Adult Hirschsprung's Disease

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Abstract Hirschsprung’s disease is rare in adults. A constant symptom of the disease is chronic refractory constipation. Barium enema showing a distal narrow segment with a cone-shaped transition zone to proximal dilatation suggests the diagnosis; ano-rectal manometry may be helpful if the anorectal inhibitory reflex is absent. The diagnosis is confirmed by the absence of ganglion cells from the rectal wall biopsy. The therapy is surgical. Combined transabdominal-transanlal procedures are the most commonly used therapeutic modalities. The case of a 34-year-old man with chronic constipation since childhood is presented. His bowel motion was once every 3-4 weeks. The diagnosis was suspected on history and finding on barium enema, and confirmed by histopathological examination. A modified Duhamel’s procedure led to a successful outcome.

Keywords: Chronic refractory constipation, Saudi Arabia

Hirschsprung’s disease (HD) is a motility disorder of the intestine, presenting most often with chronic constipation. The pathologic substrate underlying the disease is the absence of ganglion cells in the submucosa and myenteric plexuses of the rectum and distal colon, which may extend proximally as far as the esophagus. The diagnosis of this condition is usually made in infancy or early childhood. In rare instances, the disease is first recognized in adolescence or adulthood. We report the first case of adult Hirschsprung’s disease (ADH) in the Saudi literature. Clinical features, diagnostic approach, treatment and outcome are described.

Case Report A 34-year-old Saudi man was admitted to the medical ward with history of chronic constipation since early childhood, which did not respond to a variety of laxatives. He was passing stools once every 3 to 4 weeks and required increasingly frequent enemas, and at times hospital admissions to induce bowel movements. Spontaneous evacuations were preceded by abdominal cramps and colic’s which would resolve shortly after defecation. On admission the abdomen was mildly distended but soft. An impassible mass occupying almost the entire abdomen was palpated. Peristaltic sounds were normal. On rectal examination the ampulla was narrow and empty. Complete blood count, blood urea nitrogen, serum creatinine and serum electrolyte, urine analysis, liver function tests as well as chest X-ray and electrocardiogram were normal. Plain abdominal X-ray showed radiographic signs of massive fecal impaction. “Unprepped” barium enema demonstrated hugely dilated colon with narrow segment distal to the rectosigmoid junction (Fig. 1); a transition zone appeared, first after preparation of the bowel prior to repeated barium enema (Fig. 2). The evacuation of the contrast material was markedly delayed.

Following the preparation by laxatives, repeated enemas and rectal washouts, the patient was subjected to a full-thickness rectal biopsy one centimetre above the dentate line. The histology revealed absence of ganglion cells. Three weeks later, a transverse loop colostomy was fashioned, which was complicated by distal stone prolapse six weeks after the operation necessitating resection.

A modified Duhamel’s pull-through procedure, using an Autosture device, was performed 2 months after the resection, and colostomy closed after another 3 months with complete recovery. Postoperative barium enema showed normal caliber and hausturation of the entire colon with slight delay in the evacuation of contrast material (Fig. 3). The patient doesn’t have difficulty with bowel function anymore, evacuating once every second day, with normal continence for feces and

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Fig. 1 - Unprepped barium enema disclosing a distal narrow segment with proximal dilatation of the colon, but without cone-shaped transition.

Fig. 3 - Barium enema demonstrating normal caliber of the colon following decompression, and modified Duhamel's procedure.

flatus. Occasionally he needs laxatives. The patient has been under regular follow-up in the outpatient clinic for the last two years. He has gained 11 kg. in weight and is enjoying a better quality of life.

Discussion Adult Hirschsprung's disease is an uncommon disorder, as it is usually diagnosed in infancy or early childhood. However, it must be considered in all cases of chronic refractory constipation associated with megacolon. Colicky abdominal pain, recurrent episodes of diarrhea, vomiting, anorexia, weight loss, fetid odor ex ore, hypochromic anemia and explosive borborygms are other symptoms of the disease. Characteristically, the history of constipation can be traced back to infancy or early childhood. However, a few cases with first onset of symptoms in adulthood have been reported.

Physical examination usually shows abdominal distention, that may be extreme, with stretched skin and perhaps visible peristalsis, or mild as in our case. Palpation may reveal an abdominal mass with doughy consistency compatible with feces-filled colon. Rectal examination frequently reveals empty and narrow ampulla with good sphincter tone, as in our patient, but fecal
Impaction is not uncommon.\textsuperscript{1,3} AHD typically remains unrecognized, despite frequent medical consultations and hospitalizations.\textsuperscript{2} We think that clinical awareness of the condition and a high index of suspicion are extremely important in navigating the diagnostic effort in the right direction. Plain abdominal X-ray often demonstrates massive fecal impaction. Barium enema usually shows a narrow rectum with a cone-shaped transition to a markedly dilated proximal colon. This characteristic transition zone is expected only if ‘unprepped’ barium enema is performed.\textsuperscript{3} Contrary to this generally accepted opinion, the transition zone appeared in our case only after bowel preparation (Fig. 2). A normal examination does not rule out the diagnosis; a 20\% false-negative rate has been reported for barium enemas.\textsuperscript{6} Measurement of rectoan inhibitory reflex has been increasingly recommended as a screening test for all patients with chronic constipation.\textsuperscript{7} It may be weak or even absent in other cases of chronic constipation such as idiopathic megarectum\textsuperscript{8} but it is always absent in HD. Its diagnostic specificity in HD is around 90\%.\textsuperscript{6}

Full-thickness rectal biopsy is currently considered the standard diagnostic test for HD. Suction rectal biopsy, commonly used in pediatric ages, is usually insufficient due to the thickness of adult rectal mucosa.\textsuperscript{2} It should be taken well above the anal verge, since ganglion cells are normally absent from the anal canal.\textsuperscript{1} The diagnosis of HD is confirmed by complete absence of ganglion cells. This study also helps classify megacolon into its various subcategories. Ganglion cells can be present but degenerated as in Chagas’ disease, or immature, or present and normal as in megacolon secondary to other organic or psychogenic disorders.\textsuperscript{9} Because of the progressive nature of the disease, surgical treatment is desirable as soon as the diagnosis is confirmed. The surgical management of ADH seems to be controversial however.\textsuperscript{2} Duhamel’s retrorectal pull-through, Soave’s endorectal pull-through and Swanson’s rectosigmoidectomy have been the most commonly performed operations.\textsuperscript{10} Low anterior resection alone and left hemicolectomy or subtotal colectomy are considered inadequate treatment for HD.\textsuperscript{3,11} Posterior rectal myectomy has been suggested as an initial approach to the condition due to its low rate of morbidity and technical ease of performance.\textsuperscript{12} Most recently, the posterior rectal myectomy is performed in suspected cases of low-segment Hirschsprung’s disease. It provides tissue to confirm the diagnosis and may even be therapeutic if the specimen shows ganglion cells in its cephalad part, but absence of ganglion cells from this part does not always necessitate further surgery.\textsuperscript{13}

The rates of major postoperative complications are 10,25 and 33\% after Duhamel’s, Soave’s and Swanson’s procedure respectively.\textsuperscript{2,11} They represent primarily anastomotic disruption leading to abscess and/or fistula formation. A 7\% rate of impotence was reported following Swanson’s procedure. The choice of surgical procedure should depend primarily on the surgeon’s experience rather than on the procedure per se. All three procedures mentioned above would yield equally good results when performed by an experienced surgeon.\textsuperscript{2} Our patient underwent a modified Duhamel’s procedure three months after the creation of a transverse colostomy, as it is the procedure of our choice in the treatment of pediatric Hirschsprung’s disease in this hospital. A preliminary colostomy was fashioned to decompress the hugely distended colon, to protect the anastomosis and to prevent contamination of the pelvis during the pull-through procedure. In fact, an adequate preoperative preparation with a colostomy is regarded as the most important contribution to a successful outcome and not the type of procedure performed.\textsuperscript{14}

References

ملخص

داء هيرشبرنغ نادر يحدث في البالغين. عرضه الشائع إمساك مزمن عصبي. وتبدأ حفنة البالغين قبل الولادة. نظرياً، يشير هذا إلى توسع داعم يحتاج إلى التشخيص. وقياس الضغط البروبيك المستقيم يساعد على التشخيص في حالة غياب المعكمس المبيض. ويؤكد التشخيص بغياب الخلايا العقدية من خزعة جدار المستقيم. والعلاج الجراحى، والعلاجات المجرة خلال البطانة والشرج هي الوسائل العلاجية الأكثر استعمالا. قدمت حالة رجل عمره 34 سنة، وعاني من إمساك مزمن منذ سن الطفولة، وكان يشير مرة واحدة إلى أربعية أسابيع. وقد تم التشخيص من التاريخ المرضي ونتائج حقنة البالغن، وأثبتت عن طريق الفحص التشريحي للمرضى النسيجي. وقد أدت عملية "دوهام المحرّة" إلى شفاء المريض.