Peutz-Jeghers syndrome with multiple endocrinial failures

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 melena. 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 tongue 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 anemia Iron; 15ng/ml TIBC; 333 ferritin; 746ng/ml Ca ; 5.1 mg/dl , phosphorous; 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 ,julenum 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