Cowden Syndrome: An Enigmatic Disease with Many Faces

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CASE PRESENTATION

HISTORY

- A 34-year-old Hispanic male presented to clinic for evaluation of iron deficiency anemia.
- His medical history was notable for a non-toxic multinodular goiter and a cerebellar lesion on imaging.
- Physical exam was remarkable for macrocephaly

FINDINGS

- Esophagogastroduodenoscopy (EGD) revealed extensive glycogenic acanthosis throughout the esophagus (Figure 1).
- Duodenal and gastric biopsies were normal. Colonoscopy revealed 37 diminutive sessile polyps.
- Histology showed these to be hyperplastic polyps with lymphoid follicles.

CLINICAL COURSE

- The patient was found to be positive for genetic change in the PTEN gene confirming our suspicion of Cowden Syndrome (CS).
- A small bowel video capsule endoscopy was later completed and found to be unremarkable.

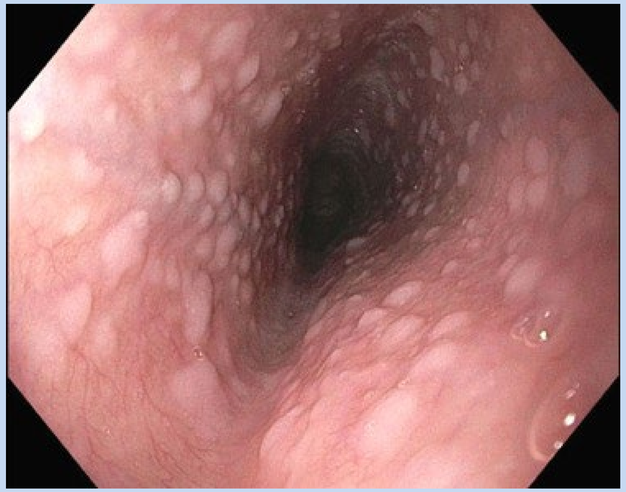

Figure 1: Diffuse glycogenic acanthosis throughout entire esophagus

DISCUSSION

- The *phosphatase and tensin homolog PTEN* hamartoma tumor syndrome is a hereditary cancer syndrome associated with mutations of the *PTEN* gene on chromosome 10.
- CS is the most common phenotype
- CS is a rare autosomal dominant disorder characterized by multiple hamartomas and hyperplastic lesions in the mucous membrane, skin, GI tract, thyroid, and brain.
- It has an estimated incidence of 1 in 250,000 people
- Diffuse glycogenic acanthosis and gastrointestinal polyposis are characteristic features of CS; seen in close to 80% and 65-93%, respectively.
- It was previously believed that CS-associated polyposis did not confer an increased risk of colorectal cancer (CRC); however, recent literature reports the risk of malignancy can reach 10-18%.
- Our patient had many features suggestive of CS including macrocephaly, cerebellar gangliocytoma, extensive glycogenic acanthosis, and numerous colon polyps.
- He will continue to undergo surveillance for CRC as well as non-GI malignancies including thyroid, skin, and renal.
- While a mutation of the PTEN gene was found, only 40-60% of patients have an identifiable mutation on genetic testing; as such, recognition of physical manifestations and endoscopic findings is critical to diagnosis.
- Esophageal glycogenic acanthosis is often a benign finding on EGD; however, its diffuse presence in conjunction with colonic polyposis should raise high suspicion for CS.