Cowden syndrome: gastrointestinal manifestations of an infrequent polyposis

A 58-year-old man, with a history of total thyroidectomy for papillary carcinoma in 2012, presented with rectal bleeding for the last 4 years, without other digestive symptoms or weight loss. His physical examination revealed macrocephaly (63-cm circumference), skin with multiple verrucous papules of 1–3 mm on the face and dorsum of his hands (which histologically were trichilemmomas), papules in the oral mucosa (hamartomatous papillomas), and a furrowed tongue. The rest of his physical examination and laboratory test results were normal. Colonoscopy showed more than 50 sessile hamartomatous polyps of 2–5 mm in the rectum and sigmoid colon. Upper gastrointestinal endoscopy showed flat whitish lesions of 3–7 mm in the distal esophagus (acanthosis glycogen). In the stomach, multiple sessile polyps of 3–5 mm (Fig. 1) were resected and histology revealed these to be hamartomas (Fig. 2). In the second portion of the duodenum, a few sessile polyps of 4–5 mm were resected (histology revealed normal mucosa). Small-bowel capsule endoscopy revealed multiple polyps of 1–3 mm in the duodenum, proximal jejunum (Fig. 3), and distal ileum (Video 1).

Cowden syndrome is a rare entity related to the PTEN gene. It is characterized by the presence of hamartomatous lesions in any location. The diagnostic criteria (International Cowden Consortium criteria) include major and minor criteria (Table 1). Major criteria include: multiple gastrointestinal hamartomas or ganglioneuromas, macrocephaly, macular pigmentation of glans penis, mucocu-
Table 1 Diagnostic criteria for Cowden syndrome (International Cowden Consortium criteria updated by the National Comprehensive Cancer Network 2018).

<table>
<thead>
<tr>
<th>Major criteria</th>
<th>Minor criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Breast cancer</td>
<td>Autism spectrum disorder</td>
</tr>
<tr>
<td>Endometrial cancer</td>
<td>Colon cancer</td>
</tr>
<tr>
<td>Follicular thyroid cancer</td>
<td>≥3 esophageal glycogenic acanthoses</td>
</tr>
<tr>
<td>Multiple gastrointestinal hamartomas or ganglioneuromas</td>
<td>Lipoma</td>
</tr>
<tr>
<td>Macrocephaly (58 cm in adult women, 60 cm in adult men)</td>
<td>Intellectual disability (IQ ≤ 75)</td>
</tr>
<tr>
<td>Macular pigmentation of glans penis</td>
<td>Papillary or follicular variant of papillary thy-roid cancer</td>
</tr>
<tr>
<td>Mucocutaneous lesions:</td>
<td>Thyroid structural lesions (adenoma, nodules, goiter)</td>
</tr>
<tr>
<td>- One biopsy-proven trichilemmoma</td>
<td>Renal cell carcinoma</td>
</tr>
<tr>
<td>- Multiple palmoplantar keratoses</td>
<td>Single gastrointestinal hamartoma or ganglioneuroma</td>
</tr>
<tr>
<td>- Multifocal or extensive oral mucosal papillomatosis</td>
<td>Testicular lipomatosis</td>
</tr>
<tr>
<td>- Multiple cutaneous facial papules</td>
<td>Vascular anomalies (including multiple intracranial developmental venous anomalies)</td>
</tr>
</tbody>
</table>

Operational diagnosis in an individual (either of the following):
1. Three or more major criteria, but one must include macrocephaly, Lhermitte-Duclos disease, or GI hamartomas
2. Two major and three minor criteria

Operational diagnosis in a family where one individual meets the revised clinical diagnostic criteria for PTEN hamartoma tumor syndrome or has a PTEN mutation (one of the following):
1. Any two major criteria with or without minor criteria
2. One major and two minor criteria
3. Three minor criteria

Competing interests
None

The authors
Daniela Mendez1, Rodrigo Dorelo1, Martín Oricchio1, Martha Otero2, Joaquin Berrueta1, Carolina Olano1
1 Clínica de Gastroenterología, Universidad de la República, Montevideo, Uruguay
2 Cátedra de Anatomía Patológica, Hospital de Clínicas, Montevideo, Uruguay

Corresponding author
Carolina Olano, MD
Hospital de Clínicas, Av. Italia S/n CP, 11600, Montevideo, Uruguay
Fax: +598-2-4872572
carolinaolanouruguay@gmail.com

References

Bibliography
DOI https://doi.org/10.1055/a-0885-9246
Published online: 16.5.2019
Endoscopy 2019; 51: E280–E281
© Georg Thieme Verlag KG
Stuttgart · New York
ISSN 0013-726X

ENDOSCOPY E-VIDEOS
https://eref.thieme.de/e-videos

Endoscopy E-Videos is a free access online section, reporting on interesting cases and new techniques in gastroenterological endoscopy. All papers include a high quality video and all contributions are freely accessible online.

This section has its own submission website at https://mc.manuscriptcentral.com/e-videos

Endoscopy_UCTN_Code_CCL_1AC_2AC