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The Evolution of Surgical Techniques in the Management of Hirschsprung's Disease: A Historial Review

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Rezumat

Evoluția tehnicilor chirurgicale în managementul bolii Hirschsprung: o analiză istorică

Harald Hirschsprung a oferit prima descriere detaliată a rezultatelor clinice la pacienții cu megacolon congenital. În secolul următor, s-au realizat progrese semnificative în înțelegerea cauzelor, îmbunătățirea diagnosticului şi dezvoltarea unor tratamente eficiente pentru boala Hirschsprung (HD). Acest articol prezintă evoluția managementului chirurgical al HD, începând cu Orvar Swenson, care a introdus prima disecție transrectală completă, şi evidențiind contribuțiile altor chirurgi inovatori a căror muncă a ajutat la conturarea abordărilor actuale pentru tratarea bolii.

Cuvinte cheie: Hirschsprung, megacolon, istorie chirurgicală

Abstract

Harald Hirschsprung provided the first thorough description of clinical outcomes in patients with congenital megacolon. Over the following century, significant advancements were made in understanding the causes, improving diagnosis, and developing effective treatments for Hirschsprung's disease (HD). This article outlines the evolution of surgical management for HD, starting with Orvar Swenson, who introduced the first complete transrectal dissection, and highlighting the contributions of other innovative surgeons whose work helped shape the current approaches to managing the disease.

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Key words: Hirschsprung, megacolon, surgical history

19th Century

In 1825, the first documented autopsy revealed a constricted rectosigmoid and an expanded proximal colon in an adult with chronic constipation, who died from acute colonic obstruction. Harald Hirschsprung (*Fig. 1*), born in Copenhagen in 1830, was a pioneering pediatrician whose work laid the foundation for modern pediatric surgery. Among his contributions was the documentation of four cases of esophageal atresia with tracheoesophageal fistula, highlighting the high fatality risk. His significant advancement came with his early technique for the hydrostatic reduction of ileocolic intussusception, reducing mortality rates compared to untreated cases, a method that remained relevant into the mid-1900s (1).

Before Hirschsprung's landmark description of congenital megacolon in 1886, over 20 reports had discussed similar conditions. In January 1886, Hirschsprung presented a paper to the Pediatric Society in Berlin, proposing that congenital megacolon was a rare anomaly in newborns. His "Malformation Theory" suggested that colonic dilation occurred shortly after birth, though he overlooked the narrow rectosigmoid as the root cause, focusing instead on the dilated proximal colon (*Fig. 2*). This misunderstanding delayed the full understanding of the disease for another 60 years. Nevertheless, his presentation became the classic description of what is now known as Hirschsprung's disease (2).

Following his foundational lecture, Hirschsprung published cases of congenital hypertrophic pyloric stenosis, while other physicians, like Osler, noted colon innervation defects. By 1895, Marfan proposed that a redundant sigmoid could cause obstruction, leading to proximal colon dilation and hypertrophy, treatable with a rectal tube. Subsequent discussions suggested colonic irrigation and colostomy as effective treatments, methods that remain relevant in some cases

Figure 1. Harald Hirschsprung
Source: https://litfl.com/harald-hirschsprung/

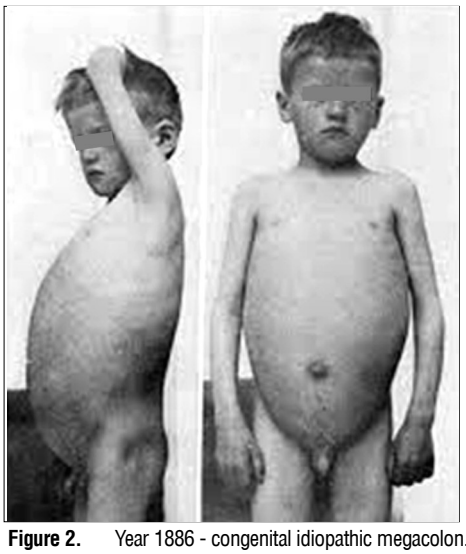

Figure 2. Year 1886 - congenital idiopathic megacolon.
Source: https://jamanetwork.com/journals/jamasurgery/article-abstract/537337

today. In 1898, Treves performed a colostomy on a young girl with chronic constipation, improving her symptoms but failing to identify the rectosigmoid obstruction as the cause, mistakenly attributing the issue to the anus (3).

20th Century

Reports of absent ganglion cells in Hirschsprung's Disease (HD) began surfacing but were initially considered rare anomalies. Fenwick argued that a tight anal sphincter due to spastic rectal contraction caused mechanical obstruction, leading to proximal colon dilation, but his treatments did not improve outcomes. Bayliss and Starling contributed to the understanding of peristalsis, focusing on how it moves material through the colon (4).

Between 1902 and 1924, a variety of treatments were explored, including resections, rectal tubes, enemas, myotomy, and drug therapies. While some methods offered temporary success, most cases of HD relapsed. In 1904, Hirschsprung authored the first book chapter on "Congenital Dilatation of the Colon." Despite his retirement that same year, his contributions remained central to understanding the disease, even as a range of opinions about its cause persisted. Hawkins, in 1907, suggested a neuromuscular defect as the cause, but many of these theories remained speculative.

Hirschsprung passed away in 1916, having left a lasting impact on pediatric surgery, with his contributions extending beyond the disease that bears his name. Between 1923 and 1944, several less invasive surgical techniques were tested but ultimately failed in the long term. In 1927, Wade and Royle proposed lumbar sympathectomy as a treatment, which temporarily restored colon function (5).

By the late 1920s and 1930s, textbooks reflected a grim prognosis for infants with HD, with most dying before the age of five. Surgeons like Ask-Upmark and Ross explored resection of the dilated colon and the effectiveness of lumbar sympathectomy but faced high mortality rates. In 1938, Robertson and Kernohan identified the absence of ganglion cells in the rectosigmoid, linking it to proximal colonic obstruction in infancy (6).

By 1940, Mayo Clinic surgeons, including Whitehouse, Bargan, and Dixon, concluded that segmental resection of the dilated colon was the most effective treatment despite a 24% mortality rate. Ehrenpreis proposed that proximal colon dilation resulted from distal obstruction but did not fully recognize the narrowed rectosigmoid as the primary cause (7).

From Hirschsprung's 1886 report to Orvar Swenson's contributions in the 1940s, over 200 case reports speculated about the disease's cause, with little consensus. However, Swenson and Bill made a significant breakthrough in 1948, demonstrating that the narrow rectosigmoid was the root cause of HD, advocating for resection of the aganglionic segment as the appropriate treatment (8).

In 1948, Zuelzer and Wilson published a series of 11 infants with classic HD symptoms, noting the absence of ganglion cells in the rectosigmoid. Despite their correct diagnosis, all infants died, leading them to recommend temporary enterostomy to assess bowel motility (9).

Throughout the 1950s, various surgical techniques, such as the Swenson pull-through procedure, emerged to treat HD. In 1951, Hiatt confirmed that the pathological site was the rectum, lacking peristalsis and normal sphincter reflexes. Swenson, in 1955, introduced rectal biopsy as a highly accurate method for diagnosing HD, marking it as superior to barium enemas (10).

In 1956, Duhamel described a posterior colorectal anastomosis, a technique designed to preserve rectal mucosa and minimize pelvic nerve injury. Although initially flawed, Grob, Genton, and Von-Tobel refined the procedure to preserve the internal sphincter. The 1960s saw further advances in rectal biopsy techniques and modifications to surgical procedures, such as Swenson's adjustments to reduce postoperative complications like enterocolitis. These developments led to significant improvements in outcomes for HD patients (11,12).

In 1964, Swenson reviewed his first 100+ cases, noting that some patients developed postoperative enterocolitis, which required sphincterotomy. He attributed this complication to leaving too much distal rectum during his initial pull-through procedure. By adjusting the resection to leave less rectum, the incidence of enterocolitis decreased significantly, while continence was preserved. Soave introduced a modification involving submucosal dissection, removing the aganglionic mucosa from the rectosigmoid and performing an endorectal pull-through of the proximal colon. Boley later modified Soave's technique by performing a primary anorectal anastomosis during the initial surgery (13,14).

In January 1948, Swenson and Bill presented evidence that the disease's cause was linked to a defective distal rectosigmoid segment, proving that simply removing the dilated proximal colon was insufficient for a cure. They introduced a curative surgery that included a temporary colostomy, which they had previously experimented with in dogs. This technique was later applied successfully in a child with Hirschsprung's Disease (HD) (15).

Zuelzer and Wilson, in February 1948, reported on 11 infants with classic HD symptoms. Despite identifying the absence of ganglion cells in the rectosigmoid, all infants died. They suggested using a temporary enterostomy to assess the non-motile bowel segment.

Following the introduction of the Swenson pull-through procedure, various modifications arose in the 1950s, confirming the defective rectosigmoid as the disease's cause. Hiatt used manometry in 1951 to confirm the rectum's pathological involvement, while Swenson, Fisher, and MacMahon introduced rectal biopsy in 1955 as a more reliable diagnostic tool than barium enemas (16).

Duhamel, in 1956, described a face-to-face posterior colorectal anastomosis to preserve the rectal mucosa, minimizing pelvic nerve injury. Rehbein improved the technique in 1958 by performing a lower resection and anastomosis (17). In 1961, Shandling refined biopsy techniques, using laryngeal biopsy forceps for submucosal biopsies without anesthesia (18).

Key developments between 1965-2000 include:

- 1965: Dobbins and Bill established rectal aspiration biopsy as an effective diagnostic tool for HD, identifying ganglion cell absence and staining for acetylcholinesterase (19).
- 1970: Ehrenpreis highlighted the complications of HD surgeries, especially involving the internal sphincter, noting that too much damage led to incontinence, while preserving too much caused recurring obstruction and enterocolitis (20).
- 1985: Laparoscopic surgery became widely adopted, including for endorectal pull-through procedures.
- 1998-1999: New trans-anal surgical approaches were introduced, avoiding abdominal entry but posing risks like higher rates of strictures and incontinence.
- 2000: The American Association for Pediatric Surgery reported a drop in neonatal mortality from 70% in 1954 to 1%, due to early diagnosis and improved surgical techniques (21).

21st Century

Between 2000 and 2003, significant progress was made in understanding the genetic and molecular mechanisms behind Hirschsprung's Disease (HD) (*Fig. 3*). Key breakthroughs included identifying multiple genes, particularly the RET gene, as central to the disease's pathogenesis. Studies in 2003 highlighted the role of dysfunctional neural crest stem cells and the RET gene's importance in cell migration. By 2004, over 10 genes were linked to HD, with RET remaining a focus due to its significant impact (22-24).

Procter and colleagues in 2003 warned against relying solely on contrast enemas for determining the level of aganglionosis, suggesting biopsies to confirm the transition zone, especially in long-segment cases. By 2005, the trans-anal pull-through procedure

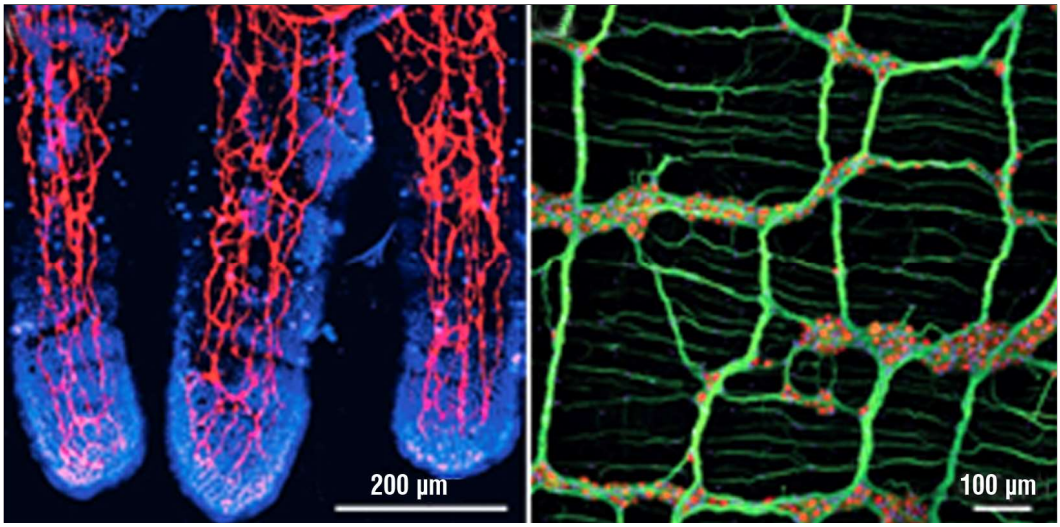

Figure 3. Hirschsprung's disease - integration of basic science and clinical medicine
Source: https://www.nature.com/articles/nrgastro.2017.149

gained popularity, but concerns emerged when applied to infants with long-segment HD, particularly when the transition zone was in the proximal colon or if complications like enterocolitis or abdominal distension were present. Some surgeons utilized laparoscopy or an umbilical incision to address these challenges. Despite these advances, the Duhamel technique remained a preferred method for treating long-segment HD due to its superior functional outcomes (25-27).

However, adverse outcomes like enterocolitis, fecal incontinence, and chronic constipation persisted, leading to research into alternative stem cell-based therapies. Around 21 genes involved in neural crest cell function and the development of the enteric nervous system were identified as being associated with HD, with mutations present in about 30% of patients. The RET proto-oncogene was linked to over 200 loss-of-function mutations in both sporadic (20%) and familial (up to 50%) cases. Reduced penetrance and variability in RET-related HD were partially explained by single-nucleotide polymorphisms. Additionally, epigenetic changes, including DNA methylation, histone modifications, and non-coding RNA, were explored as contributors to HD pathology (28-30).

These genetic and molecular findings have significantly influenced the understanding, diagnosis, and treatment of HD.

Conclusions

- The evolution of surgical techniques for treating Hirschsprung's disease represented a remarkable advance in paediatric medicine. From the first pioneering interventions to modern minimally invasive methods, each step has contributed to increasing success rates and reducing complications. Studying the history of these techniques highlights the importance of continued innovation and research to improve patient outcomes.
- The history of surgical treatment for Hirschsprung's disease illustrates an eloquent example of how determination and medical progress can radically transform the prognosis of a complex disease. Each new technique has brought significant improvements, giving patients a better quality of life and more effective treatment. Continued research and the adoption of new technologies are essential to further optimise surgical interventions.

- Looking back at the history of surgical treatment for Hirschsprung's disease, it is evident that progress has been made through collaborative efforts between surgeons, researchers and technological innovations. Modern techniques, which provide safer and more effective interventions, are the result of decades of dedication and study. In the future, it is anticipated that new discoveries in genetics and biotechnology will continue to revolutionise surgical approaches.
- The development and refinement of surgical techniques for Hirschsprung's disease reflects not only scientific progress, but also a deep commitment to the health and wellbeing of patients. Each advance was built on previous lessons and experiences, thus providing a solid foundation for future innovations. Looking ahead, there is justifiable optimism that new techniques and discoveries will continue to improve the treatment of this disease.
- The evolution of the surgical treatment for Hirschsprung's disease is a testament to medical progress and the human ability to overcome complex challenges. From the first exploratory surgeries to today's advanced procedures, each stage has brought significant improvements in the management and outcomes of this condition. The future promises even more innovation, driven by a deeper understanding of the disease and the emerging technologies.

Conflict of Interest

The authors declared no potential conflicts of interest.

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