Peutz Jeghers Syndrome

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First described in 1921 by a Dutch Physician Jan Peutz

In 1949 Harold Joseph Jeghers and K H Katz published Peutz - Jeghers Syndrome in New England.Journal of Medicine

Melanotic macules are seen in the skin. Lips ,oral cavity ,hands and feet
There is a family history
Hamartomatous Polyps in the GIT ,Benign with low Malignant potential

Molecular testing
STK11 /LKB1 gene mutation

These patients have a risk of Developing cancers in the breast ,colon ,stomach, rectum and Pancreas

Incidence
1 in 20000
to
1 in 300000
Births

Autosomal dominant

1 Benign hanartomatous polyps in GIT
2 Pigmented macules in the lips and oral mucosa ,
Melanosis

Bowel obstruction
Intussusception
Anemia
Polyps

In 1998 a tumor suppressor genes STK11 /LKB1
Autosomal dominant

Arborisation of smooth muscle within lamina propria

Melanosis and melanocytic macules

Intussception
Obstruction
Anemia
Polyps
Ovarian cysts
Ovarian sex chord Stromal Tumors

Polyps seen in Small intestine ,colon ,stomach
Also in the lungs ,nose ,Urinary Bladder rectum

PJ Syndrome diagnosis
1 two or more PJ Polyps
2 any no of PJ Polyps with family history
3 dark spots with.family history
4 any no of PJ Polyps and dark spots

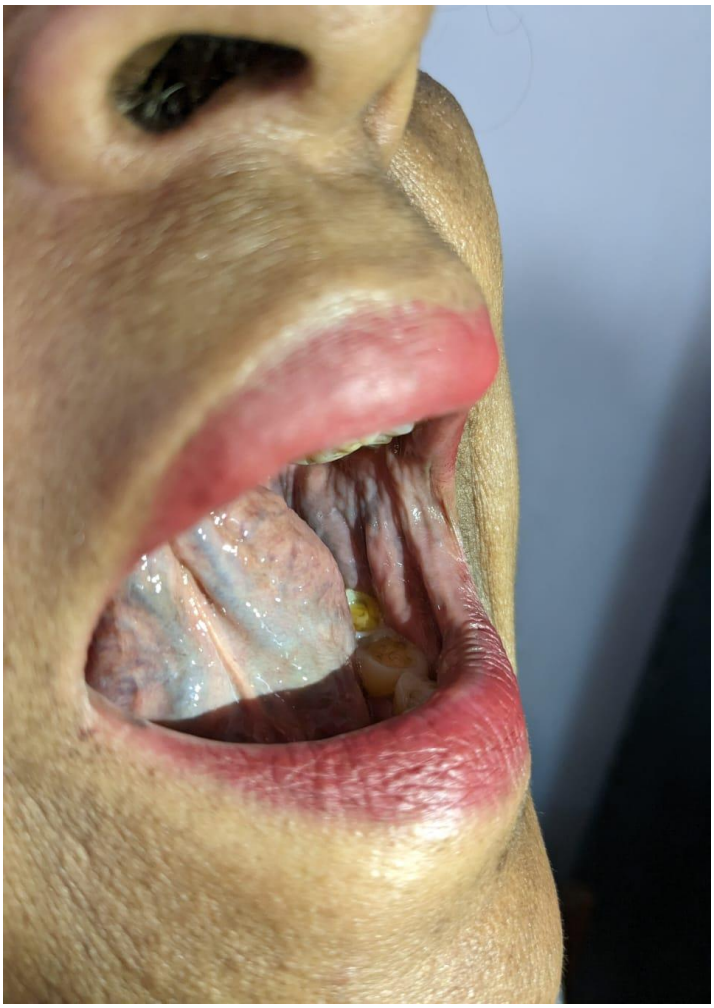
Genetic testing
Genetic counselling

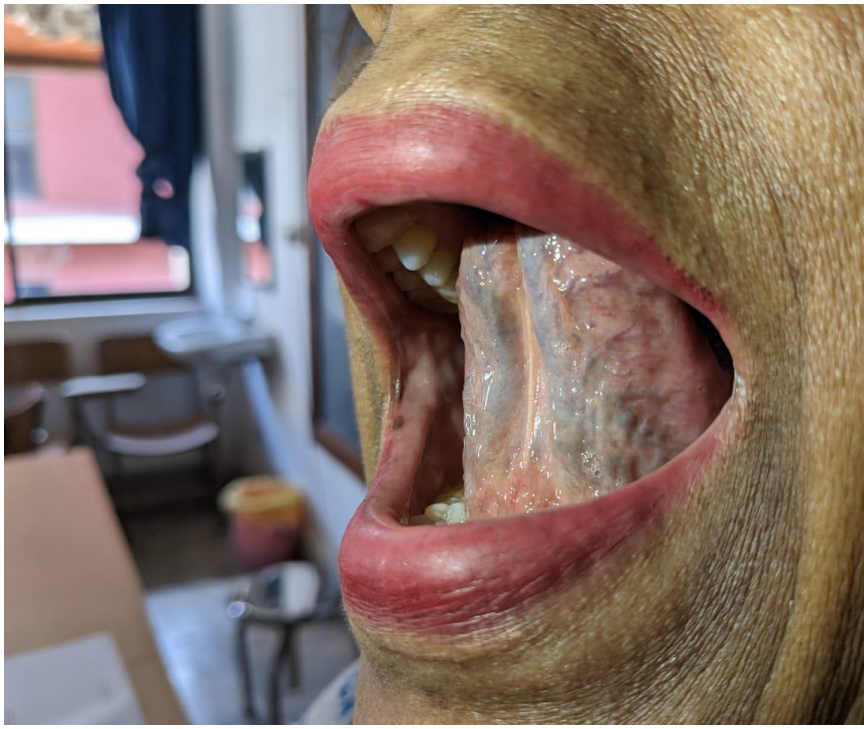
Dr RG Wiseman Pinto
Dr Amala Kudchadkar

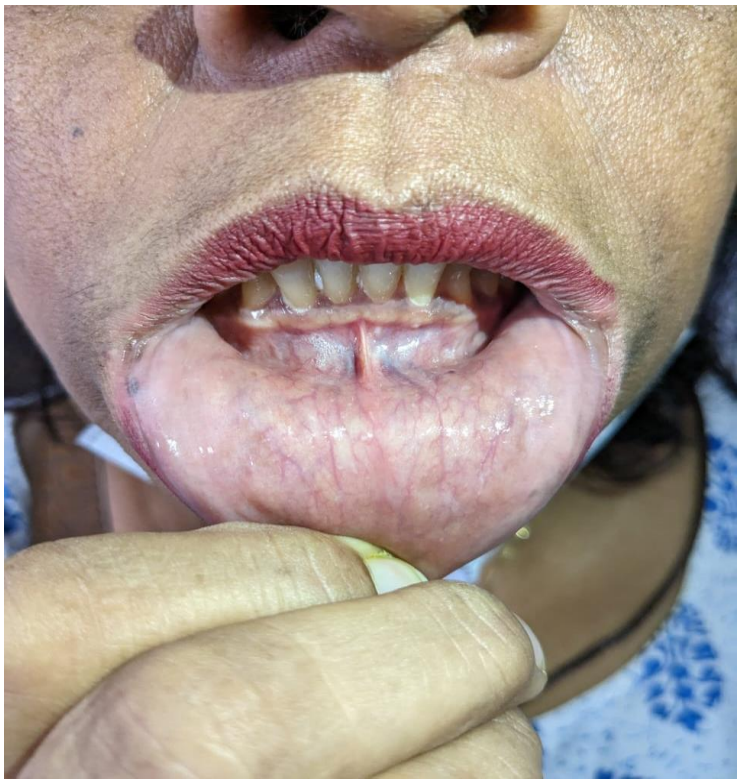
Dr JF Alvares

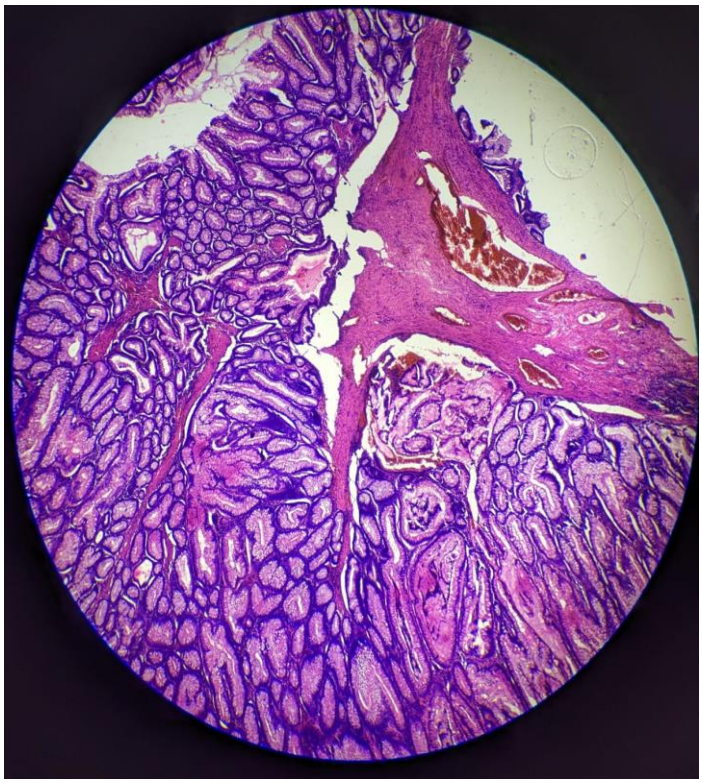
Peutz -Jeghers Polyp
In Sigmoid Colon
61 y Female

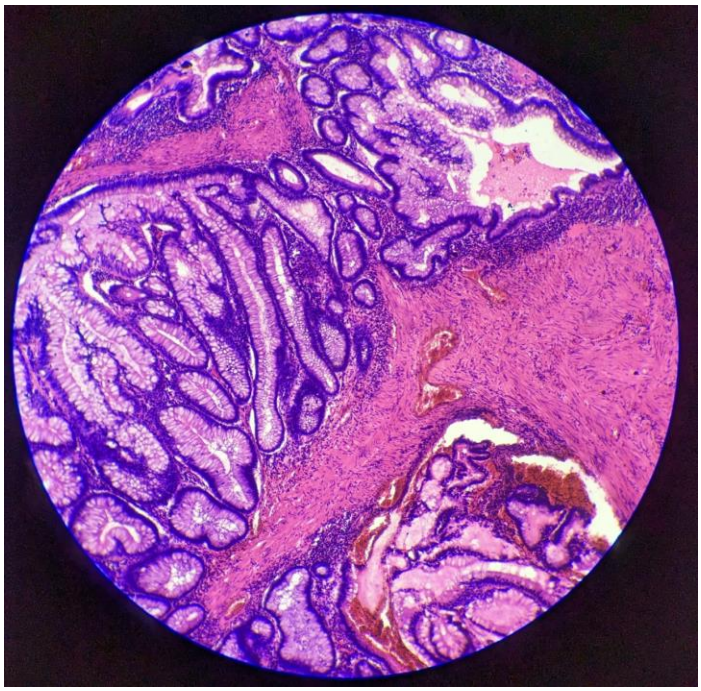
Endoscopic biospy done and sent by
Dr JF Alvares
Victor Hosp Margao
Peutz - Jeghers Syndrome
Muco cutaneous pigmented macules and endoscopic pics of polyp

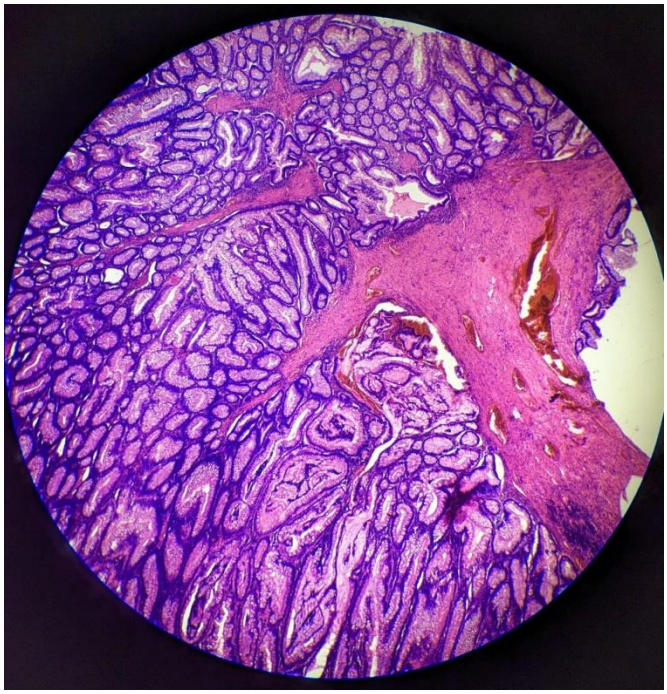












GASTRO-DUODENOSCOPY REPORT

Findings

Cricopharynx is normal

Esophagus shows normal mucosa

Fundus and body of stomach are normal

Antrum - Erosions present in the antrum, antral biopsy taken for RUT for H - Pylori

Duodenal bulb is normal

DII is normal

Conclusions

Antrum - Antral erosions, antral biopsy taken for RUT for H - Pylori

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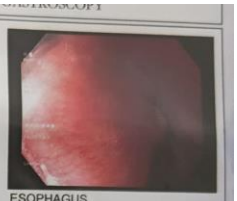
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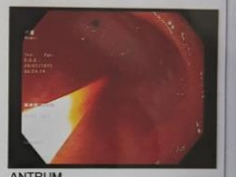
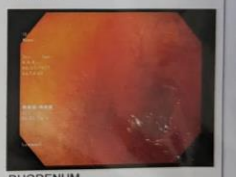
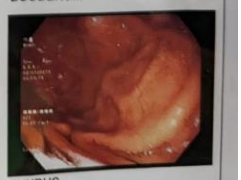

ANTRUM


DUODENUM


FUNDUS

COLONOSCOPY REPORT

Findings

A large pendanculated polyp seen in the proximal sigmoid colonoscopic polypectomy done - no post complication

Conclusions

Colonic polyp - Colonoscopic polypectomy done

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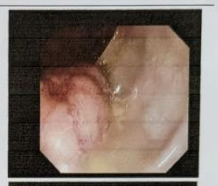

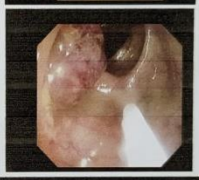

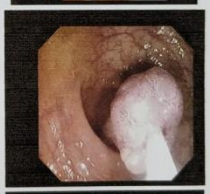

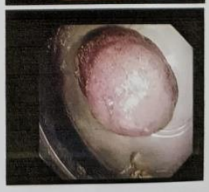

Procedure: COLONOSCOPY

VIDEOCOLONOSCOPY REPORT

Findings

Colonoscope passed upto cecum

At 30 - 32cms from the anal verge there is a large pedanculated polyp with a long stock in the sigmoid colon

Rest of the colonic mucosa is normal

Conclusion

Pedanculated sigmoid polyp

Plan for colonoscopic polypectomy

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