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Answers to Medical Quiz

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A1. The figure shows proliferation and overgrowth of nerve ganglion cells (S100 positive), nerve fibers and supporting cells in the descending colon, highly suggestive of polypoid intestinal ganglioneuroma[1].

The differential diagnosis for GI ganglioneuroma includes: GI schwannoma, GI neurofibroma, GI perineurioma, mucosal Schwann cell hamartoma and mucosal benign epithelioid nerve sheath tumor[2-4].

A2. There are three groups of ganglioneuromas (GN) of the intestinal tract: polypoid GN, ganglioneuromatous polyposis and diffuse ganglioneuromatosis[2].

A3. The polypoid GN of the gastrointestinal tract can be found in patients with familial adenomatosis coli, Cowden's disease, tuberous sclerosis, juvenile polyposis, von Recklinghausen's disease and multiple endocrine neoplasia type II[2].

Diffuse GN can be associated with MEN (multiple endocrine neoplasia) type II and neurofibromatosis type 1; however, no known systemic disorders have been reported in the literature with ganglioneuromatous polyposis subgroup[5-7].

A4. Depending on their size and location, polypoid GN can cause variety of symptoms, such as, abdominal pain, obstruction, constipation, ileus, appendicitis and weight loss. Incidental finding during endoscopy, surgery or autopsy has also been reported[8].

DISCUSSION

Ganglioneuromas are rare tumors, which occur in patients older than 10 years[9]. No gender difference has been noted, mostly located in the posterior mediastinum, retroperitoneal, adrenal glands, cervix and pelvis[9]. However, cases have been reported to occur in other locations, such as, the skin and gastrointestinal tracts, as in our case[10,11].

Ganglioneuroma could be solitary or multiple and Shekita et al classified ganglioneuromas (GN) of the GI tract into three groups: polypoid GN, ganglioneuromatous polyposis and diffuse ganglioneuromatosis. Mutation in the RET proto-oncogene with methionine to threonine substitution at codon 918 (M918T) has been reported to cause GI ganglioneuromatosis[12-14]. GI ganglioneuromas are generally associated with a variety of symptoms and can occur in association with other conditions.

The histologic diagnosis of ganglioneuromas is based on the identification of ganglion cells by Hematoxylin and Eosin stain. S-100 is to confirm the neural origin of these tumors and could support the diagnosis.

Microscopically, ganglioneuromas show proliferation of spindle-shaped cells, ganglion cells, Schwann cells and nerve fibers.

The relationship between GI ganglioneuromas and malignancy remains unknown, although some colorectal adenocarcinoma cases occurring in patients with intestinal ganglioneuromatosis have been described in the litreature[15,16].

CONCLUSION

We present an extremely rare case of polypoid intestinal ganglioneuroma occurring in a man who presented with left upper quadrant pain and change of bowel habits. The treatment for this variant is endoscopic resection and because of the association of polypoid ganglioneuroma and other systemic diseases, careful follow-up and screening for possible related syndromes and malignancy is highly recommended[17].

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