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Unusual Presentation of Neuroendocrine Tumor with Multiple Endocrine Neoplasia 1 Syndrome: A Rare Case Report

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ABSTRACT

Neuroendocrine tumor with multiple endocrine neoplasia type 1 (MEN1) syndrome is detected in a middle-aged lady presented with history of fall leading to multiple fractures. On per rectal examination a polypoidal structure was found. Further investigations confirmed the diagnosis of neuroendocrine tumor.

Key words – Rectal polyp, Rectal neoplasm, Neuroendocrine Tumor, MEN1

INTRODUCTION

Neuroendocrine tumor shows neuro and endocrine properties**.** [1] Neuro properties is based on identification of Dense Core Granules (DCG) and endocrine properties means secretion and synthesis of monoamines. [1]

These tumors begin in neuroendocrine cells. They occur in the digestive tract, lungs and brain. In the digestive tract, they may involve the stomach, pancreas, small intestine, colon and rectum. [1]

The most common site for NET is gastrointestinal region accounting for 60 - 67% of tumor**.** [2] Among all the colon and rectum tumors, rectal NET accounts for approximately 1-2%. [4] About 50 % of rectal NET are asymptomatic and are diagnosed incidentally or presents as polyp in colonoscopies**.** [4,6] Current incidence is approximately 2 per lakh population with a female preponderance. [2]

MEN1 (multiple endocrine neoplasia) is an autosomal dominant inherited tumor of endocrine gland also known as Wermer Syndrome. It is due to loss of menin gene on chromosome 11q13. Menin is a tumor suppressor gene associated with cellular growth and function**.** [3]
MEN1 causes neoplastic proliferation in pituitary, parathyroid and pancreas.

Presenting here is an accidently diagnosed case of rectal polyp (NET) in MEN1 syndrome which presented to our hospital with history of fall leading to multiple non healing fractures.

Clinical History-

A 38 years old female was admitted to our hospital with history of sudden fall leading to non-healing multiple fractures. Per rectal examination revealed a small rectal polypoidal growth.
Hematological and the biochemical tests were within normal range with mild anemia (9 gm/dl) and slightly raised serum calcium levels (12.8 mmol/L).

Colonoscopy was performed which revealed a 3.5 mm rectal nodule 3-4 cm away from anal verge.

CT scan was done in view of non-healing fracture that revealed Parathyroid Adenoma, Hyperparathyroidism and Pancreatitis.

Patient was admitted in view of multiple fractures, but these accidental findings led to the diagnosis of

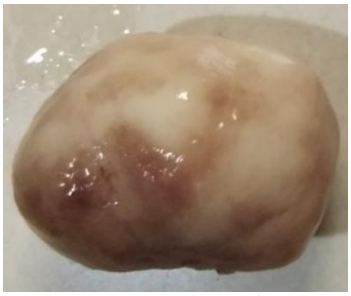
neuroendocrine tumor with MEN1 syndrome. Hence, patient was subjected to surgical excision of the polyp and the tissue was sent for histopathological examination.

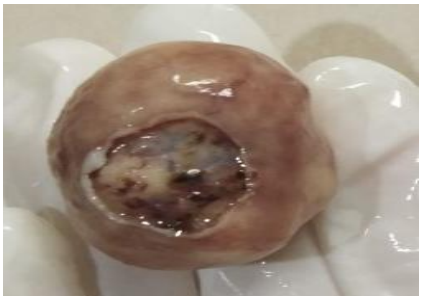
Pathological Findings-

Gross Examination - Received a single, soft to firm, grey-white, globular tumor measuring 3.5 cm in diameter.

External surface was smooth with one ulcerated area measuring 2cmX1.5 cm.

Cut surface: homogenous, grey-white area with yellowish tinge.





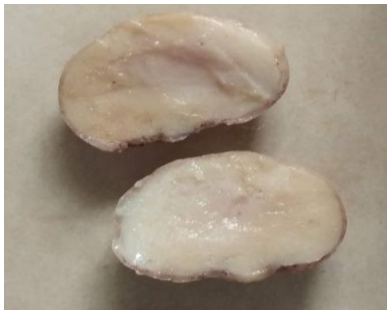


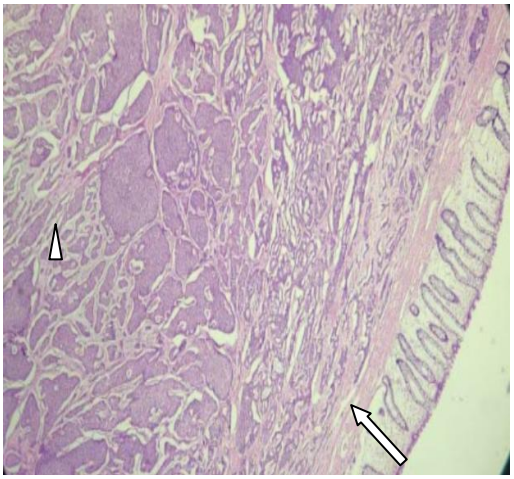
Figure 1: Gross appearance of rectal polypoidal mass.
Figure 2: Gross appearance of ulcerated surface of rectal mass
Figure 3: Cut surface of rectal mass- Tan Homogenous areas

Microscopic examination-

Entirely processed lesion showed lining mucosa and crypts lined by mucus secreting columnar epithelium. The lamina propria and muscularis mucosa were unremarkable.

There was diffusely infiltrating neoplasm in the submucosa and muscularis propria composed of uniform cells arranged in solid pattern (organoid), islands (insular), pseudo glandular, festooning, trabeculae, syncytial and ribbons.

MICROSCOPY-



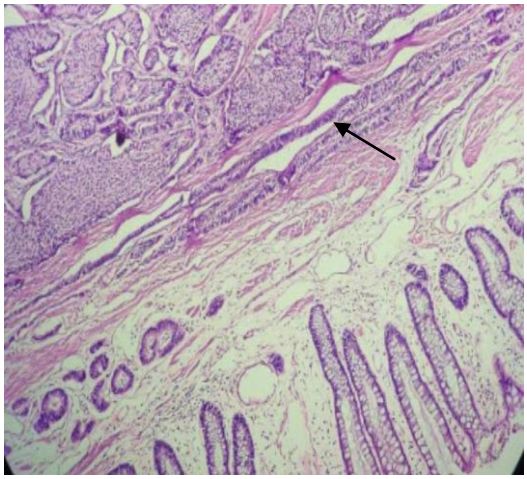


Figure 4- Shows epithelial lining (arrow) & Tumor Islands (arrow head).
Figure 5- Shows submucosal tumor.

The individual tumor cells were uniform, having moderate cytoplasm and indistinct cell borders. The nuclei were round to oval with powdery salt and pepper chromatin. Mitotic activity of 3/10 HPF was noted. However, no necrosis or dysplasia of the overlying epithelium was identified. The base of the polyp was involved by the tumor. No lymphovascular or perineural invasion was seen. Hence, it was diagnosed as Well Differentiated Neuroendocrine Tumor - Grade 1, Rectum- Base of polyp involved by tumor. No lymphatic, vascular or perineural invasion seen.

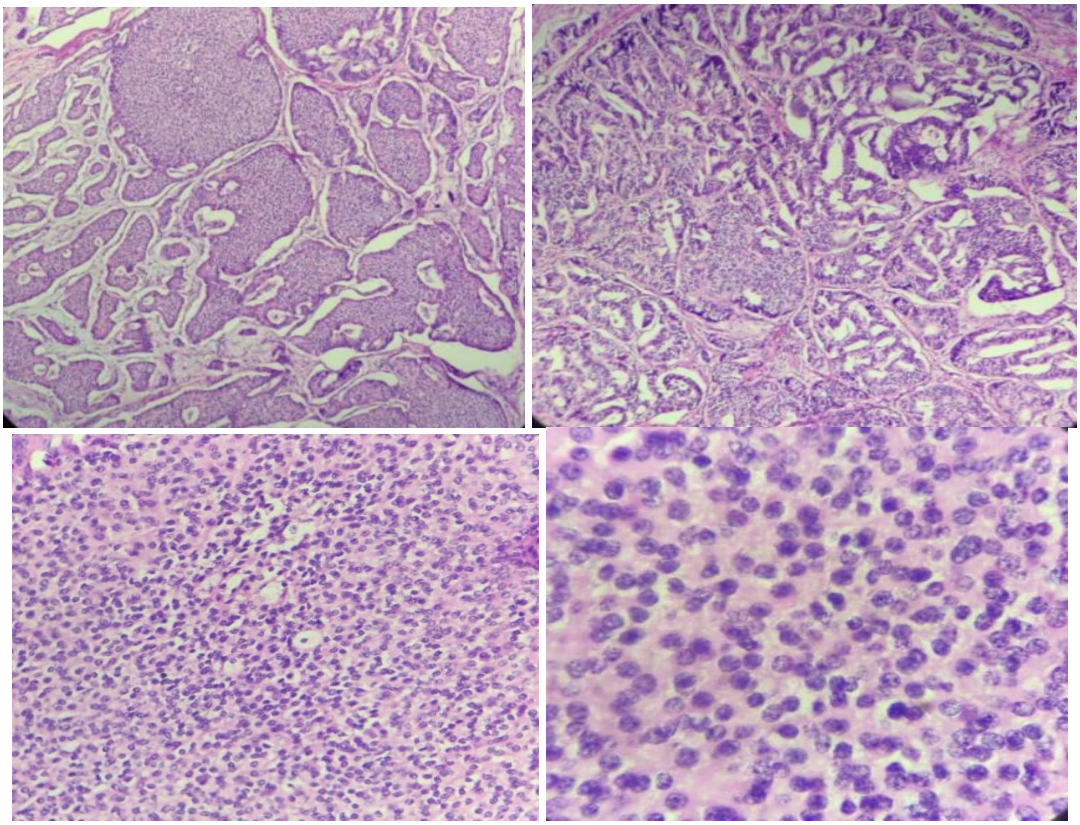


Figure 6 - Tumor cells arranged in solid, organoid pattern(4x)
Figure 7- Tumor cells arranged in pseudo glandular pattern(10x)
Figure 8 & 9 - (40x or 100x) Uniform Tumor cells and nuclei show salt and pepper chromatin.

Immunohistochemistry-

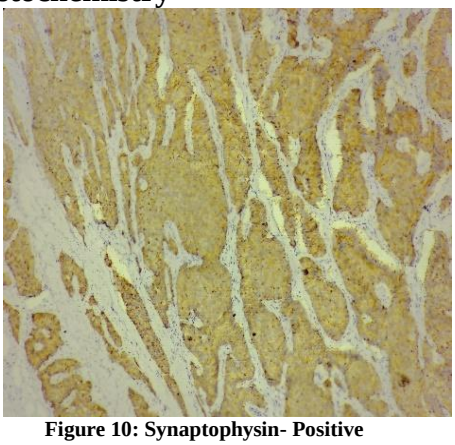


Figure 10: Synaptophysin- Positive

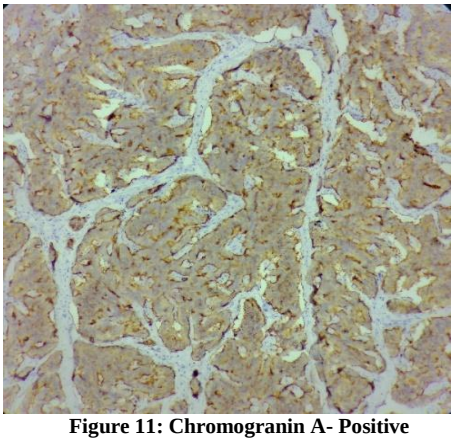


Figure 11: Chromogranin A- Positive

DISCUSSION

NET can occur anywhere in body but most commonly occurs in digestive system. They can be benign or malignant. NET is neuronal and endocrine hormonal tumors. The five years survival rate for localized disease is 87% with an excellent prognosis. [7] Most of the benign cases are asymptomatic, small, slow growing tumor. Generally, rectal NET appears as a single, smooth submucosal nodule or polyp with normal appearing mucosa. [6] Neuroendocrine tumors are derived from enterochromaffin cells which are located throughout the intestine within the crypts of Lieberkühn. The NET typically occurs deep within these crypts. As these tumors progress, they invade the muscularis mucosa and sub mucosa resulting in there subepithelial appearance.

MEN1 is clinically diagnosed by the presence of two or more associated tumors.

It is called familial if a patient is associated with one tumor and a first degree relative with MEN1. [5] It is diagnosed as genetic if a patient has MEN1 gene mutation but no clinical or biochemical MEN1 mutation (a mutant gene carrier). [1]

30% of the pediatric NET is genetically inherited and are associated with MEN1 syndrome, von Hippel Lindau syndrome, Neurofibromatosis type 1 and others. [8] The NET of rectum is rare but are being identified due to widespread use of screening with colonoscopy. They show indolent behavior, but they can be malignant and can metastasize. Lesions showing similar microscopic features include - Small cell carcinoma lung, Lymphoma, Melanoma, Carcinoid tumor, Insulinoma, Gastrinoma, Renal cell carcinoma, Medullary thyroid carcinoma, Pheochromocytoma, PECOMA. Most of the NETs show positivity for Chromogranin A and Synaptophysin.

The current treatment for rectal NET of low grade that is confined within the submucosa is complete resection. Other factors to take into account are poor differentiation of tumor, invasion to underlying muscles, high mitotic figures, high Ki-67 index along with lymphatic/ vascular or neural infiltration. Distance of polyp from anal verge should also be considered while resection.

CONCLUSION

We report a case of unusual presentation of NET as rectal polyp. Although rectal neuroendocrine tumors are rare, still it should be kept in mind in the differentials for rectal polyp. Despite their indolent behavior, these tumors can be malignant and can metastasize. These tumors can occur many years after resection, hence follow up is recommended.

Conflict of Interest: None

Abbreviation- MEN1 (Multiple Endocrine Neoplasia 1), NET (Neuroendocrine Tumor), DCG (Dense Core Granules)

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