Colo-colonic intussusception as the first manifestation of familial adenomatous polyposis – index case

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DESCRIPTION
A 19-year-old man presented at the emergency department with asthenia and anorexia for 2 weeks, associated with nonspecific abdominal pain. The patient reported diarrhoea for the last 6 days, without blood neither mucus. The physical examination showed signs of dehydration (dry mucous membranes) and the abdomen was soft and depressible, with diffuse discomfort on palpation. A slightly granular texture was noted on digital rectal examination.

The abdominal ultrasound showed a hypoechoic mass in the right hypochondrium, corresponding to a colo-colonic intussusception on CT, associated with multiple polyps throughout the whole colon (figure 1A–D). These polyps were confirmed by colonoscopy performed 1.5 hours after the CT, showing a larger polyp located in the ascending colon (hepatic angle), corresponding to the location of the intussusception detected previously, that was no longer present. The pathological study of two polyps diagnosed tubulovillous adenomas and the genetic study revealed a mutation in the APC gene, compatible with Familial Adenomatous Polyposis (FAP). The patient has undergone prophylactic proctocolectomy and the surgical specimen showed multiple adenomas throughout the colon, with millimetre dimensions. A larger polyp with 30 mm was confirmed in the ascending colon, which had low-grade dysplasia (figure 2A–D).

FAP is an inherited autosomal dominant disease, caused by germline mutations of the APC gene (chromosome 5q21), corresponding to the development of hundreds to thousands of adenomas throughout the colon and rectum. It occurs with a frequency of 1 in 10 000 individuals.1–3

The polyps appear at the second decade of life, with a mean age of 15 years.4 If the patients are left untreated, they almost inevitably develop colorectal cancer (CRC) by the age of 35–40 years.4

This case report describes a patient with FAP, with a spontaneous mutation – index case – associated with the occurrence of a colo-colonic intussusception. To the best of our knowledge, there are no previously published reports addressing the association between FAP and colo-colonic intussusception.

Intussusception is the invagination of a bowel loop with its mesenteric fold (intussusceptum) into the lumen of a contiguous portion of bowel (intussusciens). Adult intussusception is uncommon and the majority of the cases result from changes...
in the normal peristalsis due an underlying lesion that acts as a lead point.5 6

The adenomas in FAP are almost all less than 5 mm in diameter and less than 1% of polyps are larger than 10 mm, likely being too small to become a leading point of an intussusception.7 This raises the curiosity for the present case, because a polyp with an uncommon size (30 mm) was disclosed in the same location of the intussusception detected at CT (figure 2B), suggesting to be the leading point of the transient intussusception.

The CT imaging findings are virtually pathognomonic for diagnosing intussusceptions in adults, corresponding to the presence of an outer intussuscipiens and central intussusceptum, with identification of the distinct anatomic features (figure 1A).8 9

The patient will undergo a surveillance programme by endoscopic studies, addressing the risk of other gastrointestinal tract tumours, and a periodic examination of the ileostomy.2 3

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REFERENCES

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