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Editorial

One of the Hereditary Polyposis Syndromes: Peutz-Jeghers Syndrome in Children

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Peutz-Jeghers Syndrome (PJS) is an inherited polyposis syndrome. It is characterized by development of multiple hamartomatous gastrointestinal polyps, mucocutaneous freckling, and an higher risk of gastrointestinal (GI) and non-gastrointestinal cancer. The risk of small bowel intussusception is as high as 68% by age 18 years [1].

Epidemiology and genetics: Its incidence is estimated between 1/8300 and 1/200,000 [2]. There is a germline mutation in the STK11 (LKB1) gene. This gene is a tumor suppressor gene and is responsible for clinical findings in PJS. STK11 gene mutations are detected in only 50 - 80% of families with PJS. 10 - 20% of patients with PJS have no family history, de novo mutation is present [1].

Clinical manifestations

- Mucocutaneous pigmentation
- Hamartomatous polyps
- Increased risk of gastrointestinal and non-gastrointestinal cancers.

Mucocutaneous pigmentation: It occurs in more than 95% of people with PJS. The lips and perioral area (94%) are the most frequently affected, followed by the palmar surface of the hands (74%), buccal mucosa (66%) and soles (62%). It may also be seen on the nose, perianal region, genital organs and rarely in the intestines [1].

Hamartomatous polyps: The majority of patients with PJS have GI hamartomatous polyps. It occurs most frequently (60 - 90%) in the jejunum of the small intestine. It can also be discovered elsewhere in the GI tract. Patients frequently present with GIS obstruction caused by intussusception or polyps, abdominal pain due to infarction, anemia due to bleeding or extrusion of the polyp through the rectum, despite the fact that 50% of them are asymptomatic at the time of diagnosis [1].

A clinical diagnosis of Peutz-Jeghers syndrome is made when any of the following is present [1,3]:

1. 2 or more PJS type hamartomatous polyps in the GI tract

2. typical mucocutaneous pigmentation in someone with a positive family history
3. any number of PJS-type polyps in someone with positive family history
4. any number of PJS-type polyps in an individual with characteristic mucocutaneous pigmentation.

Genetic analysis: Patients with PJS who satisfy the diagnostic requirements should unquestionably have a genetic test for the STK11 gene mutation. Genetic testing is recommended for asymptomatic children at risk, beginning at the age of three, and earlier if symptoms are present in children in the risk category [1]. Parents with PJS transmit the disease to their children at a rate of 50%. Genetic counselling is recommended to parents for pre-implantation genetic diagnosis to detect the STK11 mutation [4].

Gastrointestinal surveillance: In asymptomatic people, endoscopic screening is advised no later than 8 years. If patient is symptomatic, it should be performed earlier. Gastroduodenoscopy, video capsule endoscopy, and colonoscopy are performed. Follow-up with endoscopy is performed every 3 years, earlier investigation should be done if there are symptoms. MR enterography should be performed if capsule endoscopy could not be performed [1].

Management of polyps [1]:

- If polyp is absent or < 10 mm, video capsule endoscopy is performed every 3 years
- If polyp is > 15 mm, endoscopic polypectomy is done
- Symptomatic small bowel polyps or polyps >10 mm should be removed to prevent polyp-related complications
- Polypectomy with balloon enteroscopy should be done in experienced hands.

GI cancers: PJS polyps do not develop into cancer in childhood. The lifetime cancer risk of patients with PJS is between 38 and 66% [5].

Chemoprevention: Many drugs (COX-2 inhibitors, Rapamycin etc.) have been used to decrease the number of polyps, but there are still insufficient evidence to recommend their use [6-8].

Bibliography

1. Latchford A., *et al*. "Management of Peutz-Jeghers Syndrome in Children and Adolescents: A Position Paper from The ESPGHAN Polyposis Working Group". *Journal of Pediatric Gastroenterology and Nutrition* 68 (2019): 442-452.
2. Huang Z., *et al*. "Clinical characteristics and STK11 gene mutations in Chinese children with Peutz–Jeghers syndrome". *BMC Gastroenterology* 15 (2015): 166.
3. Beggs AD., *et al*. "Peutz-Jeghers syndrome: a systematic review and recommendations for management". *Gut* 59 (2010): 975-986.
4. Xu X., *et al*. "Multidisciplinary management for Peutz-Jeghers syndrome and prevention of vertical transmission to offspring using preimplantation genetic testing". *Orphanet Journal of Rare Diseases* 17 (2022): 64.
5. Van Lier MG., *et al*. "High cancer risk in Peutz-Jeghers syndrome: a systematic review and surveillance recommendations". *The American Journal of Gastroenterology* 105 (2010): 1258-1264.
6. Udd L., *et al*. "Suppression of Peutz-Jeghers polyposis by inhibition of cyclooxygenase-2". *Gastroenterology* 127 (2004): 1030-1037.

7. Wei C., *et al*. "Suppression of Peutz-Jeghers polyposis by targeting mammalian target of rapamycin signaling". *Clinical Cancer Research* 14 (2008): 1167-1171.

8. Robinson J., *et al*. "Oral rapamycin reduces tumour burden and vascularization in Lkb1þ/- mice". *The Journal of Pathology* 219 (2009): 35-40.

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