Peutz-Jeghers syndrome complicated with intussusception in late pregnancy

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A 19-year-old woman who was 34 weeks pregnant presented at The Affiliated Hospital of Southwest Medical University (Luzhou, China) in September, 2018, with a 3-day history of abdominal pain, accompanied by vomiting and constipation. Physical examination revealed hyperpigmented macules on her lips (figure, A), and an abdominal bulge, abdominal tenderness, and hyperactive bowel sounds. A non-contrast CT scan indicated the presence of intussusception (figure, B) and intrauterine late pregnancy. The patient reported that her mother had undergone bowel resection twice because of intussusception and died of intestinal cancer.

Emergency caesarean and exploratory laparotomy were done. Jejunum intussusception and multiple intestinal polyps were confirmed during surgery. Partial resection and end-to-side anastomosis of the small intestine were done. Pathological evaluation of the resected segment revealed multiple intestinal hamartomatous polyps.

Based on the hyperpigmentation on the lips, multiple intestinal hamartomatous polyps, and family history, a diagnosis of Peutz–Jeghers syndrome was confirmed. The patient recovered well after the initial surgery. 3 months later, she underwent digestive endoscopic polypectomy, with resection of about 20 bigger polyps. She was recommended to have a digestive endoscopy every year, and was advised that she might need further endoscopic polypectomy if necessary. Her premature baby survived delivery at 34 weeks and there was no mucocutaneous hyperpigmentation on the baby's body. However, because Peutz–Jeghers syndrome is an autosomal dominant genetic disease and the incidence in offspring is 50% if one of the parents is a symptomatic heterozygote, the unborn baby should undergo genetic testing for the condition in weeks 18–22 of pregnancy.

Peutz–Jeghers syndrome is a tumour susceptibility syndrome that is related to many malignant tumours, including cancers of the digestive tract, breast, and reproductive system, and can easily lead to acute intussusception. Early diagnosis, staging, and treatment, and regular follow-up are important. In patients with mucocutaneous hyperpigmentation who are suspected of having the condition, full digestive tract radiography or digestive endoscopy should be done, followed by endoscopic treatment if the presence of multiple polyps in the digestive tract is confirmed.

Contributors
All authors cared for the patient. ZL and YZ did the operation. ZL and MS acquired the figures and drafted the original manuscript. ZL and HJ devised the original idea for the report. All authors critically reviewed the article. Written informed consent to publication was obtained.

Declaration of interests
We declare no competing interests.

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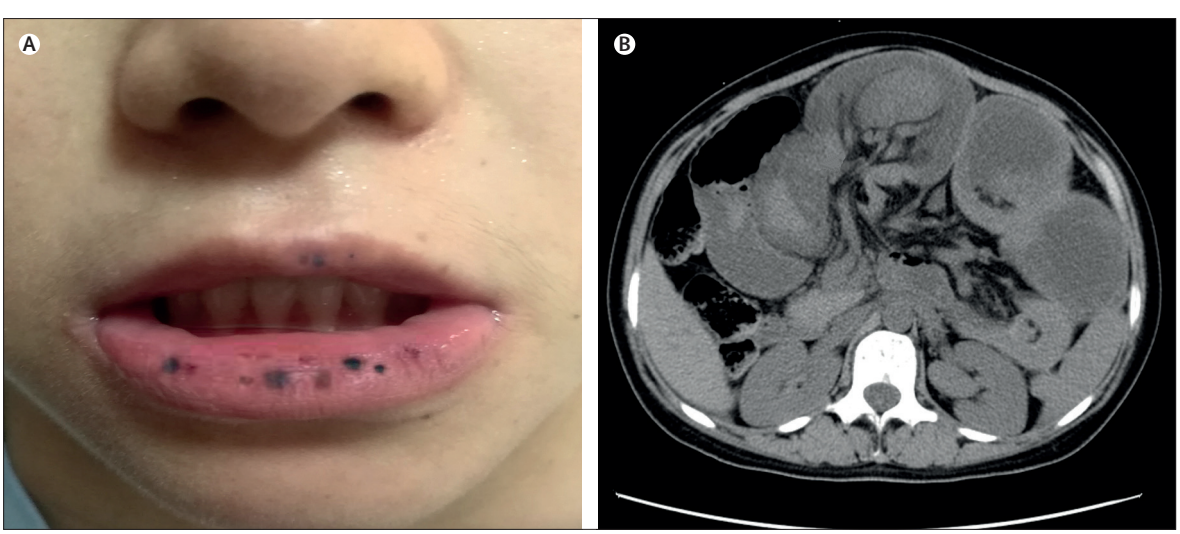

Figure: **Peutz-Jeghers syndrome complicated with intussusception in late pregnancy**
(A) Hyperpigmented macules on the patient's lips. (B) Cross-sectional CT scan showing intussusception.