Volume 11, Issue 4, April – 2026 International Journal of Innovative Science and Research Technology
ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/26apr1050

Hirschsprung Disease: Pathophysiology, Clinical Manifestations, Diagnostic Evaluation, and Nursing Management

Jisha Yohannan

Assistant Lecturer, T John College and School of Nursing
Bangalore.

Publication Date: 2026/04/27

Abstract: Hirschsprung disease (HD), also known as congenital aganglionic megacolon, is a developmental disorder of the enteric nervous system characterized by the congenital absence of parasympathetic ganglion cells in the myenteric (Auerbach) and submucosal (Meissner) plexuses of the distal colon and rectum. This absence results in functional intestinal obstruction and extreme proximal colonic dilatation. The recto-sigmoid colon is the most frequently affected segment. HD has a well-established predilection for the male sex. Clinical presentation varies significantly by age of onset: neonates typically exhibit delayed meconium passage, abdominal distention, and bilious vomiting, while older children present with chronic constipation, failure to thrive, and progressive abdominal distention. Diagnosis is confirmed through a combination of contrast enema studies, anorectal manometry, and definitive rectal biopsy demonstrating the absence of ganglion cells. Surgical management includes initial colostomy for intestinal decompression, followed by definitive resection of the aganglionic segment with primary anastomosis. Nursing assessment, perioperative care, and family education are central pillars of comprehensive management.

Keywords: *Hirschsprung Disease, Congenital Aganglionic Megacolon, Paediatric Nursing, Enteric Nervous System, Colostomy, Rectal Biopsy, Neonatal Obstruction.*

How to Cite: Jisha Yohannan (2026) Hirschsprung Disease: Pathophysiology, Clinical Manifestations, Diagnostic Evaluation, and Nursing Management. *International Journal of Innovative Science and Research Technology*, 11(4), 2084-2086. https://doi.org/10.38124/ijisrt/26apr1050

I. INTRODUCTION

Hirschsprung disease (HD) is a congenital anomaly of the large intestine first described in 1888 by the Danish physician Harald Hirschsprung. The condition is defined by the absence of intrinsic ganglion cells in the distal segment of the colon, arising from a failure of neural crest cell migration during embryogenesis. These ganglion cells, which form the enteric nervous system, are essential for coordinating peristaltic movement. Their absence leads to a permanently contracted, non-functioning bowel segment that obstructs normal faecal transit and causes massive dilatation of the proximal, normally innervated colon—a state referred to as megacolon.

HD represents one of the most common causes of neonatal intestinal obstruction, with an estimated incidence of approximately 1 in 5,000 live births. The disorder demonstrates a marked male predominance, with affected males outnumbering females at a ratio of approximately 4:1. Early recognition, prompt diagnostic evaluation, and timely surgical intervention are critical to preventing life-threatening complications, including enterocolitis and intestinal perforation. This paper provides a comprehensive clinical overview encompassing the definition, epidemiology, clinical features, diagnostic approach, surgical management, and nursing implications of Hirschsprung disease.

II. DEFINITION AND PATHOPHYSIOLOGY

Hirschsprung disease is defined as the congenital absence of parasympathetic ganglion cells in both the myenteric (Auerbach) plexus and the submucosal (Meissner) plexus of the distal colon and rectum. This aganglionic segment is unable to undergo normal peristalsis and remains in a state of tonic contraction. As a result, the proximal, ganglionated colon becomes obstructed and undergoes progressive dilatation, giving rise to the clinical picture of megacolon.

The pathophysiological basis of HD lies in the disruption of enteric neural crest cell (ENCC) migration. Normally, ENCCs migrate from the vagal region of the neural crest in a rostro-caudal direction to colonize the entire gut by

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/26apr1050

the twelfth week of gestation. A failure or premature arrest of this migration results in aganglionosis of the terminal bowel. The recto-sigmoid segment is affected in the majority of cases (approximately 75-80%), while total colonic and total intestinal aganglionosis represent less common but more severe variants.

III. EPIDEMIOLOGY AND INCIDENCE

HD occurs in approximately 1 per 5,000 live births globally. The condition is significantly more common in males than in females, a pattern most pronounced in the classic short-segment form. HD may occur as an isolated finding or in association with other congenital anomalies. Down syndrome (trisomy 21) is the most common syndromic association, occurring in approximately 2-10% of HD cases. Mutations in the RET proto-oncogene account for the majority of familial cases, and genetic counselling is recommended for affected families.

IV. CLINICAL MANIFESTATIONS

➢ *Presentation in Neonates*

In neonates, symptoms typically manifest at birth or within the first seven days of life. The hallmark early sign is failure to pass meconium within 48 hours of birth, which should raise strong clinical suspicion for HD. Additional neonatal features include:

- Delayed or absent meconium passage beyond 48 hours of birth
- Progressive abdominal distention secondary to proximal colonic dilatation
- Constipation as the dominant bowel symptom
- Feculent or bilious vomiting resulting from intestinal obstruction
- Intermittent diarrhoea and signs of dehydration in complicated presentations

➢ *Presentation in Older Children*

In children in whom the diagnosis is not established in the neonatal period, HD typically presents with chronic gastrointestinal symptoms:

- Chronic, refractory constipation resistant to dietary and pharmacological management
- Progressive abdominal distention with visible peristaltic waves
- Absence of the urge to defecate due to absent anorectal sensation
- Alternating constipation and diarrhoea, occasionally with overflow soiling
- Anorexia, abdominal discomfort, and irritability
- Gross malnutrition, iron deficiency anaemia, and failure to thrive

V. DIAGNOSTIC EVALUATION

The diagnosis of HD is established through a stepwise approach incorporating clinical assessment, radiological investigations, physiological testing, and histopathological confirmation. No single test is independently sufficient; diagnosis requires integration of multiple modalities.

➢ *Digital Rectal Examination*

Digital rectal examination reveals a tight, empty rectum in a neonate with abdominal distention. An explosive release of gas and meconium upon withdrawal of the examining finger ('squirt sign') is highly suggestive of HD, resulting from sudden decompression of the dilated proximal colon past the aganglionic segment.

➢ *Plain Abdominal Radiography*

Plain X-ray of the abdomen typically reveals distended loops of large bowel proximal to the aganglionic zone, absence of gas in the rectum, and in severe cases, air-fluid levels indicating functional obstruction. Plain radiography serves as a useful screening tool but is not diagnostic in isolation.

➢ *Barium (Contrast) Enema*

Contrast enema demonstrates a characteristic 'transition zone'—a funnel-shaped area between the narrow aganglionic distal segment and the dilated proximal colon—which is the pathognomonic radiological sign of HD. The study should be performed without prior bowel preparation to avoid distortion of the transition zone. A 24-hour delayed film showing retained contrast is an additional supportive finding.

➢ *Anorectal Manometry*

Anorectal manometry assesses the recto anal inhibitory reflex (RAIR), whereby rectal distension normally causes reflex relaxation of the internal anal sphincter. In HD, this inhibitory reflex is characteristically absent due to the lack of ganglion cells. The absence of RAIR is highly sensitive and specific for HD in children, though technically challenging in neonates.

➢ *Rectal Biopsy*

Suction rectal biopsy remains the gold standard for definitive diagnosis. Specimens are subjected to histochemical staining, and diagnosis is confirmed by the demonstrable absence of ganglion cells in the submucosal plexus, accompanied by hypertrophic nerve bundles and elevated acetylcholinesterase activity. This finding is pathognomonic of aganglionosis and guides the level of surgical resection.

VI. MANAGEMENT

➢ *Initial Surgical Management: Colostomy*

The primary objective of initial surgical management is intestinal decompression. A colostomy is created in the normally ganglionated portion of the colon proximal to the aganglionic segment. This procedure achieves three principal goals: decompression of the obstructed proximal colon, diversion of faecal material to allow preparation for definitive surgery, and restoration of adequate nutritional intake by relieving obstruction. Intraoperative frozen sections confirm the presence of ganglion cells at the colostomy site.

➢ *Definitive Surgical Repair*

Definitive management consists of resection of the entire aganglionic, non-functioning, and dilated bowel

ISSN No:-2456-2165 https://doi.org/10.38124/ijisrt/26apr1050

segment, followed by end-to-end anastomosis between the normally innervated proximal colon and the residual normal anorectum. Established operative techniques include the Swenson, Duhamel, and Soave pull-through procedures. In contemporary paediatric surgery, single-stage laparoscopic-assisted pull-through without a preliminary colostomy is increasingly performed in appropriately selected neonates and infants.

VII. NURSING ASSESSMENT AND MANAGEMENT

➢ *Comprehensive Nursing Assessment*

Thorough nursing assessment forms the cornerstone of holistic care in HD. A detailed history addressing bowel habits, dietary intake, and associated clinical problems is essential. Key assessment parameters include:

- Neonatal period: Timing of first meconium passage, constipation characteristics, stool consistency and frequency, degree of abdominal distention, and feeding tolerance
- Older children: Chronicity and pattern of constipation, evidence of failure to thrive, growth parameters, signs of malnutrition and anaemia, and abdominal examination findings
- Family history: Genetic predisposition, prior surgical history, and psychosocial context

➢ *Perioperative Nursing Care*

Perioperative nursing encompasses preoperative preparation (bowel irrigation, intravenous fluid management, prophylactic antibiotics), intraoperative monitoring, and postoperative care. Postoperative nursing responsibilities include wound assessment, stoma care education, monitoring for complications such as anastomotic leak or Hirschsprung-associated enterocolitis (HAEC), and documentation of return of bowel function. Pain assessment and management are integral throughout the perioperative period.

➢ *Family Education and Discharge Planning*

Comprehensive family education is an indispensable component of nursing management. Nurses provide structured teaching on stoma care and appliance management, dietary guidance, early identification of HAEC signs (fever, abdominal distention, explosive diarrhea), and long-term follow-up expectations. Psychological support for both child and family—particularly regarding adjustment to a stoma and future surgical procedures—should be incorporated throughout the care continuum.

VIII. CONCLUSION

Hirschsprung disease is a significant congenital disorder of the enteric nervous system demanding a coordinated multidisciplinary approach for optimal outcomes. Early clinical recognition—particularly in neonates presenting with delayed meconium passage and abdominal distention—is essential to initiating timely diagnostic evaluation. Histopathological confirmation via suction rectal biopsy remains the diagnostic standard, and surgical management culminating in definitive resection with anastomosis is curative in the majority of patients.

Nursing professionals occupy a critical role at every phase of the HD care pathway: from initial clinical assessment through perioperative management to long-term family education. A thorough understanding of the pathophysiology, clinical spectrum, and management principles of HD equips nursing practitioners to deliver high-quality, evidence-based, and compassionate care to affected children and their families.

REFERENCES

[1]. Amiel, J., Sproat-Emison, E., Garcia-Barcelo, M., Lantieri, F., Burzynski, G., Borrego, S., & Lyonnet, S. (2008). Hirschsprung disease, associated syndromes and genetics: A review. Journal of Medical Genetics, 45(1), 1-14.
[2]. Burkardt, D. D., Graham, J. M., Short, S. S., & Frykman, P. K. (2014). Advances in Hirschsprung disease genetics and treatment strategies. Clinical Pediatrics, 53(1), 71-81.
[3]. Langer, J. C. (2013). Hirschsprung disease. Current Opinion in Pediatrics, 25(3), 368-374.
[4]. Moore, S. W. (2016). Hirschsprung disease: Current perspectives. Open Access Surgery, 9, 39-50.
[5]. Pini Prato, A., Gentilino, V., Giunta, C., Avanzini, S., Parodi, S., Mattioli, G., & Gandullia, P. (2013). Hirschsprung disease: Do risk factors of poor surgical outcome exist? Journal of Pediatric Surgery, 48(1), 197-205.
[6]. Swenson, O. (2002). Hirschsprung's disease: A review. Pediatrics, 109(5), 914-918.
[7]. Taguchi, T., Leerapun, T., & Ohshiro, H. (2017). Surgical treatment of Hirschsprung disease. Surgery Today, 47(3), 266-272.