Overview of Hirschsprung Disease: A Narrative Literature Review

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1. Introduction

Hirschsprung disease, megacolon aganglionic congenital, is a functional obstruction of the large intestine. It is the most common cause of large bowel obstruction, accounting for about one-third of all gastrointestinal obstructions in infants. The incidence is approximately 1 in 5000 live births, with an increased incidence in males, siblings of children with Hirschsprung's disease, and children with Down syndrome or other congenital malformations. Familial occurrence is 5% to 20%. The exact cause is unknown, but several factors interact, including gene mutations and epigenetic mechanisms.1-3 This literature review aimed to describe the pathophysiology, clinical symptoms, and treatment of Hirschsprung disease.

Pathophysiology of Hirschsprung disease

Hirschsprung disease is a congenital malformation resulting from the failure of neural crest cells to migrate into the gastrointestinal (GI) tract. The absence of ganglion cells intrinsic to the parasympathetic nervous system in the submucosal plexus and myenteric plexus (Meissner’s and Auerbach’s plexuses) along the length of the colon results in a lack of nerve stimulation so that the muscular layer of the colon wall fails to push feces through the colon and causes functional obstruction. In 80% of cases, segment aganglionic is limited to region rectosigmoid (Hirschsprung short segment disease); in about 5% of cases, the entire colon is devoid of ganglion cells, and the ileum may be involved. The abnormally innervated colon interferes with stool movement, causing the proximal colon to
distend, and is, therefore, called megacolon (Figure 1). In rare cases, the entire intestine may be involved, which is known as total intestinal Hirschsprung disease.4-9

Figure 1. Congenital aganglionic megacolon (Hirschsprung disease).

Clinical manifestations

Infants usually become symptomatic during the first 24 to 72 hours after birth with delayed passage of meconium. Mild to severe constipation is a common manifestation of Hirschsprung's disease with poor diet, poor weight gain, and progressive abdominal distention. However, diarrhea may be the first sign, as only water can flow around the affected stool. With Hirschsprung's disease segment super short, mild constipation is usually the only symptom. These children may not be diagnosed until adulthood.

The most serious complication in the neonatal period is Hirschsprung-associated enterocolitis (HAEC), which can occur preoperatively and postoperatively. Intestinal inflammation is associated with fecal impaction, altered gut microbiota, impaired mucosal barrier function, and innate immunity by bacterial translocation. The dilated bowel stretches and partially occludes the surrounding blood and lymphatic vessels, causing edema, ischemia, mucosal infarction, and significant outflow of fluid into the intestinal lumen. The result of excessive liquid stools. Infarction and destruction of the mucosa allow enteric microorganisms to penetrate the intestinal wall. Often, gram-negative sepsis occurs, accompanied by fever and vomiting. Severe and rapid fluid and electrolyte changes can occur, resulting in hypovolemic or septic shock or death.10-15

Evaluation and treatment

Radiocontrast enemas and anorectal manometry are screening tools for the diagnosis of Hirschsprung's disease. A definitive diagnosis is made by rectal biopsy, which shows no ganglion cells in the submucosa of the colon. Surgery is the definitive treatment in all cases of Hirschsprung's disease, with resection of the aganglionic segment and anastomosis of the bowel proximal to the anus, known as the pull-through procedure. Laparoscopic or open approaches can be used. In general, the prognosis for Hirschsprung's disease is satisfactory for children undergoing surgical treatment. Bowel training can be extended; however, most children achieve bowel continence before puberty, whereas others develop long-term constipation or fecal incontinence.16-20

2. Conclusion

Hirschsprung disease, megacolon aganglionic congenital, is a functional obstruction of the large intestine. This disorder is caused by multifactorial, especially the role of genetic aspects.

3. References

1. Martucciello G. Hirschsprung’s disease, one of the most difficult diagnoses in pediatric surgery: a review of the problems from clinical practice


