Hirschsprung's Disease As a Cause of Severe Iron Deficiency Anemia in an Adult: Case Report and Review of the Literature

CİDDİ DEMİR EKSİKLİĞİ ANEMİSİNE NEDEN OLAN ERİŞKİN HİRSCHSPRUNG HASTALIĞI: VAKA TAKDİMİ VE LİTERATÜRÜN GÖZDEN GEÇİRİLMESİ

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Summary

Objective: Hirschsprung's disease is a rare congenital disorder in adulthood. Symptoms in adult life include constipation, anorexia, vomiting, weight loss, and tiredness. Hirschsprung's disease usually results in long term morbidity and shortened life span. Persistent anemia is a very rare symptom in adults Hirschsprung's disease. We aimed to present a case of iron deficiency anemia in a patient secondary to adult Hirschsprung's disease.

Case Report: A 19-year old female presented with the symptoms of Hirschsprung's disease that emerged late in adult life with severe iron deficiency anemia.

Conclusions: The diagnosis of adult Hirschsprung's disease should be suspected in patients with a history of chronic constipation and persistent anemia. We describe the findings detected by computed tomography (CT) and magnetic resonance (MR), but these findings are not specific and rectal biopsy is required for definitive diagnosis.

KeyWords: Hirschsprung's disease, Constipation, Anemia

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Özet

Amaç: Hirschsprung hastalığı erişkinlerde nadir olarak görülür.Konstipasyon , bulantı, kusma, kilo kaybı ve halsizlik semptomları olabilir. Hirschsprung hastalığı uzun dönemde hayat süresini kısaltır. Bu hastalarda dirençli anemi nadiren görülür. Bu olguda demir eksikliği anemisinin nadir bir nedeni olan adult Hirschsprung hastalığını sunmayı amaçladık.

Olgu Sunumu: Ciddi demir eksikliği anemisi olan, 19 yaşında erişkin Hirschsprung hastası takdim edildi.

Sonuçlar: Kronik konstipasyon ve demir eksikliği anemisi saptanan hastalarda adult Hirschsprung hastalığından şüphe edilmelidir. Tomografi ve manyetik rezonans bulguları bu hastalık için spesifik olmadığından kesin tanı için rektal biopsi gereklidir.

Anahtar Kelimeler: Hirschsprung hastalığı, Konstipasyon, Anemi

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Hirschsprung's disease (HD) is characterized by motor dysfunction of the alimentary tract including the internal anal sphincter and a congenital absence of ganglion cells in the involved bowel segment (1,2). The segment of bowel involved can be variable, from a few centimeters to the entire colon and rectum. In about three- fourths of the cases, involvement is limited to the rectosigmoid colon. The affected segment of the intestine lacks the ability to relax and move bowel contents along.

Some patients with a milder form of Hirschsprung's disease present later in life. These patients often have a short aganglionic segment (3). The disease is rare in adulthood. Symptoms include constipation, abdominal swelling, and vomiting. Anemia is not a specific finding associated with Hirschsprung's disease. To our knowledge, only one case of persistent anemia secondary to Hirschsuprung's disease has been reported. We report a patient with chronic constipation secondary to newly diagnosed short segment Hirschsprung's disease who had been treated for ten years because of iron deficiency anemia.

Case Report

A 19-year old female presented with diffuse abdominal pain, fatigue, palpitations, and life-long history of constipation, passing stools every 1-2 weeks. Patient did not describe pica. She had required hospitalization at the age of 8 for a fecal impaction. She complained of diffuse abdominal pain for five months prior to admission to our hospital. Constipation worsened despite increased laxative use. Physical examination revealed a moderately distended abdomen and pale appearance. There was no tenderness and bowel sounds were hyperactive. Digital examination of the rectum revealed no mass or fecal impaction.

Laboratory evaluation displayed severe hypochromic and microcytic anemia (hemoglobin, 7.9 gr/dl, hematocrit, 26.6%, mean red cell volume, 62.1 fl), WBC, 8200/ mm3 (65% neutrophils, 30% lymphocytes, 3% monocytes, 2% eosinophils, anisocytosis, microcytosis). Reticulocyte count was 9%. Bone marrow examination showed erythroid hyperplasia. Examination of the stool for pus, bacterial and parasitic organisms and for the presence of fat was negative.

The serum iron level was also low at 16 ng/dl as well as ferritin [1.5 (9-20) ng/dl]. Iron-binding capacity was 466 ng/dl. LDH, total biluribin and all other laboratory results were normal. Occult blood in stool was positive. The patient stated to have a regular menstrual cycle. Oesophagogastroduodenoscopy and a barium examination of the small bowel were normal. CT and MR colonography exams were performed on the same day with a standart colonoscopic bowel preparation. For these examinations a liquid enema was used. Enema was prepared by mixing 20 ml gadolinium and 100 ml iodine within 1000 ml saline. Once colonic distention was achieved, CT colonography was performed with 8 mm collimation and a table feed of 8 mm by spiral CT (PQS, Picker, USA). Then MR colonography was performed with 1,5 T MR unit (Edge, Picker, USA). T1-weighted gradient echo (GRE) sequences were used in MR colonography. A narrow rectum with markedly dilated sigmoid colon and a short segment stenosis at the rectosigmoid junction were demonstrated by CT colonography (Figure 1) and MR colonography (Figure 2). Anal manometric study revealed ab-

Figure 1. CT colonography showing typical transitional zone at the rectosigmoid junction (arrow) and the narrow rectum with markedly dilated sigmoid colon.

Figure 2. MR colonography showing a narrow segment, with an enormously dilated segment above.

Figure 3. The biopsy revealed the absence of ganglion cells (hematoxylin and eosin x 40).

absence of the normal anorectal inhibitory reflex and increased rectal tonus. Full-thickness biopsy was obtained under general anesthesia, from recto-sigmoid junction by myectomy. Histopathologically, a full-thickness biopsy of the stenotic bowel disclosed no ganglion cells within the nerve plexus (Figure 3). Colonoscopically, there was mucosal hyperemia and mucosal erosions in the descending and sigmoid colon regions. The patients was treated with cleansing enemas for 3 days and improved. She refused surgery after the diagnosis. Oral iron was administered.

Discussion

Hirschsprung's disease is a congenital disease with an incidence of 1 out of 5000 births. Two thirds of cases are diagnosed in the first three months of life and only a few cases remain undetected after 5 years of age (1,4). Rarely, the patients may be undiagnosed until adulthood. Patients have chronic intractable constipation requiring enemas, cathartics, and may give a history of requiring digital evacuation. Fecal incontinence is not a feature of the adult patients. A familial tendency has been reported. The rectal ampulla will frequently be empty with stool felt high in the rectum (5,6). The delay in diagnosis of the present case was due to failure to recognize the symptoms of Hirschsprung's disease.

The aganglionic segment is the result of failure of migration of ganglion cell precursors from the neural crest in to the hindgut. The involved area is not innervated by non-adrenergic non-cholinergic excitatory or inhibitory nerves, and does not relax, and eventually, the colon proximal to the aganglionic segment dilates (4,7,8). In typical Hirschsprung's disease, the aganglionosis starts from the anal valve, spreads proximally. As reported in our case, in less than 10% of the patients, aganglionosis may occur more proximally and in some cases ascending colon may be involved (3). Hirschsprung's disease should be established on well prepared permanent sections (9). Forty-seven biopsy- proved adult HD cases at ages between 18-74 (mean age 35 years) were reported in the literature and the male-to-female ratio was 4:1 (7). The barium enema is frequently the initial diagnostic procedure where in 80% of the cases, a cone-shaped transitional zone leading into a narrow segment was seen (7,10). The plain film of the abdomen demonstrates large amounts of faeces within a grossly dilated colon. Adequate bowel preparation may be impossible and unnecessary. Since barium retention could occur, we used alternative diagnostic procedure, such as MR and CT colonographies.

CT colonography and MR colonography without the use of sedation, intervention or compression are well tolerated by patients compared with other full colonic examinations such as barium enema radiography or conventional colonoscopy. The potential for fecal tagging and the elimination of anerous bowel cleansing regimens, as well as extension to a multiorgan tumor screening, may further improve the acceptance of colonography (11,12).

The diagnosis must be carefully confirmed in adults in order to differentiate the condition from idiopathic megacolon (13). As was in our case, radiological findings, a life time history of refractory constipation, anal manometry and full-thickness biopsy should all confirm the diagnosis. Powell reported a 14- year-old patient with HD and persistent anemia for 6 months (10). The anemia was due to ischemia caused by compromise of the vasculature by colonic distention, superficial inflammation and ulcerations of the mucosa by fecalomas.

In conclusion, Hirschsprung's disease should be suspected in adults with abnormal bowel habits, especially a history of constipation dating back to infancy and unexplained iron deficiency anemia.The images obtained by CT and MR colonography provide diagnostic criteria, albeit not specific, and anal manometric study with histopathological analysis are required for exact diagnosis.

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