CASE REPORT: RARE FORM OF HIRSCHSPRUNG’S DISEASE

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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).
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.

**References**

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Santrauka


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