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Resumo	Plexiform neurofibromas (PN) represent an uncommon phenotype of neurofibromatosis type 1 (NF-1) in which neurofibromas arise from multiple nerves as bulging and deforming masses involving also connective tissue and skin. These neoplasms rarely affect the colon and rectum. Co-occurrence of PN and neuroendocrine tumors (NET) with primary involvement of the rectum, and medullary thyroid cancer (MTC) in patients with NF-1 is a previous undescribed condition. In this context, we presented a case of primary a PN associate with NET of the upper rectum, in a patient with NF-1, previous submitted to a thyroidectomy due an MTC whose genetic sequencing found a novel pathogenic variant in the neurofibromin 1 (*NF-1*; deletion of exons 2-30) gene. **Case Report:** A 49-year-old woman, with familial history of NF-1 complaining modification of her bowel habits, abdominal cramps, and tenesmus, for six months. She was undergone a thyroidectomy due to MTC 8 years ago. She was submitted to a colonoscopy, which identified a raised submucous lesion, with 5 cm in diameter, located in the upper rectum. During colonoscopy, a deep biopsy of the lesion was performed and the histopathological evaluation and immunohistochemically panel confirm the PN diagnosis. The patient was referred to a laparoscopic anterior retosigmoidectomy. Conventional histopathological examination using the hematoxylin-eosin technique suggested presence of two neoplasm tissues. An immunohistochemical panel was done for etiological confirmation, which

	demonstrated an intense immunostaining of S-100 protein from the largest and superficial lesion, and positivity to chromogranin and *somatophysin* of the minor and deeply lesion *confirming the rectal NET diagnosis*. The proliferative activity rate analysis using Ki-67 antibodies showed that both tumors was a low rate of mitotic activity (<1%). Genetic sequence panel identify a novel pathogenic variant in the *NF-1* (deletion of exons 2-30) gene and a variant of uncertain significance in *POLE* (DNA Polymerase Epsilon, Catalytic Subunit; c.1370C>T; p.Thr457Met) gene. These findings confirming a rare association of PN and NET of the rectum and a MTC, in-patient with NF-1 syndrome with a pathogenic variant not previously described in the *NF-1* gene. The patient's postoperative evolution was uneventful, and she remains well, without signs of tumor recurrence, two years after surgical excision. **Conclusion:** Our study demonstrated the first description of co-occurrence of MTC, PN and NET of the rectum in a patient with NF-1 due a new mutation in the *NF-1* gene. The immunohistochemical staining and the inclusion of a multi-cancer genetic panel established the diagnosis.
Fomento	