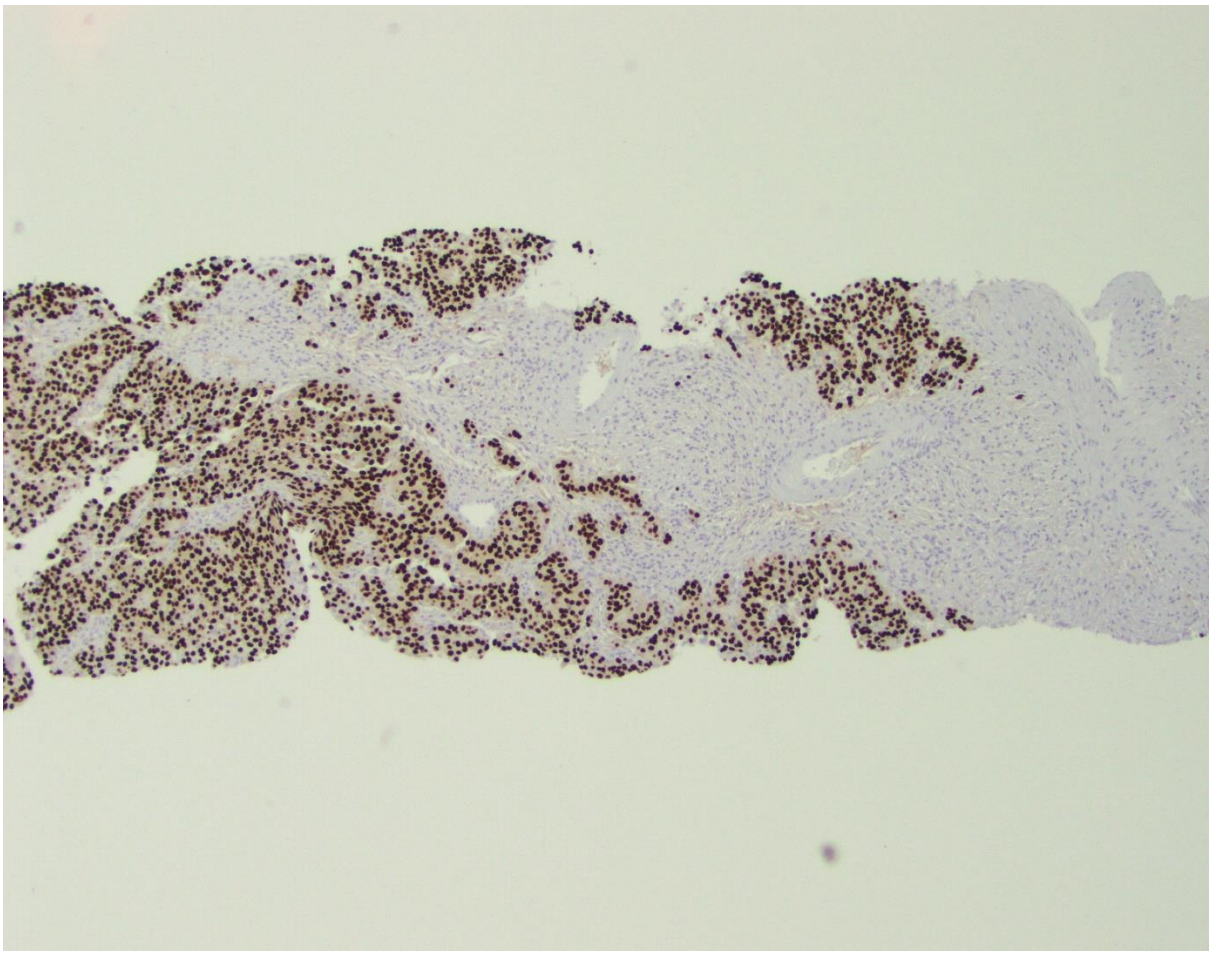
Some areas show pseudopapillary pattern (H&E, 200X)



Some tumor cells show spindling features (H&E, 400X)



The tumor cells are diffusely positive for synaptophysin (100X)



The tumor cells are diffusely positive for SOX-10 (100X)

Immunohistochemistry

Positive stains: AE1/AE3, synaptophysin, SOX-10

Negative stains: Cam5.2, chromogranin, CK7, CK20, CDX2, p40, DOG1, CD117, Melanoma cocktail, Sall4

Diagnosis

GI Neuroectodermal tumor (GNET)

Differential diagnoses

- GIST
- Neuroendocrine tumor

- Metastatic melanoma
- Monophasic synovial sarcoma
- Desmoplastic Small Round Cell Tumor (DSRCT)
- Clear cell sarcoma involving gastrointestinal tract
- Malignant gastrointestinal neuroectodermal tumor (GNET)

Discussion

Malignant gastrointestinal neuroectodermal tumor (GNET) is an extremely rare disease originating from neural crest cells in the digestive tract. It was previously known as clear cell sarcoma-like tumor of the GI tract (CCSLTGT). It primarily affects adults, with the age at diagnosis typically ranging from the second to the seventh decade of life. It can affect any parts of the GI tracts, but most commonly it affects the small intestine. Clinical manifestations vary widely and are often nonspecific, encompassing symptoms such as abdominal pain, weight loss, fatigue, and gastrointestinal bleeding. Radiologically, these tumors present as intraluminal masses with associated features such as bowel wall thickening, ulceration, and mesenteric lymphadenopathy.

The tumor is centered within the wall of the bowel, with secondary involvement of mucosa and submucosa. It can be transmural often ulcerating mass, or it can be exophytic/polypoid or circumferential firm, solid tan to gray-brown cut surface and may show hemorrhage or necrosis. Histologically, GNETs exhibit a diverse range of patterns, including solid, nested, pseudopapillary, microcystic, trabecular, and glandular formations. One of the most distinctive features of GNET is the presence of osteoblast-like multinucleated giant cells admixed with the neoplastic cells. Tumor exhibits round to oval primitive looking cells with infrequent spindling, moderate amount of eosinophilic cytoplasm, variable clear cell morphology, uniform vesicular nuclei with small nucleoli and sometimes nuclear pseudoinclusions may be seen.

Immunohistochemistry plays a critical role in establishing the diagnosis, with GNETs typically showing positivity for markers such as S100, neuron-specific enolase (NSE), synaptophysin, and CD56. The expression of these neuroendocrine markers supports the neural crest origin of GNETs and helps distinguish them from other gastrointestinal tumors.

GNET carries a poor prognosis, and systemic therapeutic options are very limited with lack of guidelines or management recommendation. Although its efficacy in GNETs is unknown, adjuvant chemotherapy is frequently used to lower the risk of local recurrence or distant metastasis. The tumor has high rates of local recurrence and metastases (to nodes and liver), high mortality and metastasis frequent at initial presentation.

References

1. Sing D, Atieh MK, Russell MA, Kittaneh M. Malignant Gastrointestinal Neuroectodermal Tumor (GENT) with Prolonged Disease-Free Survival after Platinum-Based Chemothrapy. Case Reports in Oncological Medicine. Vol, 2020.

2.Saeed S, Grezenko H, Nisar L, et al. (July 07, 2023) A Rare but Aggressive Malignancy: A Case Report of a Gastrointestinal Neuroectodermal Tumor (GNET). Cureus 15(7): e41509. doi:10.7759/cureus.41509