Peutz-Jeghers syndrome: capsule endoscopy to stage disease

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A 13-year-old girl with recurrent abdominal pain and rectal bleeding was referred to our department. Her 36-year-old father had been diagnosed with Peutz-Jeghers syndrome (PJS) after previous surgical interventions for intestinal occlusion. On physical examination, she had hyperpigmented macules on her lips and buccal mucosa. Genetic testing showed a mutation in exon 6 of the gene STK11. A video capsule endoscopy was done to stage the disease. A 7 mm polyp in the stomach (figure A; video 1), and a 12 mm pedunculated polyp in the proximal jejunum, were seen (figure B; video 2). Smaller polyps were found sparsely in the jejunum and proximal ileum. Screening and surveillance is crucial in Peutz-Jeghers syndrome because of the risk of complications related to polyps, and the association with cancers. Our patient’s case shows the feasibility, security, and sensitivity of use of capsule endoscopy to stage disease.

Figure: Peutz-Jeghers syndrome
Video capsule endoscopy showing (A) pedunculated polyp in the stomach; and (B) pedunculated polyp in the proximal jejunum.