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SCIENTIFIC LETTER

Schwann cell hamartoma associated with hyperplastic colon polyp: An exceptional finding

Hamartoma de células de Schwann asociado a pólipo hiperplásico de colon: un hallazgo excepcional

A 73-year-old man had a past history of hypercholesterolemia, high blood pressure, hypothyroidism, and squamous cell carcinoma of the larynx treated with surgery, radiotherapy, and chemotherapy. He had no personal or family history of neurofibromatosis (NF), Cowden syndrome, or multiple endocrine neoplasia type 2B (MEN 2B). His habitual treatment was acetylsalicylic acid, levothyroxine, valsartan, and pitavastatin.

The patient was asymptomatic and underwent a complete and adequately clean colonoscopy that was ordered as a follow-up of adenomas. Diverticula were seen in the left colon, along with numerous 3 to 10 mm polyps, all of which were resected with a cold snare. The histologic results were hyperplastic polyps, except for one 5 mm flat-elevated polyp with no erosions or ulcerations on its surface (Fig. 1A), resected in the transverse colon. An associated mucosal Schwann cell hamartoma (MSCH) was identified. This diagnosis was made after the histology and immunohistochemistry (IHC) studies were performed, ruling out other possibilities.

Microscopic examination (Fig. 1B, C) revealed colonic mucosa composed of elongated glands, with serrated morphology in the upper and middle thirds, covered by a columnar epithelium with abundant intracytoplasmic mucin and rounded nuclei located in the intermediate or basal layer, without atypia. Focally, there was stromal expansion due to neural lineage proliferation (S-100+) (Fig. 1D) made up of spindle cells that had fibrillary eosinophilic cytoplasm and nuclei with irregular oval-shaped contours. Some of the nuclei were hyperchromatic and had a certain pleomorphism, but they lacked mitotic activity and marked atypia.

After 18 months of follow-up, the patient remains asymptomatic and has not presented with any complications.

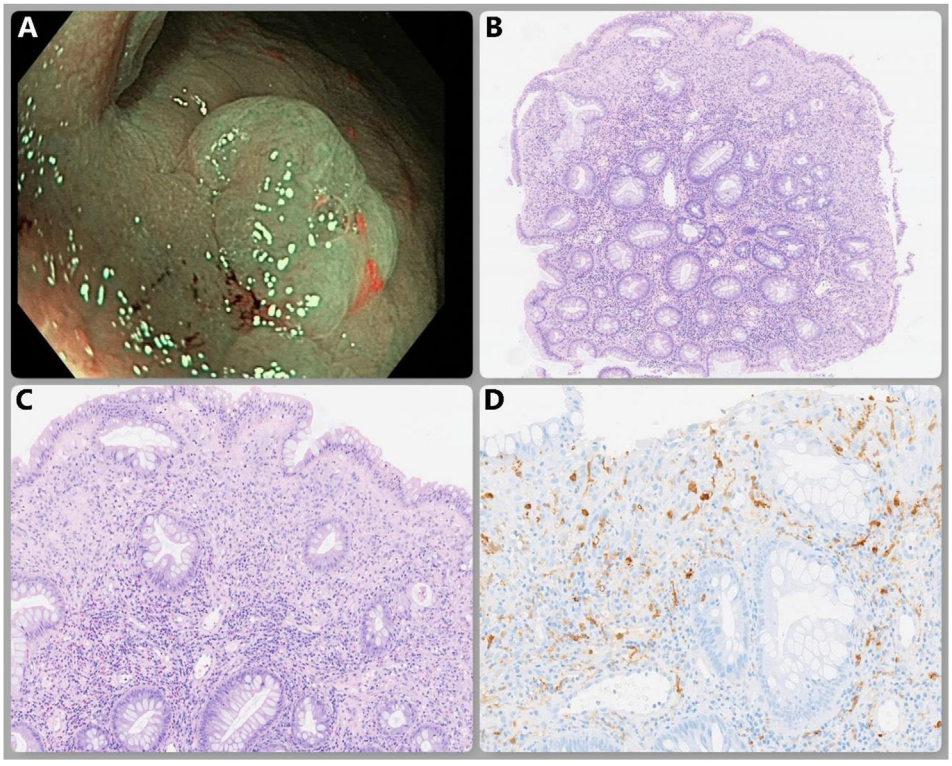

Figure 1 A) Endoscopic image with NBI of a 5 mm polyp in the transverse colon; B) Histology with H&E staining (x5 magnification); C) x10 magnification; and D) S100+ immunohistochemistry.

MSCH is a benign entity of mesenchymal origin that was first described in 2009 by Gibson and Hornick.[1] It consists of neural proliferation without the presence of ganglion cells. It has been identified in different gastrointestinal organs, such as the stomach, gallbladder, and duodenum, but it is most frequently located in the colon.[2–4]

Its endoscopic appearance tends to be that of a 1 to 6 mm polyp.[2,3] When these lesions are evaluated with optical magnification, they have a surface with a Kudo I and II pattern and a non-neoplastic profile,[5,6] but they can also appear as small intramucosal nodules, protruding subepithelial lesions, or even as normal mucosa (an unexpected result of random biopsies).[3,6,7]

MSCHs are extremely rare, with fewer than 100 cases presently reported in the medical literature.[3] They appear to be more frequent in middle-aged women and usually coexist with other colon polyps, such as adenomas or hyperplastic polyps.[4,8]

Cases of MSCHs have been reported in patients with ulcerative colitis, primary sclerosing cholangitis, multiple sclerosis, and acute myeloid leukemia, but its etiology is still unknown.[2,4] They have been postulated to be secondary to an inflammatory process in areas of previously exposed or erosive mucosa.[2]

Found in a majority of asymptomatic patients, MSCH is frequently an endoscopic incidental finding.[8] Nevertheless, it has also been described in patients with diarrhea, melena, abdominal pain, and anemia, as well as in a case of intestinal intussusception.[1,2,8]

The differential diagnosis includes neuroma, perineuroma, schwannoma, mucosal benign epithelioid nerve sheath tumor (MBENST), granular cell tumor (GCT), gastrointestinal stromal tumor (GIST), and colonic leiomyoma.[5,7,9]

No Schwann cells are identified in neuromas or perineuromas (the former present with axon fibers and the latter do not). The presence of Schwann cells and abundant axons leads to suspected neurofibroma that would have positive IHC ganglion cell markers (CD34). If the axons are scarce or absent, MBENST, an intestinal schwannoma, or MSCH would be suspected. Schwannomas are polypoid masses that are not limited to the lamina propria.

IHC is key to making the differential diagnosis: MBENST is positive for CD34 (it also has an infiltrative, epithelioid morphology), GIST is positive for CD117, and leiomyoma is positive for smooth-muscle actin (SMA).[10] Perineuromas are positive for epithelial membrane antigen (EMA) or claudin-1.[2] GCT is more frequent in the esophagus but can be present in the right colon and PAS + and S-100+ cytoplasmic granules would be identified.[9]

MSCH is differentiated from all of the above because it is positive for S-100 and negative for claudin-1, CD34, CD117, and SMA.[9]

Unlike neurofibromas (associated with NF1), neuromas (associated with MEN 2B), and some ganglioneuromas (Cowden syndrome, juvenile polyposis syndrome, MEN 2B, and NF), MSCH is never associated with hereditary syndromes,[9] which emphasizes the importance of a correct differential diagnosis, given the implication in terms of family member genetic counseling.[7,10]

At present, no cases of neoplastic degenerations in MSCH have been reported. Considered a benign lesion, the benefit of a follow-up colonoscopy after polyp resection is doubtful, and if it were to be performed, the most adequate time interval is not known. Given the rareness of the disease, there are no clinical practice guidelines for the management of MSCH.[4] Therefore, it is necessary to report each individual case that is diagnosed, to broaden the knowledge about this entity.

Ethical considerations

The authors declare that no experiments on humans or animals were conducted in relation to this research. The work center protocol on the publication of patient data was followed. The authors declare that neither the images nor the text includes data that could identify the patient, guaranteeing that complete patient anonymity was observed. Nevertheless, informed consent was obtained for the publication of this case.

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Declaration of competing interest

The authors declare that there is no conflict of interest.

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Boerhaave syndrome: A case report of successful conservative treatment

Síndrome de Boerhaave: reporte de caso de tratamiento conservador exitoso

Boerhaave syndrome is the spontaneous rupture of the esophageal wall. It accounts for 8 to 56% of all esophageal perforations and is a potentially life-threatening syndrome.[1]

A 30-year-old man with no previously known comorbidities was admitted to the hospital due to 7 episodes of intense vomiting that had begun one hour earlier, after having eaten an abundant amount of food. The patient presented with intense epigastralgia and retrosternal pain (8/10), as well as dyspnea and odynophagia. Physical examination revealed he was conscious, complaining, polypneic, tachycardic and diaphoretic, with subcutaneous emphysema in the lateral regions of the neck and upper chest. No other alterations were observed. The laboratory work-up reported hemoglobin 16.6 g/dl, hematocrit 51%, leukocytes 26,800 $\times$ $10^3/\mu l$, immature neutrophils 14%, glucose 234 mg/dl, and C-reactive protein 60 mg/l. A contrast-enhanced computed tomography (CT) scan of the neck identified extensive subcutaneous emphysema in the base and lateral regions of the neck (Fig. 1a) and a contrast-enhanced chest CT scan showed pneumomediastinum and pneumopericardium (Fig. 1b). Boerhaave syndrome was suspected, and the patient was kept in a fasting state. Endovenous analgesia and broad-spectrum antibiotic therapy were started. On day 7 of hospitalization, the patient showed clinical and laboratory parameter improvement. Upper gastrointestinal endoscopy was performed that revealed an area of linear erythema in the left wall of the mid-to-distal esophagus (Fig. 2a) and a chest x-ray showed no alterations (Fig. 2b). Oral diet was restarted, the patient's progression was favorable, and he was discharged.

Boerhaave syndrome is the spontaneous rupture of the esophageal wall secondary to retching, vomiting, or any vigorous straining that increases the intraluminal pressure.[2] It affects 3.1 per 1,000,000 patients annually, with morbidity and mortality rates of up to 50%.[2,3] Clinically, 14% of patients seek medical attention due to chest pain, vomiting, and subcutaneous emphysema (the Mackler triad), but symptoms can be nonspecific, making the diagnosis a challenge.[4,5] Contrast-enhanced CT has high diagnostic sensitivity (92-100%), detecting mediastinal involvement, through the presence of air or secondary abscesses.[6] After diagnosis, treatment should be early, significantly decreasing mortality (below 10%).[3,6] Some patients may be candidates for conservative treatment, as long as they are hemodynamically stable, the esophageal rupture is contained, and mediastinal contamination is limited.[7] Said treatment encompasses null oral intake, broad-spectrum antibiotics, and the start of proton pump inhibitor therapy.[6,8] If the patient requires an intervention, endoscopy is a less invasive option that enables the placement of clips and covered metallic stents, as well as endoluminal vacuum therapy. However, surgery is indicated as the first therapeutic option, in patients with large perforations, septic shock, or an associated esophageal disease.[8,9]

In conclusion, Boerhaave syndrome is a rare esophageal disease, with a great risk of complications and death. Timely suspicion and diagnosis, together with individualized treatment, improve the prognosis.

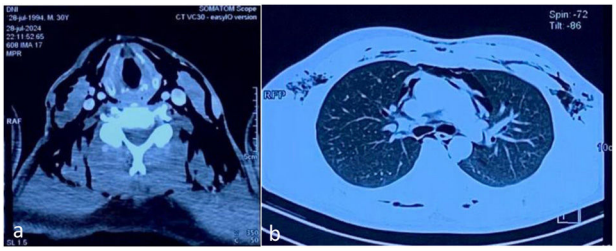

Figure 1 a) Subcutaneous emphysema in the base and lateral regions of the neck; b) Pneumomediastinum and pneumopericardium.

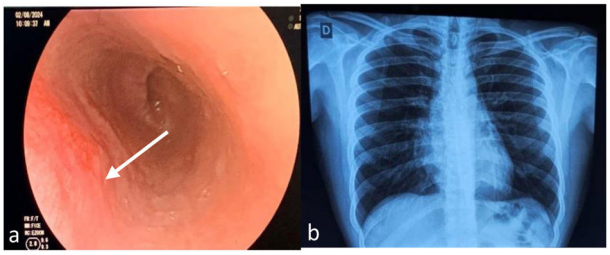

Figure 2 a) Area of linear erythema in the left wall of the mid-to-distal esophagus; b) Normal chest x-ray.