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IMAGE

Cowden syndrome

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Cowden syndrome (CS), first described in 1963, is an autosomal dominant disorder associated with mutations in the phosphatase and tensin homolog (PTEN) gene, incomplete penetrance, and variable expressivity. Gastrointestinal manifestations include intestinal ganglioneuromas and esophageal acanthosis [1]. The clinical criteria proposed by the International Cowden Consortium are used to diagnose the syndrome. The Cleveland Clinic has suggested a PTEN score calculator based on clinical manifestations to select patients for PTEN mutation testing [2]. Due to CS association with malignant diseases, early diagnosis and periodic follow up examination in patient and their family members is necessary.

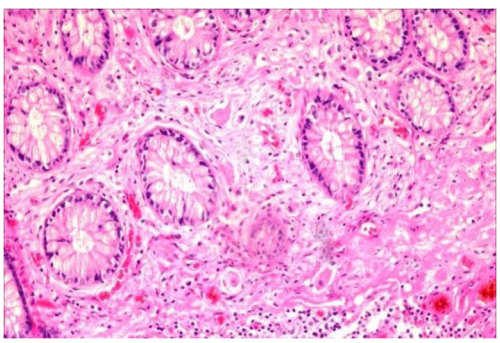

Fig. 1 Colonic polyps showing multiple nests predominantly in the lamina propria that contain neural tissue and ganglion cells suggestive of ganglioneuromas

The images are from a 38-year-old male investigated for intermittent hematochezia. Past history was significant for lipoma, dysplastic nevi, and family history for colon cancer in father and breast cancer and benign thyroid nodules in sister. This patient had macrocephaly, with multiple polyps at colonoscopy which were all ganglioneuroma on biopsy (Fig. 1). Upper gastrointestinal endoscopy revealed diffuse esophageal glycogenic acanthosis (Fig. 2) and gastric hyperplastic polyp, and the patient was referred to genetic counseling and was positive for PTEN mutation.

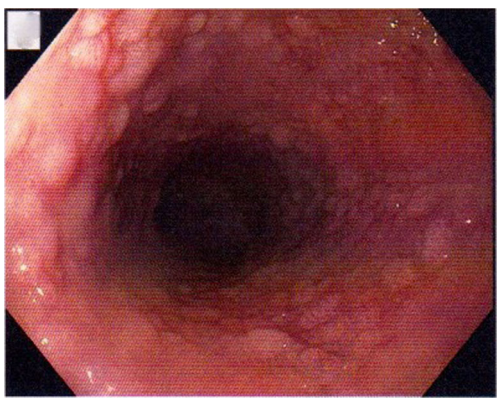

Fig. 2 Esophagogastroduodenoscopy showing esophageal glycogenic acanthosis

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