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Familiar Occurrence of Desmosis of the Colon – Report of Two Cases and Literature Review

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Abstract

Gastrointestinal motility disorders are relatively common in children and adolescents. The diagnosis is often difficult and time-consuming as the etiology of the problems includes different pathological entities such as enteric neuropathies, myopathies and collagenopathies. Hirschsprung's disease (HD) is one of the most common and best recognized disorders, particularly in infants. Whereas, desmosis of the colon is a rare condition characterized by total or focal lack of connective-tissue net of the circular and longitudinal muscles without any abnormalities within enteric nervous system (ENS). Its diagnosis is based on full-thickness intestinal biopsy or post-operative material followed by histological staining with pricrosirius red. We present two adolescent girls who were familiar related (the same mother, different fathers) and both of them suffered from chronic constipations. HD was excluded in these cases and surgical removal of impaired part of intestines was performed. Material from post-operative specimens proved desmosis of the colon in each patient.

Keywords: Gastrointestinal motility disorders; Hirschsprung's disease; Desmosis of the colon; Biopsy

Introduction

Gastrointestinal motility disorders are relatively common in children and adolescents. Many diseases such as enteric neuropathies, myopathies and collagenopathies, may cause constipation and abdominal pain in the pediatric population. Therefore diagnosis is usually difficult and time-consuming. Rare but serious cause of chronic constipation is desmosis of the colon. The condition was first described by Meier-Ruge [1]. He presented 14 patients suffering from chronic constipation, hypoperistalsis and dilation of the colon. In his group, histopathology of the resected bowels revealed complete or incomplete lack of the mesh network of collagen [2,3]. This entity may be congenital or acquired. Congenital form of desmosis is called aplastic and it occurred mostly in premature infants with low birth weight [4]. Acquired type, also called atrophic, is more common and its mechanism is related to pathologic inflammation of muscularis propria due to Crohn disease or diverticulitis for example. Enzymes such as collagenases are released and they destroy the connective tissue of the bowel wall [4]. The disease may be found both in children and adults. Two forms of atrophic desmosis were described – complete, with involvement of the taenia and incomplete, without involvement of the taenia [4]. Diagnosis of chronic constipation consists of radiologic examination, anorectal manometry and rectal biopsy. The last method is believed to be the current international diagnostic gold standard [3,4]. The exact etiology of pseudo-obstructions has not been identified yet. This disorder is defined as a clinical syndrome caused by severe impairment in the ability of the intestines to push food through. It is characterized by the clinical signs and symptoms without any lesion in the intestinal lumen. We present two adolescent girls who were related (the same mother, different fathers) and both of them suffered from chronic constipation. Hirschsprung's disease (HD) was excluded in these cases and surgical removal of impaired part of intestines was performed. Material from post-operative specimens revealed desmosis of the colon in each patient. This is an assumption that specific genetic background of the entity may exist.

Case 1

In 2006, 12 year old girl was admitted to The Children's Memorial Health Institute. She presented with chronic constipation that started 10 years ahead after appendectomy performed due to acute appendicitis. In 1999 she was operated on because of abdominal abscess complicated by necrosis of omentum. At admission wide diagnostic approach was introduced. Ultrasound examination was within normal, rectoscopy revealed no abnormalities, however, barium enema examination showed reduced caliber of the rectum suggesting aganglionosis. Therefore, after multidisciplinary consultation (gastroenterologist, surgeon, and radiologist) decision of colectomy was established. During the procedure, intestinal biopsy was obtained to assess the extent of operation. Histopathological examination of diagnostic material (serially cut slides) proved presence of normal ganglion cells. The result was consistent with negative acetylcholine esterase reaction. Left-side hemicolectomy was done. Histopathological examination of post-operative material was within normal with presence of ganglion cells in mysenteric plexus and submucosal plexus (Figure 1).

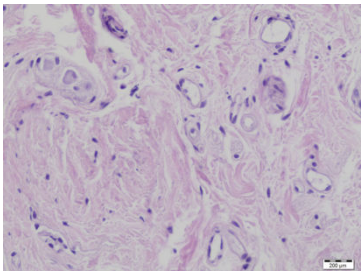

Figure 1: Presence of normal ganglion cells in myenteric plexus, H&E, original magnification X600.

After the surgery, constipation remained thus ileostomy was exteriorized 15 cm after ileocecal valve. The girl was discharged from the hospital in good general condition but final diagnosis was not established than. In 2008 she was re-admitted complaining of abdominal pain and constipation. Suction biopsy was taken but it was within normal as previous one. Total colectomy according to Duhamel-Martin method with the removal of terminal ileum was performed. Several months later she returned to the hospital presenting with burning sensation in her chest and sore throat and regurgitation of food. Gastroesophageal reflux disease (GERD) was suspected; therefore endoscopy of upper alimentary tract was performed. Although the examination revealed no abnormalities, she was treated with proton-pump inhibitor and underwent another ileostomy. We decided to analyze not only recent post-operative material but also retrospectively samples taken from colon and perform additional staining with picrosirius red. It showed complete lack of the mesh network of collagen in the intestinal wall (Figure 2). Clinical symptoms together with histological stain supported the diagnosis of complete desmosis of the colon. The girl is 22 year old now and she has remained in good general condition.

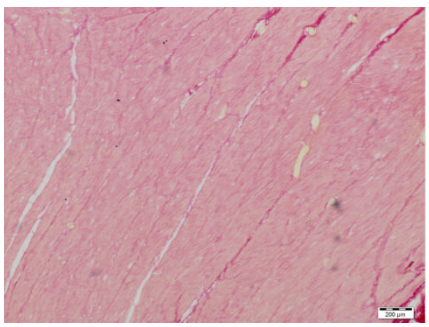

Figure 2: Complete lack of the mesh network of collagen in the intestinal wall, Picrosirius red, original magnification X100.

Case 2

In 2015, 16-years old girl was presented to the Department of Surgery, The Children's Memorial Health Institute, with chronic constipation accompanied by weight loose. Symptoms started 4 years ahead. During hospitalization, Malone stoma was created and intestinal biopsy was taken at the sometime. Histopathological evaluation of full-thickness bowel gut was within normal. Nonetheless, the girl did not tolerate stoma well and it turned out that it was narrowed. In view of her history - stepsister (the same mother, different fathers) was diagnosed with desmosis coli, suction biopsy from caecum was stained with Picrosiriu red proving desmosis of the coli (Figure 3). Total colectomy was surgical treatment of choice. One year later, the girl was re-admitted to our hospital for follow-up visit. She has remained in good general condition, and she put on weight.

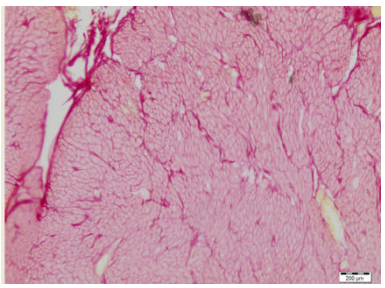

Figure 3: Total lack of connective - tissue net of the muscles in the intestinal wall, Picrosirius red, original magnification X100.

Discussion

Diagnosis and treatment of pseudo-obstructions in children, especially in pre-term infants, is difficult and time-consuming. There are several entities that may cause constipations and abdominal pain including enteric neuropathies, myopathies and collagenopathies [5,6]. Quick establishment of proper diagnosis is mandatory as management of patients depends of etiology of the disorder. In most cases, avoiding surgery is possible. One of very rare but serious causes of constipation is desmosis of the colon. Literature on the subject consists of 6 full-text papers [7,8], the most recent one from 2012. It means that the problem should be better investigated. Therefore, we presented two related girls who were diagnosed with desmosis of the colon. We believe, that these cases are interesting and instructive both for physicians and pathologists, as the patients suffered from chronic constipation with first manifestation in adolescence, the histopathology of their surgical materials revealed complete lack of connective tissue network in the circular and longitudinal muscle layer. Apart from similar clinical course, the patients have the some mother; this may be an assumption that desmosis coli may have specific genetic background. Unfortunately, we could not perform genetic tests, because older sister stayed abroad. Acquired type (atrophic) should also be taken under consideration in these cases. However, it is less probable, as pathologic inflammation due to different diseases was diagnosed in neither of the girls. Hübner et al. [4] described in 2001 four cases of desmosis coli in unrelated patients. His observations were consistent with ours in respect of clinical follow-up. In his group, three out of four patients needed lifelong ileostomy, only one gained nearly normal bowel movements but it was the case of focal desmosis of transverse intestine. Both our patients had finally removed all large intestines. The aim of this paper is also to highlight the most important diagnostic clues that may elude as entity is very rare. It is crucial to remember that it may concern both small and large bowels and colectomy does not guarantee the treatment in all cases. Besides, in pre-term infant's ganglion cells may be immature and difficult to find in light microscopy, therefore, exclusion of aganglionosis should not be rash. The first girl underwent total colectomy but we also had post-operative material (stoma) from her terminal ileum. We retrospectively performed staining with

picrosirius red in samples from small and large bowels. This should be particularly done in symptomatic patients with normal microscopic features of the large intestine. Even though the name suggests only colonic involvement, it can appear also in small intestine or be present in small intestine exclusively. Burns et al. [1] made an attempt to describe the development of enteric nervous system and its role in intestinal motility in fetal and early postnatal stages, but it is still not entirely clear matter and seems to be very individual. Proper microscopic diagnosis of desmosis coli requires full-thickness intestinal biopsy with subsequent picrosirius red stain. It is recommended to stain samples from small and large bowels as the disorder may be focal and complete and incomplete forms exist. It seems that the later present with milder clinical course and has better prognosis. The final diagnosis is based both on clinical and histopathological characteristics. We would like to introduce diagnostic algorithm of pseudo-obstructions in our hospital. Suction colonic biopsy with ACHE reaction should be the first step. If the result is normal, full-thickness intestinal biopsy with picrosirius red staining should be performed. If it also reveals no abnormalities, additional IHC reactions toward other possible disorders are necessary. Presented cases support this kind of approach. Nonetheless, further studies on the subject with larger groups of patient, both pediatric and adults are needed. Primary future goal is to better understand the etiology of the disease with special focus on genetic aspect.

Conclusion

Desmosis of the colon is a rare entity. One should take it under consideration in patients with constipation and presence of normal ganglion cells in intestinal wall. Full-thickness biopsy is mandatory for proper diagnosis.

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