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CASE REPORT: RARE FORM OF HIRSCHPRUNG'S DISEASE

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Key words: Hirschsprung disease, total colonic aganglionosis, Down syndrome.

Summary
Hirschsprung disease (HD) is a developmental disorder characterized by the absence of ganglia in the distal colon, resulting in a functional obstruction. Incidence of total colonic aganglionosis (TCA) is 1 in 500 000 and it accounts for 5-10% of all cases of HD. HD should be suspected in patients with typical clinical symptoms and a high index of suspicion is appropriate for infants with a predisposing condition such as Down Syndrome (DS), or for those with a family history of HD. The treatment of choice for HD is surgical, such as Swenson, Soave, and Duhamel procedures. The goals are to resect the affected segment of the colon, bring the normal ganglionic bowel down close to the anus, and preserve internal anal sphincter function. We present a clinical case report of TCA in a child with Down syndrome (DS) and review of literature.

Introduction

Hirschsprung disease (HD) is a developmental disorder characterized by the absence of ganglia in the distal colon, resulting in a functional obstruction [1]. Introduction Total colonic aganglionosis (TCA) is a rare condition in which aganglionosis involves the entire colon. Incidence of TCA is 1 in 500 000 and it accounts for 5-10% of all cases of HD. HD should be considered in any newborn who fails to pass meconium within 24-48 hours of birth [2]. Contrast enema is useful in establishing the diagnosis, but full-thickness rectal biopsy remains the criterion standard [3]. The definitive treatment is to remove the aganglionic bowel and to restore continuity of the healthy bowel with the distal rectum, with or without an initial intestinal diversion [4].

The aim of this study was to present a clinical case report and literature review of TCA in a child with Down syndrome (DS).

Case report

A full-term 24 hours old male neonate was admissioned to Children's Hospital, Affiliate of VU Hospital Santaros Clinics, he presented with symptoms of distal intestinal obstruction: bilious emesis, abdominal distension, and failure to pass meconium or stool. Upon arrival, the condition was severe due to intoxication. Gestational age 37 weeks. + 5d. ; Apgar 10/10. Radiograph revealed decreased air in the rectum and dilated proximal bowel loops. Characteristic dysmorphic features of Down syndrome were noticed: epicanthal folds, flat nasal bridge, upward-slanting palpebral fissures, short neck and generous nuchal skin, single transverse palmar crease. 8 days after admissiom down syndrome was confirmed with a genetic test (karyotype 47, XY +21). After 10 days a decision was made to perform a surgical intervention due to deteriorating condition of neonate. Right transrectal laparotomy was performed. Dilated small bowel loops were noted upon the opening of the abdominal cavity, no obstruction have been found - it was only reported that a 5-7 cm long segment of ileum proximally to ileocecal valve appears narrowed. The appearance of the large intestine was normal. The contents of the small intestine were pushed into the large intestine and, since Hirschprung's disease was suspected, full-thickness biopsies from caecum, ascending colon, hepatic flexure, transverse colon and splenic flexure were taken. Finally, a diverting double ileostomy was established in order to decompress the colon.

After surgical intervention postsurgical care were performed. The patientent was treated with antibiotics: Penicillin 330k IU x 2 IV; Gentamicin 13,2 mg x 1 IV; Nystatin 100k IUx 3 PO. Two days after right transrectal laparotomy the partial enteral feeding was renewed. The contents of the small intestine were transferred to the large intestine (via enterostoma) and came out through the rectum and anus. After month partial parenteral nutrition discontinued and CV catheter was removed. Upon discharge general condition was satisfactory. Body weight was growing steadily. The denitive diagnosis of Hirschsprung disease was confirmed by a

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full-thickness biopsy demonstrating the absence of ganglion cells in the submucosal and muscular layers of the intestine.

At 3 year of age patient underwent total colectomy with ileorectal anastomosis: lower midline laparotomy was performed and the entire large intestine proximal to the rectum as well as a distal part of the ileum were removed. Ileostomy and the aganglionic rectal stump were closed. The normoganglionic bowel was pulled down directly in front of the sacrum. The posterior rectal wall was incised above the dentate line, entering the previously dissected retrorectal space. The normoganglionic bowel was pulled through the rectal incision in the posterior wall of the rectum and the anastomosis between the aganglionic rectum and the normoganglionic pulled-down bowel was created.

Discussion

At least 12 genetic mutations have been identified in patients with HD [5]. The predominant gene affected is the RET proto-oncogene. In one study, RET variations were found in 82% of patients with TCA, as compared with 33% of those with short segment disease [6]. The strong association between Down syndrome and HD may be partly explained by mutations in the Down Syndrome Cell Adhesion Molecule gene (DSCAM), since it results in over-transmission of a hypomorphic RET allele [7]. DS is the most frequent chromosomal abnormality associated with HD - it is present in 2 to 16 % of individuals with HD [8]. HD should be suspected in patients with typical clinical symptoms and a high index of suspicion is appropriate for infants with a predisposing condition such as DS, or for those with a family history of HD [5]. Occasionally, affected infants may present with enterocolitis, a potentially life-threatening illness in which patients have a sepsis-like picture and can progress to toxic megacolon [9]. The treatment of choice for HD is surgical, such as Swenson, Soave, and Duhamel procedures. The goals are to resect the affected segment of the colon, bring the normal ganglionic bowel down close to the anus, and preserve internal anal sphincter function [10]. The traditional operation was an abdominoperineal pull-through in two or three stages, in which patients initially underwent a diverting colostomy with definitive repair performed later. However, most centers now perform the procedure in one stage - that does not appear to increase complication rates. Laparoscopic-assisted and transanal repairs are now preferred over the open procedures [11]. The results seem to be equal to the traditional abdominoperineal pull-through with the added benefit of earlier resumption of full feeds, less pain, shorter hospitalization, and less conspicuous scars. Patients with total colonic involvement require modified procedures to exclude the aganglionic colon while preserving maximal absorptive epithelium. The goal of these procedures is to bypass the dysfunctional bowel while maximizing the chance of postoperative nutritional function and growth [12].

Conclusion

The most common long-term complications are fecal incontinence, constipation, and enterocolitis. Patients with trisomy 21 or other syndromes are more likely to have constipation or incontinence. Total colon aganglionosis has a much higher rate of complications and mortality before and after definitive treatment, compared with the more common forms of HD in which a smaller portion of the colon is affected. TCA also has a much higher risk for enterocolitis and poor functional results, including incontinence after surgery.

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RETA HIRŠPRUNGO LIGOS FORMA: ATVEJO PRISTATYMAS

I. Skalskis

Raktažodžiai: Hiršprungo liga, totalinės storosios žarnos aganglionozės, Dauno sindromas.

Santrauka

Hiršprungo liga (HL) yra raidos sutrikimas, kuriam būdingas ganglijų nebuvimas storosios žarnos distalinėje dalyje, sukeliantis funkcinę obstrukciją. Totalinės storosios žarnos aganglionozės dažnis yra 1 iš 500 000 ir sudaro 5-10 proc. visų HL atvejų. HL reikia įtarti pacientams, kuriems pasireiškia būdingi šiai patologijai klinikiniai simptomai ir tuomet, kai kūdikiui diagnozuojamas Dauno sindromas, ar jei giminėje buvo pasireiškusi ši liga. Pagrindinis HL gydymas yra chirurginis, pavyzdžiui, Swenson, Soave ir Duhamel procedūros. Tikslas – pašalinti pažeistą gaubtinės žarnos segmentą, priartinti normalią ganglioninę žarną prie išangės ir išsaugoti vidinę analinio sfinkterio funkciją. Straipsnyje pristatomas Dauno sindromu (DS) sergančio vaiko totalinės storosios žarnos aganglionozės klinikinis atvejis ir literatūros apžvalga.

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