The diagnosis of Peutz-Jeghers syndrome

J. L. González Muñoz, M. Angoso Clavijo, C. Esteban Velasco, A. Rodríguez Pérez, L. Muñoz Bellvis and A. Gómez Alonso

Services of Surgery and Digestive Diseases. Salamanca University Hospital. Salamanca, Spain

A 23 year-old woman with chronic anemia showed abdominal pain four hours before being admitted. Her family history included a grandfather with small-bowel polyposis, a father with skin melanosis, and a brother diagnosed with Peutz-Jeghers syndrome. He developed a solitary giant rectal polyp that prolapsed.

On physical examination, the most remarkable sign encountered was the presence of hyperpigmented maculae on the face, lips, and oral mucosa (Fig. 1). She was operated on for ileocolic invagination. Forty centimeters of ischemic ileum were removed, and the pathological study showed twelve polyps with different sizes (Fig. 2).

The postoperative period was uneventful, and the patient was discharged at day seven. Later on we completed the study with oral endoscopy and colonoscopy. Dozens of millimetric gastric polyps were seen, and four colonic polyps were removed. The pathological diagnosis was hamartomatous and adenomatous polyps (Fig. 2).

The definite diagnosis of Peutz-Jeghers syndrome requires the presence of hamartomatous polyps and two of the following items –family history, skin hyperpigmentation, and small-bowel polyps (1). Our patient met all of these criteria. Furthermore, she had oral maculae, which are specific of this syndrome, and her case was similar to the original one reported by Peutz in 1921, except for nasal polyposis (a non-diagnostic feature) (2).

We emphasize the extensive phenotypic expression in our patient, and the scarce one in her brother. This is due to a different penetrance of the STK-11 gene mutation, the presence of de novo mutations, and genetic mosaicism (3).

Due to their high risk of intestinal (small-bowel, colon, pancreas, stomach) and extraintestinal (breast, ovary, uterus, testicle) carcinomas these patients should be closely monitored during follow-up.

REFERENCES