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History of surgery for Hirschsprung disease: a view from Melbourne

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The modern story of Hirschsprung disease (HSCR) begins in 1886 in Berlin, when Harald Hirschsprung, a professor of pediatrics in Copenhagen, presented a case report of two infants with constipation who died and had colonic dilatation and hypertrophy.

In the first case, the infant suffered from constipation from birth that had only responded to laxatives and enemas. Before his demise at 11 months of age, he spent 2 months in the hospital under active treatment for his constipation before developing diarrhea. The megacolon regressed with treatment, but then recurred after discharge just before he died from sepsis (what we would now recognize as toxic megacolon and enterocolitis). The second case was similar, affecting a boy with constipation that had been present from birth, leading to acute enterocolitis and death. In both cases, the proximal colon was grossly dilated, and in the second case, a rectal examination had revealed an empty rectum with no evidence of a stricture.[1]

In 1970, Ehrenpreis,[2] the head of pediatric surgery at the Karolinska Institute, wrote a monograph on HSCR. He described the search for a cause after its recognition in the 1880s, which took a multitude of observers and 60 years of speculation and imagination before it was finally understood in 1948.

It is an intriguing question as to why it took so long to unravel the pathophysiology of HSCR, but it is easy to overlook the fact that in the first half of the 20th century the understanding of the anatomy of the nerve supply and physiology of the bowel was rudimentary. It was well known that sympathetic nerves inhibited intestinal tone and peristalsis and contraction of the anal sphincter. By contrast, parasympathetic stimulation produced the opposite effect. However, it was not until the post-war years that it was firmly established that the intestine can contract and undergo peristalsis without external innervation in response to mechanical stimuli. In addition, these findings were complemented by an improved understanding of the role of the internal neuronal networks that form the myenteric (Auerbach) plexus and the submucosal (Meissner) plexus.

The intrinsic plexuses of the colon were first identified in a patient with HSCR in 1901, presumably proximal to the aganglionic distal segment.[3] A neuronal cause was suspected, but there were many alternative hypotheses, such as a primary 'malformation' to explain the megacolon and the possibility of a presumed distal obstruction from a mechanical cause. Infants were being investigated with the new study of a barium enema, which initially demonstrated no megacolon in the neonatal period but with megacolon as a later finding. This suggested that the megacolon in HSCR was a secondary, functional response to distal obstruction.

In 1948, Zuelzer and Wilson[4] described 11 patients with clinical, radiological, and pathological findings of presumptive HSCR. In six of these patients, there was a proximal megacolon associated with an undilated and aganglionic distal colon. Shortly after, Swenson and Bill[5] demonstrated that the distal colon was in a 'spasm' in a series of 20 patients with HSCR. They were able to show this by modifying the then-standard method of barium enema. Instead of just filling the entire colon with barium, they just filled the distal colon at low pressure until the contrast entered the dilated segment, thereby revealing the spastic, tonically contracting distal colon clearly.

Around the same time, Whitehouse and Kernohan[6] showed that the ganglion cells were absent in the distal colon in 11 patients. These findings were also associated with an abundance of non-myelinated nerve fibers between the circular and longitudinal muscle layers of the colon, where the ganglion cells should have been. Then, in 1949, Bodian *et al*.[7] from our own institution (The Royal Children's Hospital, Melbourne) described 73 cases of megacolon and were able to demonstrate that only 39 patients had the clinical and pathological evidence of HSCR, while

34 had idiopathic megacolon.[8] This finally explained the heterogeneity and mixed results of previous reports, which were an amalgam of two different disease entities.

The diagnosis of an aganglionic distal colon was confounded by the discovery that ganglion cells are normally absent from the entire length of the internal anal sphincter.[9] One of the early studies to define the normal distribution of the ganglion cells was conducted by Magnus,[10] who was a junior surgeon in Douglas Stephens' surgical research unit in Melbourne. She studied the histology of eight subjects, ranging in age from 1 week to later childhood, and found no ganglion cells distal to the level of the anal valves (the so-called dentate line). Above this level, there was a variable segment of 1–8 mm containing occasional ganglion cells, leading to a normal network of ganglion cells proximal to this.

The recognition of abundant non-myelinated nerve fibers in the aganglionic segment was important for the understanding and later development of one of the modern diagnostic tests for HSCR.[6] The embryology of the enteric nervous system was first studied in 1954 by Yntema and Hammond,[11] who found that the internal plexuses of ganglion cells were derived from the vagal neural crest cells. These embryonic nerves enter the primitive pharynx and then migrate distally through the developing intestine to reach the end of the rectum. Later, the extrinsic autonomic nerves migrate into the intestine through the mesentery and then form synapses with the intrinsic nerves in the ganglia.

In HSCR, the lack of ganglion cells causes the extrinsic nerves to proliferate, effectively searching for a ganglion to make a synapse. Once a synapse is established, all of the neurofibrils normally degenerate, but in the aganglionic colon they persist. This hyperplasia of the extrinsic nerves enabled the development of the acetylcholinesterase staining method as a diagnostic test for HSCR, as pioneered by Peter Campbell in the 1970s in the Department of Pathology at The Royal Children's Hospital, Melbourne.[12] This staining technique was based on biopsies obtained using the newly developed suction rectal biopsy gun pioneered by Noblett,[13] a pediatric surgeon at The Royal Children's Hospital, Melbourne.[14]

As discussed previously, the use of a limited, low-pressure barium enema was not only crucial in advancing the understanding and diagnosis of HSCR but also augmented the treatment of HSCR. Working at the Children's Hospital of Boston, Swenson and Bill, in their study of 20 patients, were able to demonstrate persistent spasm in the distal colon, which they treated successfully with a bypassing proximal colostomy. They then demonstrated that local excision of the contracted (and aganglionic) segment, with anastomosis of the proximal, innervated colon to the anus was a definitive cure for the otherwise fatal condition. This established Swenson's rectosigmoidectomy as the first successful treatment for HSCR.

Once local excision of the aganglionic segment was recognized as a viable treatment, there was a sudden proliferation of variations to the procedure, with the aims of (1) removal of some or all of the aganglionic colon/rectum, (2) anastomosis of the proximal innervated colon above the anal canal, (3) preservation of the important autonomic nerves in the pelvis to avoid causing impotence, and (4) avoidance of stretching of the anal sphincters to preserve long-term continence. As the procedures were being done in older infants because of the risks of anesthesia, the increasing depth of the pelvis became an issue for the pelvic part of the procedure. This put the structures in the lateral wall of the pelvis, particularly the autonomic nerves, as well as the bladder, urethra, and pre-pubertal prostate at risk.

Swenson's own results were excellent, but other less experienced surgeons were unable to replicate these results. This led to a search for alternative procedures that lowered the risk of complications to pelvic structures. Duhamel[15 16] pioneered the retrorectal and transanal pull-through procedure, using a side-to-side anastomosis to avoid pelvic nerve damage. Rehbein[17] modified the anterior resection procedure, while retaining a significant portion of aganglionated rectum. Soave[18 19] then developed and presented the endorectal procedure to bypass damage to the pelvic structures outside the wall of the rectum, which were at risk in a less elegantly performed Swenson procedure. This technique was enhanced by Boley,[20] with the suggestion of a primary coloanal anastomosis. It is important to note that the concept of the Soave procedure was first devised and published over a decade earlier by Yancey *et al*.[21] However, due to racial issues at the time, his work was not given the recognition it deserved, a situation that is now being rectified. We encourage our surgical colleagues to now refer to this procedure as the Yancey-Soave pull-through.

In Melbourne, the Duhamel procedure was the standard operation through the 1960s and 1970s, but then Franco Soave came as a visiting professor in 1978 to demonstrate his technique. His endorectal rectosigmoidectomy via laparotomy became the standard procedure at our hospital until the introduction of the transanal approach and its laparoscopic modifications. The transanal approach was first described by De la Torre-Mondragón and Ortega-Salgado[22] in 1998, following its successful introduction in five patients. Similar successful findings were described the following year by Langer *et al*.,[23] and the transanal approach became widely accepted. The technique was further enhanced by the popularization of the laparoscopically assisted pull-through first described by Georgeson *et al*.[24]

As has been shown by a multitude of authors, the short-term and long-term outcomes for each of these procedures are relatively similar. It is most likely the case that the technique that is most regularly performed by the individual surgeon, as long as that surgeon is exposed to a suitable number of patients, is the best technique for that surgeon's patients. It is our responsibility to determine the outcomes for our patients, even if those outcomes will not be known for another decade or more. For those readers interested in alternative perspectives regarding

the history of surgical approaches to HSCR, the authors recommend the reviews by Sergi[25] and Swenson.[26]

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