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CASE REPORT

Desmosis coli as an extremely rare cause of constipation in children – case report and mini-review

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ABSTRACT

Desmosis coli (DC) is an extremely rare condition of unknown pathogenesis. Only a few cases have been reported in the literature. It is characterised by the absence of connective tissue between muscle layers in the colon. Chronic constipation is the main manifestation of DC; thus, it largely resembles Hirschsprung disease. Moreover, neither DC nor Hirschsprung disease responds to conventional treatment. This paper describes a case of a patient with a diagnosis of DC. It was written to highlight the importance of proper diagnosis and to raise awareness of this uncommon disorder. A mini-review of the literature on DC is also provided.

KEY WORDS:

constipation, peristalsis, gastroenterology, paediatrics, intestinal motility disorders.

INTRODUCTION

Constipation is a relatively common condition affecting children. Functional constipation, which affects 0.5–32.2% of the paediatric population, is the most frequent type [1, 2]. Rarely, its cause is organic, and amongst the organic causes, Hirschsprung disease (HD) remains the most common [3, 4]. *Desmosis coli* (DC) can manifest very similarly to HD because both diseases lead to chronic constipation in children that does not improve after conventional treatment [3–6]. *Desmosis coli* is an extremely rare condition, and only a few cases have been reported. Moreover, the pathogenesis of DC remains unclear. Difficulties with stool passage cause a lack of connective tissue between the muscle layers of the colon [4–6].

We report a case of a patient whose symptoms could not be managed with conventional treatment. Histopathological examination revealed that DC was the cause of his symptoms. We wrote this work to raise awareness of this uncommon disorder and to provide a mini-review of the literature on DC.

CASE REPORT

A 7-year-old boy was admitted to the Department of Paediatric Gastroenterology and Nutrition with suspicion of abdominal distention and chronic anaemia. On admission to hospital he presented chronic constipation, faecal incontinence, abdominal pain, enlarged abdominal circumference, and decreased body mass (10^{th} percentile). The symptoms had started a year prior to admission. The abdominal pain occurred also during the night. As of 2 days prior to admission, he had been vomiting as well. The physical examination revealed only asthenic physique, pallor of the skin, tachycardia, distended abdomen, and palpable faecal masses. Blood tests showed decreased haemoglobin (8.5 mg/dl), mean cell volume – 68.1 fl, mean cell haemoglobin – 20.8 pg, mean cell

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haemoglobin concentration – 30.6 g/dl, ferritin level – 11.1 ng/ml, transferrin – 403 mg/dl, and total iron binding capacity – 672 μg/dl. C-reactive protein and sedimentation rate were within normal limits. Due to the history of microcytic anaemia and constipation, the serology of celiac disease was performed. Anti-endomysial, anti-tissue transglutaminase, and anti-deamidated gliadin peptide antibodies were negative.

Abdominal X-ray was performed and revealed distention of the colon with air-fluid levels (Figure 1). Ultrasounds revealed dilation of the sigmoid colon. Contrast enema showed extension of the rectum. Hirschsprung disease was ruled out during anorectal manometry – recto-anal inhibitory reflex was elicited, and normal pressure parameters of the anal canal and normal defaecation dynamics were observed.

These findings led to the suspicion of mechanical obstruction presenting as constipation. An enema was performed and macrogols were prescribed. After the normalisation of bowel movements, the patient was discharged home with the recommendation of taking macrogols and iron supplementation until the next appointment in our department.

The patient returned to the hospital 6 months later for a follow-up visit with additional tests. The symptoms of faecal incontinence, constipation, and abdominal pain seemed to be resolved, but enlarged abdominal circumference remained. A diet analysis showed high carbohydrate content in the diet. Dietary changes were proposed, e.g. limitation of sugary products.

More than one year later, he was admitted to our department due to abdominal pain and bleeding from the lower gastrointestinal tract. The blood tests revealed microcytic anaemia. The gut scintigraphy ruled out Meckel's diverticulum. Additionally, an endoscopic examination was performed, but the colonoscopy was nondiagnostic due to residual stool masses despite proper standard bowel cleansing. Therefore, extensive preparation to the subsequent colonoscopy was initiated with high doses of macrogols administered for 4 consecutive days. The examination revealed an ulceration of the rectum caused by the former presence of hard masses of stool. No histopathologic abnormalities were present after evaluation of mucosal samples. Colon transit time was elongated; the test showed significantly dilated colon and rectum (Figure 2).

Given the findings described above, we decided to take a full-thickness colon wall biopsy. Histopathological examination revealed adequate intestinal wall innervation. However, a specific pattern of abnormalities of the connective tissue characteristic of DC was found, and the diagnosis of DC was made. Total colectomy without a pouch was performed successfully. During follow-up visits, normal bowel movements are observed. Presently there are no symptoms of faecal incontinence, and the abdominal pain has resolved.

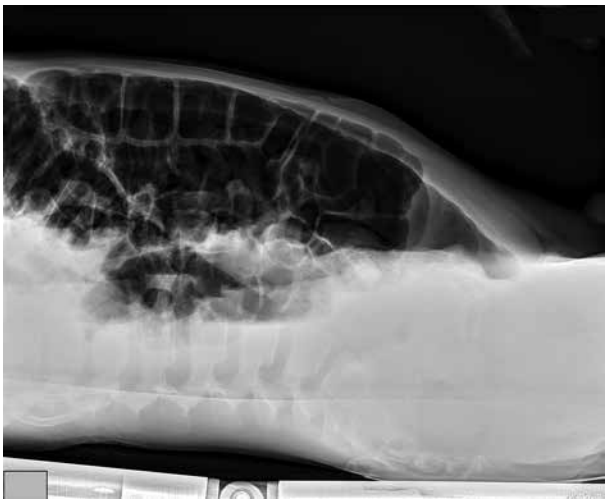

FIGURE 1. Abdominal X-ray showing distention of the colon with air-fluid levels

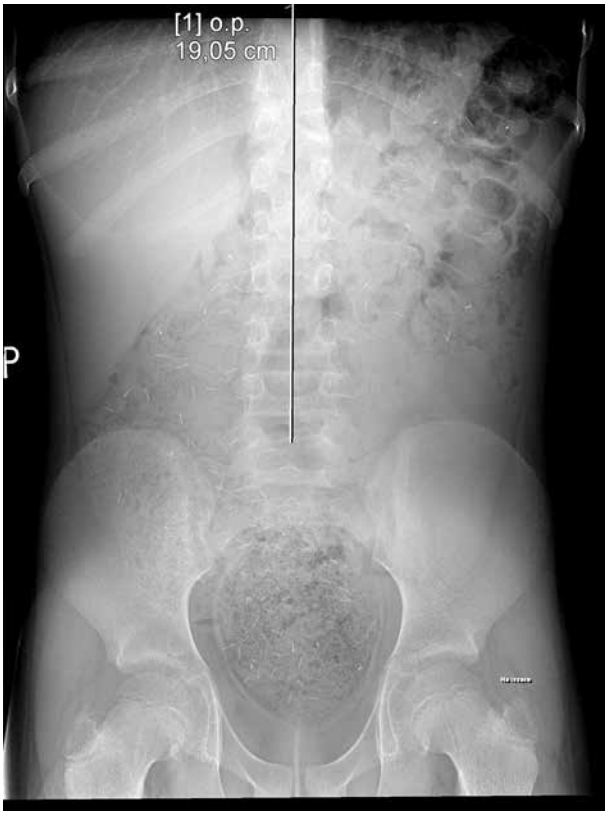

FIGURE 2. Colon transit time test showing significantly dilated colon and rectum

DISCUSSION

Desmosis coli is a congenital disorder that is diagnosed in children. The term was first used by Meier-Ruge in 1998 to describe an extremely rare condition in the gastrointestinal tract [7]. *Desmosis coli* is characterised by the lack of a connective tissue (collagen) layer between the circular and longitudinal muscle fibres of the colon. Unlike in HD, myenteric and submucosal ganglion cells are always present, although their number can be dimin-

ished. The final diagnosis of DC can be made based on histopathological findings [4–7].

Very little is known about the aetiology of DC. *RET* gene mutation is suspected to play a role in the pathogenesis of the disease, but there is not enough evidence to confirm that alterations in a specific gene or any combination of genes may lead to this disorder [6].

The incidence of the disease is also unknown, and few cases have been reported in the literature. The youngest patient whose case was reported in the literature was a child of 14 months and the oldest was 17 years of age. No predominance of any sex was observed [4, 5].

Two distinct subtypes of DC have been reported in the literature: congenital primary type and secondary adulthood onset. The first is known as aplastic desmosis and the second, more common subtype is called atrophic desmosis. Atrophic desmosis can be secondary to Crohn's disease or necrotising enterocolitis [4].

Desmosis coli manifests as chronic constipation that does not improve after conventional therapy. It usually appears in early childhood and can worsen over time. It depends on the severity of constipation at the time when the patient starts searching for medical help. The diagnostic process begins earlier when DC leads to malabsorption and results in low body weight. Distention of the colon and thus increased abdominal circumference are usually observed from the very beginning. At school age faecal incontinence becomes a social problem, and therefore the diagnosis is sought. The lack of collagen network may cause dysfunction of the propulsive activity of the colon, which can even lead to aperistalsis. This occurs because the connective tissue plays an important role in coordinating contractions of intestinal muscle fibres. Cases reported in the literature show that after few years of chronic constipation pseudo-obstruction and perforation can appear. Other non-specific symptoms include vomiting and abdominal pain [4–7].

Differential diagnosis of DC must include HD. The symptoms of both diseases are very similar, and the only way to distinguish between the 2 conditions is full-thickness biopsy with a proper histopathological evaluation, preferably by an experienced pathologist. Calretinin is a marker that can be helpful in differentiating between these disorders, because it is positive in biopsies from patients with ganglion cells. Moreover, a specific staining method called picrosirius red is necessary to mark the presence of connective tissue, which is negative in DC [3, 8–10].

Other conditions that must be ruled out include allied disorders of Hirschsprung disease, which clinically resemble HD. They are compared in Table 1.

Finally, other conditions that can cause similar symptoms include functional constipation (the most common type of constipation overall), dietary factors, hypocalcaemia, anorectal malformations, visceral myopathy, neural tube defects, inflammatory causes (ulcerative colitis,

TABLE 1. Comparison of gastrointestinal motility disorders

Disorder	Histopathological features	Marker
Hirschsprung disease	The absence of ganglion cells in the myenteric and submucosal plexuses of the intestine [10]	Calretinin (negative), increased AChE activity
Desmosis coli	Lack of connective tissue layer between the circular and longitudinal muscle fibres of the colon [4]	Picrosirius red staining (negative)
Intestinal neuronal dysplasia type A	Hypoplasia or aplasia of the adrenergic enteric nervous system [11]	Increased AChE activity
Intestinal neuronal dysplasia type B	Hyperplasia of the parasympathetic submucosal plexuses (hyperganglionosis, giant ganglia, ectopic ganglion cells) [12]	Increased AChE activity
Immaturity of ganglia	Immaturity of the submucous and myenteric plexus [13]	Negative or weak positive SDH reaction
Hypoganglionosis	Decreased number of nerve cells (to approximately 60%) in the bowel wall [10]	Increased AChE activity
Megacystis microcolon intestinal hypoperistalsis syndrome	Normal ganglion cells, thin longitudinal muscle, reduction in α-smooth muscle actin, proliferation of connective tissue [14]	–
Internal anal sphincter achalasia	Normal ganglion cells in rectal biopsy, absence of ganglia in anal canal [15]	–
Paediatric intestinal pseudo-obstruction	Small intestinal involvement: myopathy and/or neuropathy and/or mesenchymopathy (abnormal ICC development) [3]	–

AChE – acetylcholine esterase, ICC – interstitial cells of Cajal, SDH – succinate dehydrogenase

Crohn's disease), celiac disease, infection (e.g. *Clostridioides difficile*), mutations (e.g. *ACTG2* mutation), and adynamic bowel syndrome [4].

Management of DC is a challenge. Pharmacotherapy with laxatives, enemas, and colostomies are all ineffective. Even when using maximum doses of drugs, neither the symptoms nor the signs, such as dilated colon, can be improved. The only treatment that might resolve the problem is surgical excision of the large intestine and performing an ileostomy. The extent of the surgery depends on the individual case [2–6]. Unfortunately, no information about the patients' follow-up can be found in the literature; hence, little is known about patients' outcomes after surgery.

CONCLUSIONS

We report our case to broaden the knowledge of organic causes of chronic constipation by describing one of the rarest: DC. We aim to raise awareness of this disorder and prompt clinicians to take it into account. *Desmosis coli* should be considered as a possible diagnosis in children with chronic constipation refractory to conventional treatment, especially with symptoms of abdominal distention, abdominal pain, incorrect body weight gain, and dilated colon despite intensive laxative treatment.

DISCLOSURE

The authors declare no conflict of interest.

REFERENCES

1. Steutel NF, Zeevenhooven J, Scarpato E, et al. Prevalence of functional gastrointestinal disorders in European infants and toddlers. J Pediatr 2020; 221: 107-114.
2. Dybowska A, Kupniewska P, Sobacka A, Kroczek K, Krogulska A, Gałązka P. A child with chronic functional constipation – a challenge for a pediatrician alone or a surgeon as well? Case seriesand literature review. Pediatr Pol 2022; 97: 125-132.
3. Chanpong A, Borrelli O, Thapar N. Hirschsprung disease and paediatric intestinal pseudo-obstruction. Best Pract Res Clin Gastroenterol 2022; 56-57: 101765.
4. Desale J, Shah H, Kumbhar V, Sandlas G. Desmosis coli – a case report and review of the literature. Dev Period Med 2017; 21: 390-392.
5. Hübner U, Meier-Ruge W, Halsband H. Four cases of Desmosis coli: severe chronic constipation, massive dilatation of the colon, and hypoperistalsis due to of changes in the colonic connective-tissue net. Pediatr Surg Int 2002; 18: 198-203.
6. Marshall DG, Meier-Ruge WA, Chakravarti A, Langer JC. Chronic constipation due to Hirschsprung's disease and Desmosis coli in a family. Pediatr Surg Int 2002; 18: 110-114.
7. Meier-Ruge WA. Desmosis of the colon: a working hypothesis of primary chronic constipation. Eur J Pediatr Surg 1998; 8: 299-303.
8. Szymańska S, Markiewicz-Kijewska M, Pyzlak M, et al. Pathogenesis of chronic constipation in a Polish group of paediatric patients – an attempt to create the optimal histopathological diagnostic protocol. Prz Gastroenterol 2019; 14: 109-111.
9. Thapar N, Saliakellis E, Benninga MA, et al. Paediatric intestinal pseudo-obstruction: evidence and consensus-based recommendations from an ESPGHAN-Led Expert Group. J Pediatr Gastroenterol Nutr 2018; 66: 991-1019.
10. Gfroerer S, Rolle U. Pediatric intestinal motility disorders. World J Gastroenterol 2015; 21: 9683-9687.
11. Rajalakshmi T, Makhija P, Babu MK, Kini U. Intestinal neuronal dysplasia type A. Indian J Pediatr 2003; 70: 839-841.
12. Toledo de Arruda Lourenção PL, Terra SA, Ortolan EVP, Rodrigues MAM. Intestinal neuronal dysplasia type B: a still little known diagnosis for organic causes of intestinal chronic constipation. World J Gastrointest Pharmacol Ther 2016; 7: 397-405.
13. Nogueira A, Campos M, Soares-Oliveira M, et al. Histochemical and immunohistochemical study of the intrinsic innervation in colonic dysganglionosis. Pediatr Surg Int 2001; 17: 144-151.
14. Wymer KM, Anderson BB, Wilkens AA, Gundeti MS. Megacystis microcolon intestinal hypoperistalsis syndrome: case series and updated review of the literature with an emphasis on urologic management. J Pediatr Surg 2016; 51: 1565-1573.
15. Obata S, Fukahori S, Yagi M, et al. Internal anal sphincter achalasia: data from a nationwide survey of allied disorders of Hirschsprung's disease in Japan. Surg Today 2017; 47: 1429-1433.