Peutz-Jeghers Syndrome

Synonyms: polyposis, hamartomatous intestinal polyps-and-spots syndrome

Peutz-Jeghers syndrome is an autosomal dominant disorder characterised by mucosal pigmentation of the lips and gums with multiple intestinal hamartomatous polyps. There is an associated marked increased risk of certain malignancies, especially gastrointestinal (GI) cancers and breast cancer.

The syndrome is co-named after Peutz, who in 1921 first recognised the association of GI polyposis and mucocutaneous pigmentation, and Jeghers, who wrote the definitive descriptive reports and drew more widespread attention to the condition in 1949.

Epidemiology

- The estimate of the prevalence of Peutz-Jeghers syndrome is about 1 in 50,000.
- Autosomal dominant condition with high penetrance.
- In up to two thirds of cases, mutations can be identified in the serine/threonine kinase gene STK11(LKB1) on chromosome 19 (19p13.3). STK11 is thought to be a tumour suppressor gene.

Presentation

- Family history: asymptomatic but requesting investigation/counselling.
- Deeply pigmented lesions on the lips (cross the vermillion border) and buccal mucosa. These may also be present on the hands and feet (particularly the palms and the soles) and around the anus and genitalia. These lesions may be most prominent in infancy and fade after puberty.
- Repeated bouts of abdominal pain in a young patient (due to obstruction or intussusception).
- Unexplained intestinal bleeding in a young patient or iron-deficiency anaemia.
- Rectal prolapse.
- Precocious puberty.
- Nasal, bronchial, biliary tract, uterine or bladder polyps.

Investigations

- FBC: may show anaemia.
- Iron studies may show iron deficiency.
- Faecal occult blood tests to check for GI bleeding.
- Endoscopy: small bowel follow-through or capsular endoscopy; colonoscopy to determine presence and location of intestinal polyps.
- Genetic analysis.

Management

- If Peutz-Jeghers syndrome is suspected on the basis of clinical criteria, the patient should be referred to a regional genetics centre for formal counselling and mutation analysis of the relevant gene.
- Surgical excision of lesions may be required:
  - Endoscopic polypectomy for diagnosis and control of symptoms.
  - Polypectomy using double balloon enteroscopy may prevent the need for repeated urgent operations and small bowel resection that leads to short bowel syndrome.

Surveillance

Colorectal surveillance: large bowel surveillance is recommended two-yearly from the age of 25 years. The intervention should visualise the whole colon and so colonoscopy is the preferred mode of surveillance.
Members of the family of an affected family where a causative gene has been identified should be referred for gene counselling and predictive gene testing. Where they test negative, there is no indication for their continued surveillance.[8]

Complications

- High cancer risk:
  - Almost 50% of patients with Peutz-Jeghers syndrome develop and die from cancer by the age of 57 years.[3]
  - Most common cancers are gastro-oesophageal, small bowel, colorectal and pancreatic.
  - There is also risk of ductal breast cancer, thyroid, lung, uterine, Sertoli cell testicular tumours or ovarian sex cord tumours.
  - The cancer susceptibility is thought to arise from mutation activating serine/threonine kinase.

- GI bleeding.
- Intussusception.
- Rectal prolapse.
- Adhesions and obstructions from repeated operations.

Prognosis[9]

Cumulative risks for developing cancer:
- by 20 years - 2%
- by 30 years - 5%
- by 40 years - 17%
- by 50 years - 31%
- by 60 years - 60%
- by 70 years - 85%

Cumulative risk of developing GI cancers (gastro-oesophageal, small bowel, colorectal and pancreatic):
- by 30 years - 1%
- by 40 years - 9%
- by 50 years - 15%
- by 60 years - 33%

Cumulative risk of breast cancer in women with Peutz-Jeghers syndrome:
- by 40 years - 8%
- by 60 years - 31%

Further reading & references

3. Mukherjee S; Peutz-Jeghers Syndrome, Medscape, Sep 2011
4. Guidelines for colorectal cancer screening and surveillance in moderate and high risk groups; British Society of Gastroenterology (May 2010 update from 2002)
5. Peutz-Jeghers Syndrome, PJS; Online Mendelian Inheritance in Man (OMIM)
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