Peutz jeghers syndrome with multiple endocrinal failures

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Introduction

Peutz-Jeghers syndrome (PJS) is a rare familial disorder with an incidence of 1 in 12-30,000 live births characterized by mucocutaneous pigmentation, gastrointestinal and extra gastrointestinal hamartomatous polyps and an increased risk of malignancy.

Case Presentation

We report a 22-year-old female hypothyroid since age of 4 , type 1 diabetic since age of 11, who presented with melaena. She gave a long history of diffuse abdominal pain without precise localization no specific relieving or precipitating factor. No family history of colonic polyps and cancer colon

Physical examination: pallor ,dark brown pigmented maculae on perioral ,lower lip,fingers well demarcated dark brown to blue –black pigmented macules on tounge and buccal mucosa,angular stomatitis (fig 1 a,b,c,d)

Soft rubbery swelling on the right and left forearm, anterior abdominal wall

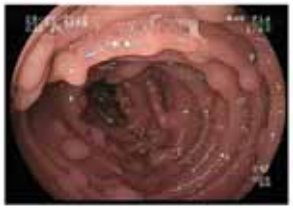
Fundus examination ; back ground diabetic retinopathy

Laboratory investigation: showed ;microcytic hypochromic aneamia **Iron**; 15ng/ml **TIBC**; 333 **ferritin**;7.46ng/ml **Ca** ; 5.1 mg/dl , **phosphrous**;1.8 mg/dl **PTH**:29.8 pg/ml ,**Vit D** ;33ng/ml **Transglutaminase IGA** : negative , **Cortisol am**; 16ug/dl **Antithyroglobulin**; negative, **antimicrosomal antibodies**;negative

Endoscopic investigation

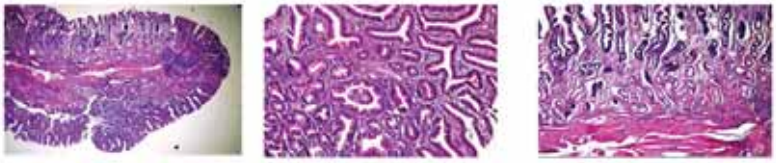
Colonoscopy .Multiple sessile polyps were detected in the rectum. Argon photocoagulation was done to a small polyp found in the rectum

Enteroscope: gastric mucosa is hyperemic shows white spots mostly the fore-runners of coming polyps ,polyp at the pyloric ring 3 mm in diameter, mucosa of the duodenum ,jujenum and ileum show ennumerable small polyps 2-4 mm



Multiple biopsies were taken from polyps ,sent for pathology report

Pathology report: hamartomatous polyps formed of proliferating glands with regular (fig 3 a,b,c) architecture with intervening twigs of smooth muscle fibers, no evidence of dysplasia or malignancy.



Pathology report of skin nodules: capsulated benign growth formed of lobules of mature fat cells , lipoma.

Conclusion

A case of peutz Jeghers syndrome with multiple lipomatosis ,she is type 1 diabetic with early childhood hypothyroidism ,vitamin D deficiency .These combinations of different aetiologies in the same patient might raise the suspension of a new syndrome waiting for other observational studies

Key words;
peutz-Jeghers syndrome, type 1 diabetes, hypothyroidism ,multiple lipomatosis