Digestive and Liver Disease 51 (2019) 910

Contents lists available at ScienceDirect

Digestive and Liver Disease

journal homepage: www.elsevier.com/locate/dld

Image of the Month

Cowden syndrome caused by a novel mutation: Endoscopy aided diagnosis

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1. Case description

A 60-year-old man with a history of epilepsy, mental retardation and recurrent meningioma was referred to our endoscopy department for an esophagogastroduodenoscopy, investigating recent weight loss. Physical examination was notable for mild frontal bossing and macrocephaly, no dermatological discrepancies were identified. Esophagogastroduodenoscopy showed numerous clear sessile lesions in the esophagus (Fig. 1), features corresponding with glycogenic acanthosis on biopsy.

Glycogenic acanthosis is characterized by multifocal plaques of hyperplastic epithelium with intracellular glycogen deposits, endoscopically appearing as glassy nodules, which may be sessile or confluent. Although frequently described as being of no clinical significance, glycogenic acanthosis is one of the hallmarks of Cowden syndrome. Timely diagnosis is primordial as this rare multi-system tumor syndrome is associated with an increased risk for colorectal cancer, thyroid cancer, endometrial cancer, urogenital cancer and as suggested recently, meningioma [1].

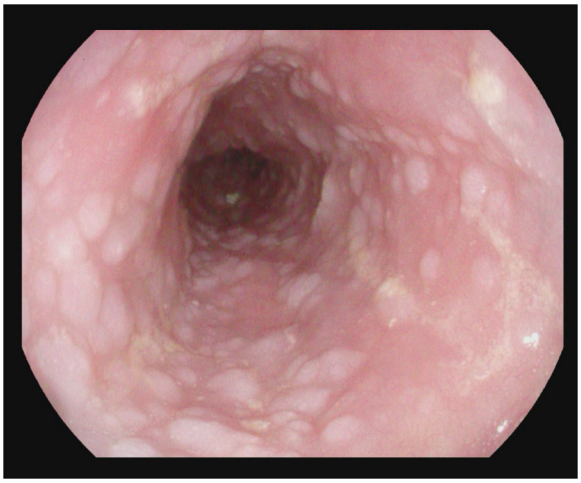

Fig. 1. Upper gastrointestinal endoscopy, demonstrating extensive sessile lesions, covering the entire esophagus. The clear, glassy aspect is typical for glycogenic acanthosis, which was subsequently confirmed by biopsy.

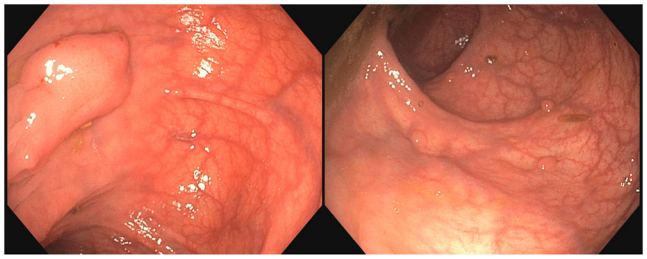

Fig. 2. Left: Endoscopic image at the ascending colon, showing a sessile polyp (LST-NG subtype, 15 × 8 mm, tubular adenoma with low grade dysplasia), for which an endoscopic mucosal resection was performed. Right: Endoscopic image at the proximal rectum. Multiple glassy, sessile polyps were identified in the transverse colon, sigmoid colon and rectum (Paris classification Is and IIa, 2–4 mm, hyperplastic polyps without dysplasia), which were removed by cold snare resection.

In our case, the conjoint occurrence of multiple meningiomata, extensive glycogenic acanthosis, mental retardation and frontal bossing was suggestive of Cowden syndrome. Mutation analysis identified a novel nonsense c.581 T > A mutation in the PTEN-gene at exon 6, leading to a pathogenic premature stop codon, confirming our diagnosis. As Cowden syndrome is also associated with colorectal polyps, a colonoscopy was scheduled, which demonstrated multiple hyperplastic colorectal polyps in the transverse colon, sigmoid colon and rectum (Fig. 2 right), as well as a larger tubular adenoma in the ascending colon, for which an endoscopic mucosal resection was performed (Fig. 2 left).

Our case illustrates that although glycogenic acanthosis is often dismissed as an incidental finding, physicians should be aware of its association with Cowden syndrome.

Funding

None.

Conflict of interest

None declared.

References

[1] Yakubov E, Ghoochani A, Buslei R, Buchfelder M, Eyüpoglu IY, Savaskan N. Hidden association of Cowden syndrome, PTEN mutation and meningioma frequency. Oncoscience 2016;3(5–6):149–55.

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https://doi.org/10.1016/j.dld.2019.03.003