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Overview of Hirschsprung Disease: A Narrative Literature Review

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A B S T R A C T

Hirschsprung disease is a congenital malformation resulting from the failure of neural crest cells to migrate into the gastrointestinal tract. The exact cause of this disorder is unknown, but it is suspected that there is an interaction of several factors, including gene mutations and epigenetic mechanisms. This literature review aimed to describe the pathophysiology, clinical symptoms, and treatment of Hirschprung disease. 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. The most serious complication in the neonatal period is Hirschsprung-associated enterocolitis (HAEC), which can occur both preoperatively and postoperatively. 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 a pull-through procedure. In conclusion, Hirschsprung disease is a congenital aganglionic megacolon or functional obstruction of the large intestine and is caused by multifactorial factors, especially the role of genetic aspects.

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 Hirschprung disease.

Pathophysiology of Hirschprung 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]

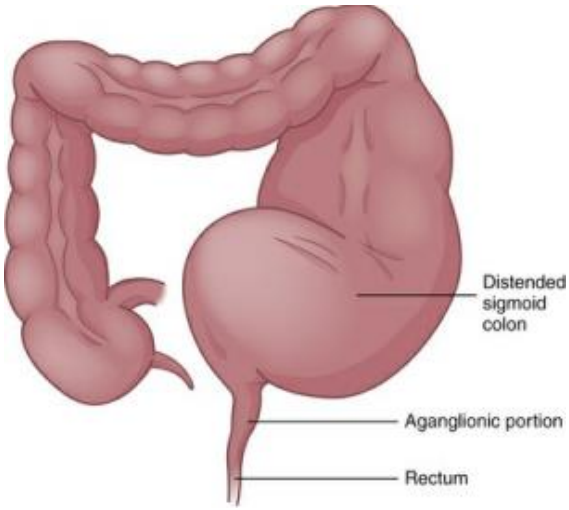

Figure 1. Congenital aganglionic megacolon (Hirschsprung disease).

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 to the bench. Eur J Pediatr Surg. 2008; 18(3): 140-9.

2. Löf Granström A, Wester T. Mortality in Swedish patients with Hirschsprung disease. Pediatr Surg Int. 2017; 33(11): 1177-81.
3. Bhatnagar SN. Hirschsprung's disease in newborns. J Neonatal Surg. 2013; 2(4): 51.
4. Butler Tjaden NE, Trainor PA. The developmental etiology and pathogenesis of Hirschsprung disease. Transl Res. 2013; 162(1): 1-15.
5. Núñez-Ramos R, Fernández RM, González-Velasco M, Ruiz-Contreras J, Galán-Gómez E, Núñez-Núñez R, et al. A Scoring system to predict the severity of Hirschsprung disease at diagnosis and its correlation with molecular genetics. Pediatr Dev Pathol. 2017; 20(1): 28-37.
6. Barshack I, Fridman E, Goldberg I, Chowers Y, Kopolovic J. The loss of calretinin expression indicates aganglionosis in Hirschsprung's disease. J Clin Pathol. 2004; 57(7): 712-6.
7. Harrison MW, Deitz DM, Campbell JR, Campbell TJ. Diagnosis and management of Hirschsprung's disease. A 25 year perspective. Am J Surg. 1986; 152(1): 49-56.
8. Menezes M, Corbally M, Puri P. Long-term results of bowel function after treatment for Hirschsprung's disease: a 29-year review. Pediatr Surg Int. 2006; 22(12): 987-90.
9. Dasgupta R, Langer JC. Hirschsprung disease. Curr Probl Surg. 2004;4 1(12): 942-88.
10. Webster W. Embryogenesis of the enteric ganglia in normal mice and in mice that develop congenital aganglionic megacolon. J Embryol Exp Morphol. 1973; 30(3): 573-85.
11. Gonzalo DH, Plesec T. Hirschsprung disease and use of calretinin in inadequate rectal suction biopsies. Arch Pathol Lab Med. 2013; 137(8): 1099-102.
12. Knowles CH, De Giorgio R, Kapur RP, Bruder E, Farrugia G, Geboes K, et al. Gastrointestinal neuromuscular pathology: guidelines for histological techniques and reporting on behalf of the Gastro 2009 International Working Group. Acta Neuropathol. 2009; 118(2): 271-301.
13. Qualman SJ, Jaffe R, Bove KE, Monforte-Muñoz H. Diagnosis of Hirschsprung disease using the rectal biopsy: multi-institutional survey. Pediatr Dev Pathol. 1999; 2(6): 588-96.
14. Santos MM, Tannuri U, Coelho MC. Study of acetylcholinesterase activity in rectal suction biopsy for diagnosis of intestinal dysganglionoses: 17-year experience of a single center. Pediatr Surg Int. 2008; 24(6): 715-9.
15. Moore SW, Johnson G. Acetylcholinesterase in Hirschsprung's disease. Pediatr Surg Int. 2005; 21(4): 255-63.
16. Rakhshani N, Araste M, Imanzade F, Panahi M, Tameshkel FS. Hirschsprung disease diagnosis: Calretinin marker role in determining the presence or absence of ganglion cells. Iran J Pathol. 2016 Fall; 11(4): 409-15.
17. Cinel L, Ceyran B, Güçlüer B. Calretinin immunohistochemistry for the diagnosis of Hirschprung disease in rectal biopsies. Pathol Res Pract. 2015; 211(1): 50-4.
18. Jiang M, Li K, Li S, Yang L, Yang D, Zhang X, et al. Calretinin, S100 and protein gene product 9.5 immunostaining of rectal suction biopsies in the diagnosis of Hirschsprung' disease. Am J Transl Res. 2016; 8(7): 3159-68.
19. Guinard-Samuel V, Bonnard A, De Lagausie P, Philippe-Chomette P, Alberti C, El Ghoneimi A, et al. Calretinin immunohistochemistry: a simple and efficient tool to diagnose Hirschsprung disease. Mod Pathol. 2009; 22(10): 1379-84.
20. de Haro Jorge I, Palazón Bellver P, Julia Masip V, Saura García L, Ribalta Farres T, Cuadras Pallejà D, et al. Effectiveness of calretinin and role of age in the diagnosis of Hirschsprung disease. Pediatr Surg Int. 2016; 32(8): 723-7.