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## Hirschsprung disease—Bowel function beyond childhood

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## ABSTRACT

Hirschsprung disease is a developmental defect of the enteric nervous system characterized by lack of enteric neurons in the distal hindgut. There are numerous reports on short-term outcomes indicating that impaired bowel function is common. Recently, several controlled studies show that bowel function outcomes are affected beyond childhood, in adolescents and adults, compared with healthy control subjects. Constipation and fecal incontinence are common. The impaired bowel function appears to have a negative impact on quality of life, although, a majority of patients have adapted to their symptoms. On the other hand, Hirschsprung disease seems to have limited impact on education and occupation in adult life.

The aim of this review was to summarize current knowledge of bowel function outcome beyond childhood in patients with Hirschsprung disease.

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## Introduction

Hirschsprung disease (HSCR) is a congenital malformation of the enteric nervous system characterized by absence of ganglion cells in the distal hindgut. The ganglion cells are replaced by hypertrophied nerve trunks. The birth prevalence of HSCR is approximately 1 in 5000 live births, with a male-to-female ratio 3.2:1.<sup>1</sup> The disorder always affects the anal canal and most commonly, 75% of the cases, involves the rectum and sigmoid colon. Less commonly a longer segment of the colon is aganglionic. In approximately 10% of the cases the whole colon and part of the small bowel is involved, total colonic aganglionosis (TCA). Total intestinal aganglionosis, affecting the whole small bowel, is extremely rare. The etiology is unknown, but HSCR is considered a multifactorial disease. A genetic basis has long been suggested by the familial occurrence in 5–20% of the patients and the association with syndromes. More recently, it has been shown that coding *RET* gene mutations account for 7–25% of sporadic cases and 40–60% of familial cases. Mutations in several other genes have also been associated with HSCR.<sup>2,3</sup> The disease most commonly results in neonatal bowel obstruction with delayed passage of meconium, distended abdomen and bile-stained vomiting, but patients may also present later with intractable constipation or Hirschsprung-associated enterocolitis (HAEC). The principal purpose of surgical management of HSCR is to remove aganglionic bowel and

anastomose normally innervated bowel to the anus, while preserving normal sphincter function. Over the last decades management has changed from original three-stage approach to recent introduction of minimally invasive one-stage procedures, which appears to be safe, efficient, and facilitates early feeding and discharge.<sup>4</sup> Total transanal endorectal pull-through and laparoscopic pull-through techniques, with various length of the muscular cuff, have gained worldwide popularity.<sup>5,6</sup> Despite improved outcomes of HSCR due to better understanding of the pathophysiology and refined surgical technique, impaired bowel function, mainly fecal incontinence and constipation, is common in childhood.<sup>7–9</sup> Many patients need specialized care to achieve social continence and to treat constipation. Some patients benefit from bowel management programs. Occasionally, botulinum toxin injections or redo surgery is required. Despite these efforts, persistent bowel function impairment is common also in adulthood.<sup>10–15</sup> In recent years there has been an interest in the effect of HSCR on quality of life and psychosocial functioning.<sup>16,17</sup> Patients with HSCR report slightly more quality-of-life problems than comparison groups.<sup>16</sup> The aim of this article was to review the bowel function outcomes in adolescents and adults. The focus is set on bowel function, although the impact on quality of life will be briefly discussed.

## Short-term outcomes

There are numerous reports of the short-term bowel function outcome after different surgical procedures for HSCR. The general opinion is that short-term outcomes do not

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differ significantly between the various surgical procedures. Many of the studies are retrospective and uncontrolled. More recently, some prospective, controlled studies have been published. The data are often “surgeon-reported” or reported by the parents, and does not necessarily reflect the patient’s opinion.

#### *Postoperative complications*

Early postoperative complications are important because, for instance, anastomotic leaks and severe strictures may have a negative impact on long-term functional outcomes. It has been shown that 11% and 14% of the patients experience at least one early complication after transanal and transabdominal Soave procedures, respectively.<sup>18</sup> Similarly, Thakkar et al showed that 15% of the patients had early complications.<sup>19</sup> Perianal excoriations are very common and can usually be treated with ointments.<sup>4</sup> Anastomotic leaks are relatively rare and have been reported to occur in 0–5.6% of the patients.<sup>4,20</sup> An anastomotic leak often results in scarring, which affects long-term function negatively. Strictures have been reported to occur in 4–9% of the patients.<sup>4,18,20</sup> Strictures can often be dilated, but occasionally a redo pull-through is required. Some patients also need redo surgery for narrowing of the muscular cuff, twisted pull-through, residual aganglionosis, or transition zone pull-through. Residual aganglionosis and transition zone pull-through is the underlying cause of persistent obstructive symptoms in one-third of the patients who need a redo pull-through.<sup>21</sup> Redo pull-through probably has a negative impact on the functional outcome, but data are slightly conflicting. There are reports indicating that outcomes are satisfactory after redo pull-through, although others have shown that both stooling scores and fecal continence are affected.<sup>21–23</sup> Adhesive small bowel obstruction was recently reported to occur in as many as 29% of patients with HSCR.<sup>24</sup>

#### *Enterocolitis*

HAEC is a serious complication of HSCR and account for both mortality and morbidity related to HSCR. It has been shown that patients with a history of HAEC have a worse functional outcome, lower bowel function score, than healthy controls.<sup>15</sup> The etiology of HAEC is complex. Recently, there has been a lot of interest in the role of the microbiota in the development of HAEC and also the association between HAEC and inflammatory bowel disease.<sup>25,26</sup> HAEC is clinically characterized by abdominal distension, explosive diarrhea, and occasionally fever and sepsis. HAEC usually occurs within the first few years after pull-through.<sup>18</sup> Postoperative HAEC has been reported to occur in 6% to 45% of the patients.<sup>4,15,18</sup> In 1989, Sherman reported episodes of HAEC in almost 30% of patients undergoing Swenson procedure. They found that the incidence had not changed over a 4-decade period.<sup>27</sup> On the other hand, Suita et al. showed that the incidence of HAEC decreased over the years. The lack of a definition of HAEC has been one reason for the wide range of reported incidence.<sup>28</sup> Pastor et al. developed a score, which was based on the opinions of a panel of experts, using the Delphi method to reach a consensus. The score has been useful for research purposes, but is more difficult to apply in clinical practice.<sup>29</sup> Treatment consists of fluid and electrolyte resuscitation, bowel irrigation, and antibiotics. In patients with mild symptoms metronidazole is usually sufficient, while patients with severe symptoms need broad-spectrum antibiotics. A lot of efforts have been made to prevent HAEC. Prophylactic rectal irrigations decrease the incidence of postoperative HAEC.<sup>30</sup> There is no strong evidence that prophylactic antibiotics prevent recurrent HAEC. There are also conflicting data with respect to

probiotics. It has been shown that botulinum toxin injections in the internal anal sphincter reduce the number of readmission related to HAEC.<sup>16,25,31</sup>

#### *Bowel function*

The general opinion is that stooling pattern changes and the number of daily bowel movement decreases with increasing age.<sup>18</sup> Rescorla et al. showed that 65% of 260 patients were very satisfied with the bowel function outcome. A lower proportion of patients followed less than 5 years were very satisfied compared to patients followed more than 10 years.<sup>32</sup> In 1995, Heij et al. reported that of 14 children less than 4 years of age 8 had spontaneous defecation, 6 had severe constipation and all were incontinent.<sup>33</sup> In 49 patients over 4 years of age 10 were continent without constipation, 22 had soiling and/or, constipation, 17 were incontinent, and one had a permanent stoma.<sup>34</sup> Several more recent studies have shown that a significant proportion of the patients have fecal incontinence and, or, constipation. Granstrom et al.<sup>9</sup> showed that 67% of patients who had undergone laparoscopic assisted transanal pull-through had some degree of soiling. Stensrud et al.<sup>35</sup> also reported that more than 50% of the patients had occasional soiling after total transanal or laparotomy-assisted pull-through. Neuvonen et al. showed that children aged 4–12 years had significantly lower bowel function score compared to healthy controls. The patients had worse rectal sensation, more abnormal defecation frequency, more fecal accidents, more problems withholding defecation, more fecal soiling, and were more socially restricted than healthy subjects.<sup>15</sup>

#### **Assessment of bowel function**

It is difficult to compare bowel function outcomes in different studies since there is no consensus on how bowel function should be assessed. Interpretation of the results is also difficult due to limited sample size and the fact that different surgical procedures have been used. The terminology may also be misleading and cause interpretation problems. For instance, in pediatric surgery, fecal incontinence usually consists of soiling (minor leakage of feces or staining of the underwear) and accidents (emptying of larger amounts of feces into the underwear or diapers). In adult colorectal surgery fecal incontinence is often categorized as leakage to solid or loose stools, urgency incontinence, and soiling (leakage of fluid). Currently, the Rintala score (bowel function score) and the Krickbeck criteria are frequently used to evaluate bowel function in HSCR patients.<sup>36,37</sup> The advantage of the Rintala score is that it has been validated in healthy normal subjects.<sup>38</sup> In many studies the outcome has been reported by the surgeon without using a validated questionnaire or scoring instrument. If the surgeon who originally performed the operation is assessing the patients’ bowel function this may introduce information bias. For adults there are numerous scoring systems for fecal incontinence, such as the Wexner score and Vaizey score.<sup>39,40</sup>

#### **Anal sphincters and the anal canal**

The basic principle of surgical procedures for HSCR is to resect the aganglionic part of the colon and to connect normally innervated intestine to the anal canal. The aim of all procedures is to avoid damaging the anal canal and to preserve the anal sphincters. When doing a transanal pull-through the dissection should always be started 5–10 mm above the dentate line, whether it is a submucosal or a full-thickness dissection. The muscle layers are divided above the internal anal sphincter. If the pull-through is done via a transanal approach it is extremely

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