PEUTZ-JEGHERS SYNDROME: One Nurse's Personal Experience

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Objectives

- Define Peutz-Jeghers Syndrome (PJS)
- Describe the management and treatment of PJS
- Discuss the patient experience associated with the diagnosis of PJS

What is PJS

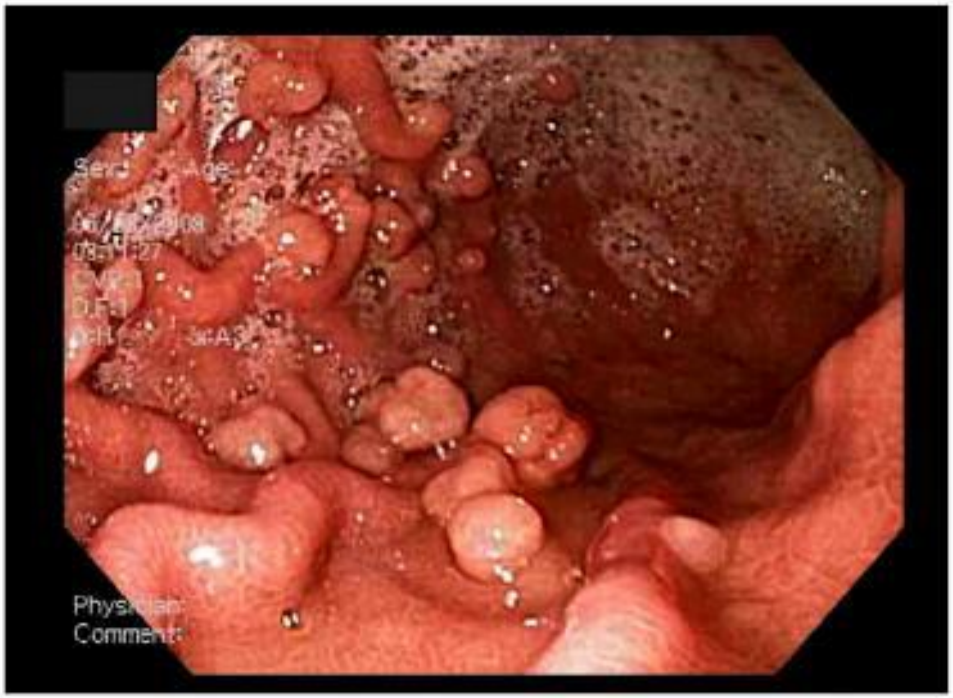
- 1921 Dr Jan Peutz
  - First published case report
- 1949 Dr Harold Jeghers
  - Detailed descriptive report

PJS

- Inherited Autosomal dominant trait
- Rare disorder
  - 1:8000 to 1:200,000
- Multiple hamartomatous polyps
- Mucocutaneous pigmentation

Clinical Manifestations

- Hamartomatous polyps/Juvenile polyps
  - Benign malformation made up of abnormal cells and tissues
  - Commonly occur in small bowel
    - Usually jejunum 60-90%
    - Stomach 15-30%
    - Colon 50-64%
  - Polyps develop in first decade of life
    - Symptomatic between ages 10-30
  - May occur outside the GI tract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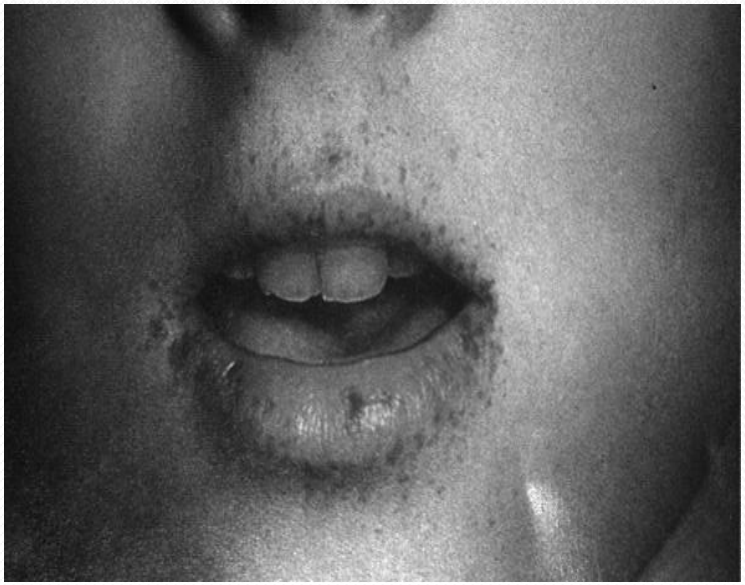
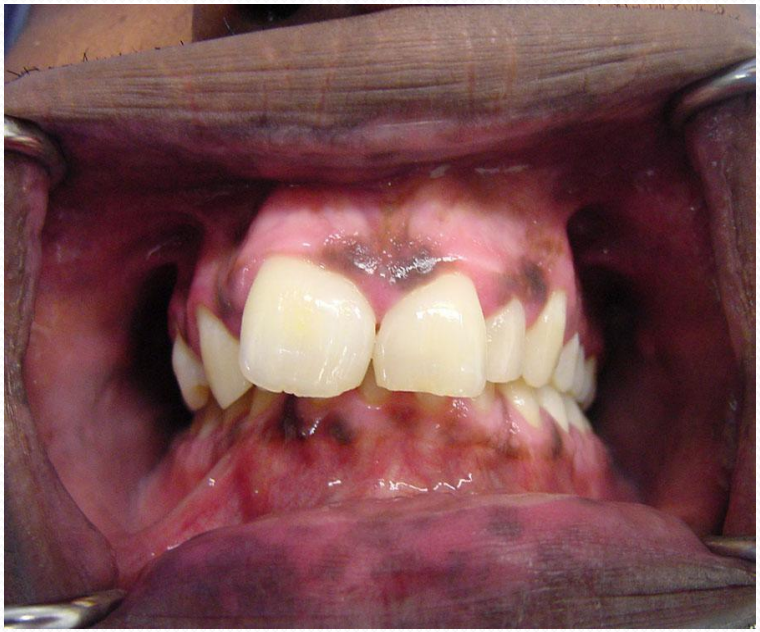
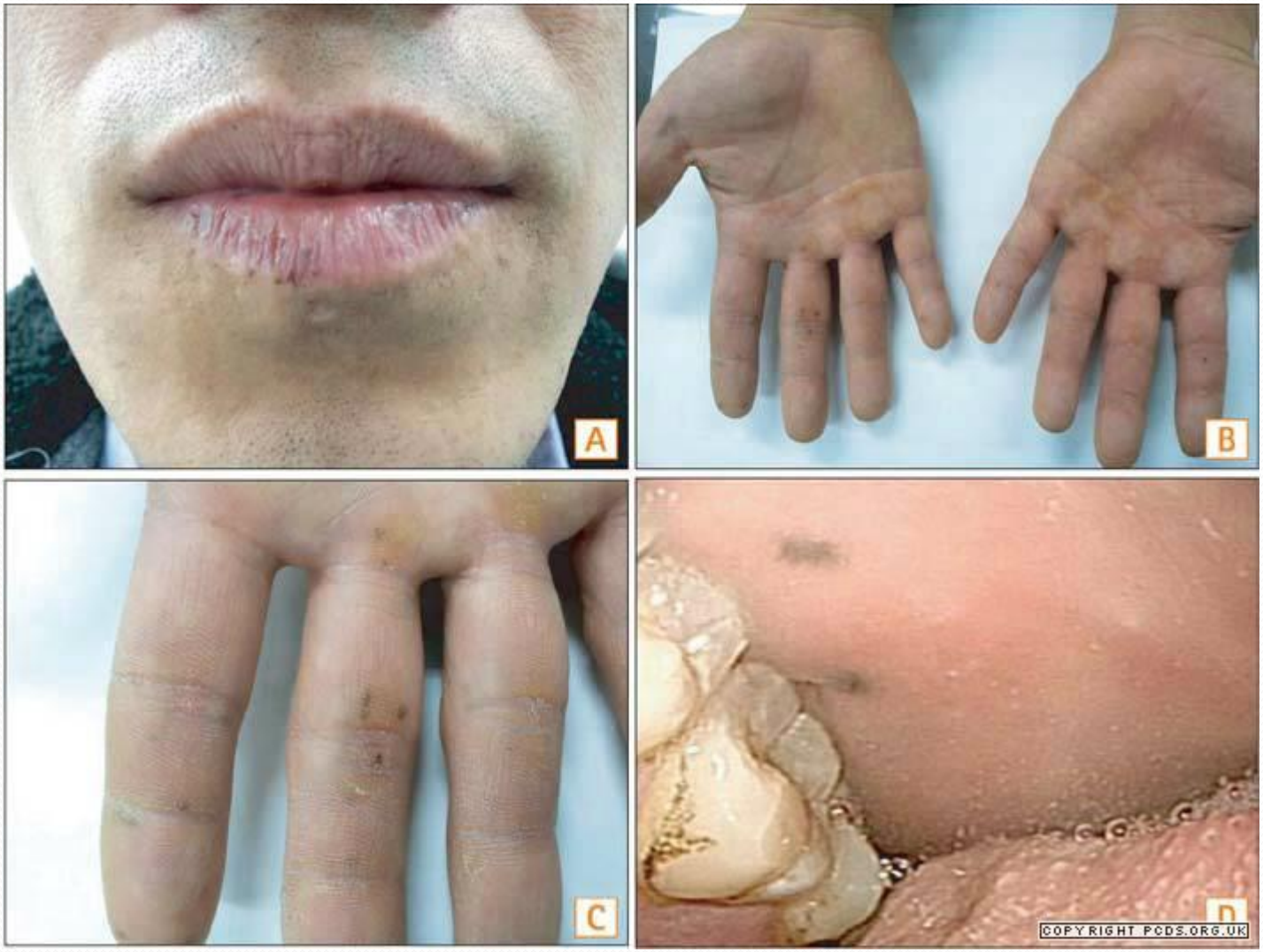
Clinical Manifestations

- Mucocutaneous pigmentation
  - Melanin spots
  - Occur in 95% of individuals with PJS
  - Flat, blue- gray to brown spots
  - Lips, perioral, buccal, nose, palms, soles, perianal
  - Fade after puberty



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Genetics

- PJS is associated with germ line mutation in the STK11 (serine/threonine kinase 11) tumor suppressor gene
  - Responsible for the clinical manifestations of PJS
  - Rare
    - Males and females equally affected

Diagnostic Criteria

- Presence of the following
  - Two or more histologically confirmed PJ polyps
  - Any PJ polyps in an individual with a family history of PJS in a close relative
  - Mucocutaneous pigmentation in an individual with a family history of PJS in a close relative
  - Any number of PJ polyps in an individual who also has mucocutaneous pigmentation

Diagnosis

- Disorders also associated with hamartomatous polyps must be ruled out
  - Cowden syndrome
  - Bannayan- Riley Ruvalcaba syndrome
  - Juvenile polyposis syndrome

- Disorders associated with mucocutaneous pigmentation must be ruled out
  - Laugier-Hunziker syndrome

Genetic Evaluation

- Completed after clinical diagnostic confirmation

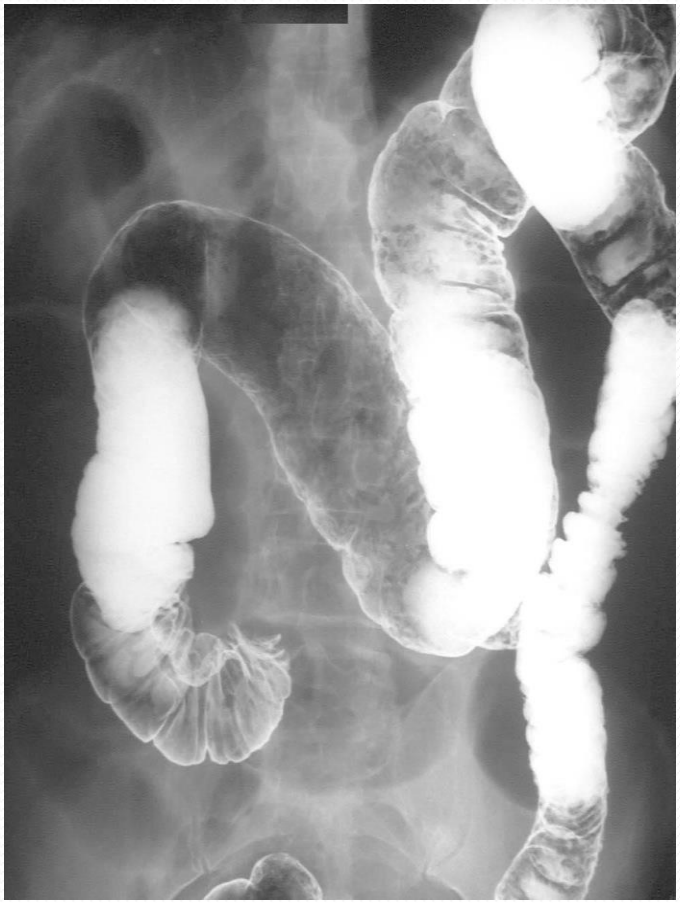
- Genetic testing for germline mutation in STK11 gene
  - Confirms diagnosis of PJS
  - Absence of STK11 mutation in individuals who meet the clinical criteria of PJS does not exclude the diagnosis
  - Genetic testing of all at risk relatives

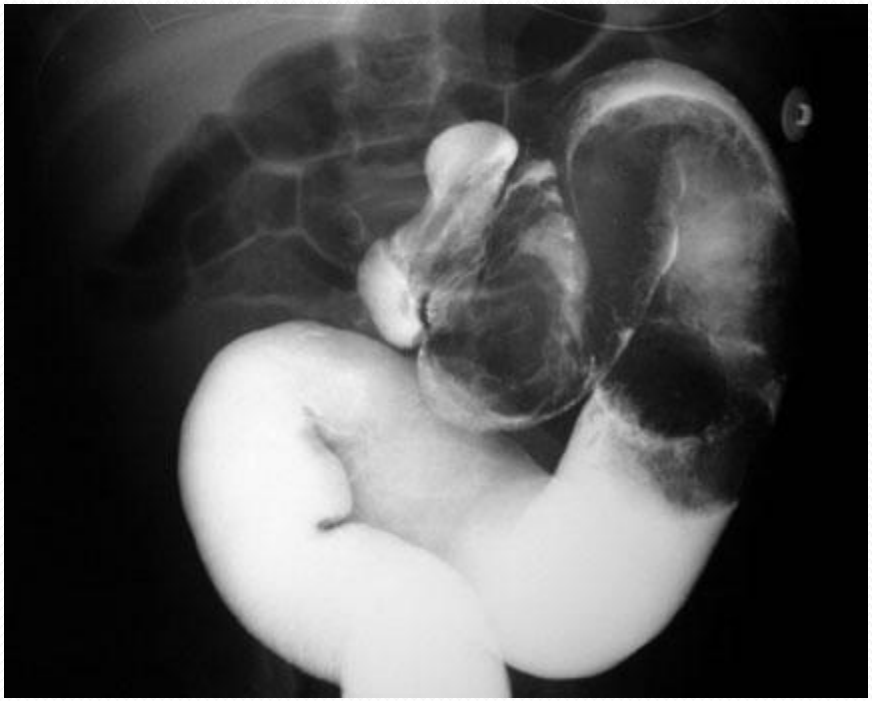
Characteristics Associated withPJS

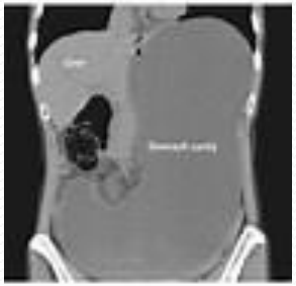
- Family history of PJS
- Recurrent abdominal pain in individuals younger than 25 yo
- Unexplained GI bleeding in young individuals
- Menstrual irregularity
- Gynecomastia
- Precocious puberty

Complications Associated with PJS

- Recurrent abdominal pain
- Gastric outlet obstruction
- SBO
- Intussusception
- IDA
- Hematemesis
- Melena and Rectal bleeding







Cancer and PJS

- Increased lifetime risk of intestinal as well as extraintestinal malignancy
- Risk increases with age
- Greater risk in females than in males
  - GI
  - Pancreatic/biliary
  - Gynecologic
  - Breast
  - Thyroid
- Most common malignancy is colorectal, followed by breast and small bowel

ACG Guidelines

- Appropriate genetic screening and counseling
- GI specialist familiar with PJS
- Urologic and gynecologic consultation
- Consider referral for psychiatric care PJS is associated with depression
- Lab studies
  - CBC, Iron studies, hemoccult, CEA
  - CA-125 every year starting at age 18
  - CA-19-9 every 1-2 years starting at age 25

Imaging Studies

- EGD
- Colonoscopy
- Enteroscopy
- EUS
- ERCP
- Capsule
- MRI
- CT with contrast
- UGI with SBFT

Monitoring and Surveillance

- Multidisciplinary
  - EGD every 2-3 years
  - Enteroscopy/capsule every 2 years
  - MRI every 2 years
  - Colonoscopy every 2-3 years
  - EUS every other year beginning at age 25
  - Mamogram every other year at age 18 then yearly at age 50
  - Annual pelvic exam and US at age 20
  - Annual testicular exam

Complications and Surgery

- Intestinal obstruction
- Abdominal adhesions
- Short bowel syndrome

Research

- Chemopreventive strategies
  - COX inhibitors
    - Shown to reduce polyposis in mice treated with celexa
  - mTOR inhibitor Rapamycin
    - Shown to reduce polyposis in mice
  - Everolimus
    - Proposed as chemopreventative agent

TWO Remarkable Patients

References

Thank you