Mucinous colorectal carcinoma in paediatric age: an unusual diagnostic

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DESCRIPTION

A 17-year-old teenager, previously healthy, with a history of a ferropenic anaemia for about 1 year, medicated with oral iron. She reports colic-like abdominal pain located in the right quadrants, starting 2 months ago, worse on decubitus. She denied other gastrointestinal symptoms, such as constipation, diarrhoea or melenas. On physical examination, a mass was palpated on the right flank, with pain on deep palpation and tenderness. Laboratory findings included normocytic normochromic anaemia (haemoglobin 10.6 g/L), increased C reactive protein (7.85 mg/dL) and sedimentation rate (60 mm/hour), with negative tumour markers, including carcinoembryonic antigen. Abdominal ultrasound revealed an ascending colon with irregular concentric parietal thickening along a longitudinal extension of 8 cm (figure 1). Abdominal and pelvic CT scan confirmed these findings and revealed bulky regional adenopathies and peritoneal implants (figure 1). She was referred to the paediatric oncology department of our institution.

Colonoscopy showed unspecific findings. An ultrasound-guided biopsy was performed and pathological anatomy revealed mucinous adenocarcinoma of colorectal phenotype.

After a multidisciplinary therapeutic decision meeting, the surgical resection was proposed and performed 10 days after being referred to our medical centre. At surgery, the cancer was localised to the ascendent colon, with bulky mesenteric adenopathies and multiple foci of carcinomatosis dispersed throughout the greater omentum. Right hemicolectomy, peritonectomy and excision of

the omentum were performed. Tumour histology showed a high-grade stage IV mucinous adenocarcinoma, with infiltration of the peritoneal surface, lymphovascular and perineural invasion—pT4a pN1b pM1c (figure 2). Adjuvant chemotherapy was proposed, which the patient has already started.

Germline genetic testing revealed pathogenic alteration in *PMS2* gene, including c.137G>T (p.Ser46Ile) and exon 14 deletion (c.2276-?_2445+?del), establishing the diagnosis of constitutional mismatch repair deficiency syndrome (CMMRD). CMMRD is a rare autosomal recessive syndrome, caused by biallelic mutations in mismatch repair genes (*MSH2*, *MLH1*, *MSH6* and *PMS2*), contributing for the accumulation of DNA mutations, that significantly increases the risk of developing cancer in children and young adults, including colorectal carcinoma (CRC).¹ The family history of cancer was obtained, showing three second-degree paternal relatives with colorectal cancer.

Mucinous CRC in paediatric age is a rare and unexpected diagnostic.

This subtype of adenocarcinoma is characterised by abundant mucinous components that comprise at least 50% of the tumour volume, differing in terms of clinical and histopathological features from other adenocarcinoma.² It is more frequently located in the proximal colon and is the most common form of paediatric CRC. The majority has a sporadic form of the disease, usually present with non-specific signs and symptoms, often lasting 3 months until diagnosis.^{3–6} This greatly contributes to delayed diagnosis, with reports estimating that

Figure 1 (A, B, E and F) CT images in portal phase. A (axial image), B (sagittal image) and F (coronal image) show exuberant thickening of the ascending colon (arrow) and locoregional adenopathies, the largest (red arrow head) being hypodense and with punctate calcifications. E (axial image)—shows peritoneal implant (yellow asterisk) adjacent to the right hepatic border. (C and D) Ultrasound images show (C) diffuse and irregular thickening of the ascending colon with internal vascularisation and (D) hypoechogenic locoregional adenopathy with punctiform calcifications.

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Figure 2 (A) Right hemicolectomy specimen, showing ulcerovegetative lesion. (B) (H&E $\times 20$) malignant epithelial lesion composed of extensive areas of mucin production; grouped cellular elements and some cells with a 'signet ring' appearance.

60%–80% of children and adolescents are diagnosed in stages 3 and 4, leading to a poor prognosis.⁵

Therefore, an effort must be made to alert health providers to this malignancy in children, increasing the rate of suspicion even through a non-specific clinic.

Colonoscopy is frequently recommended during investigation of CRC. However, just as in this case report, the biopsy can also be guided by other imaging modalities, such as ultrasound or CT.

The mainstay of treatment is surgery, including complete tumour resection, its lymphatic bed, and any other involved organs, which provides the greatest impact on the overall survival of the patients.^{7,8}

The role of adjuvant chemotherapy and radiotherapy in children and adolescents has not been established but is recommended in advanced stages.⁸

Prognosis of the disease is influenced by factors such as aggressive histological subtypes, mucinous adenocarcinoma, advanced tumour grade and advanced stage of the disease.^{8,9}

Learning points

- Mucinous colorectal carcinoma (CRC) is the most common histological type of CRC in the paediatric population with a known poor prognosis.
- It is a rare and unexpected diagnostic, which greatly contributes to delayed diagnosis, frequently with dissemination.
- It is important to increase the index of suspicion among doctors for an earlier diagnosis, which is the most important favourable prognostic factor.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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