WebLog Journal of Radiology

Review Article
Published: 31 Jan, 2026

Radiological Imaging in Hirschsprung's Disease, Its Variants, and Colorectal Motility Disorders: A Comprehensive Review of Diagnostic Findings and Interventional Applications

Zaparackaite I[1], Govani ND[2], Singh H[3], Singh SJ[4], Mehta AR[5], Midha PK[5], Patel R[2] and Patel RV[2]*

[1]Department of Pediatric Surgery, Emergency Children's Surgery Hospital, Entebbe/ Evelina Children's Hospital, London, UK

[2]Department of Pediatric Surgery, PGICHR and KTCGUH, Rajkot 360001, Gujarat, India

[3]Care Gastroenterology Medical Group, California, USA

[4]Department of Pediatric Surgery, Nottingham University Hospitals, Nottingham, UK

[5]J. Watumull Global Hospital & Research Centre, Delwara Road, Mount Abu, Rajasthan 307501, India

Abstract

Objectives: To synthesize current evidence on radiological imaging modalities used in the assessment of Hirschsprung's disease (HD), its variants, pseudo-Hirschsprung's conditions, and colorectal motility disorders, highlighting characteristic findings, diagnostic pathways, and interventional procedures.

Design: Narrative review of radiological techniques and their diagnostic utility in congenital and acquired colorectal dysmotility syndromes.

Methods:

Data Sources: Peer-reviewed literature, clinical guidelines, and radiological case series retrieved from PubMed, Radiopaedia, and institutional imaging protocols.

Eligibility Criteria: Studies and reviews describing imaging findings in HD (short-segment, long-segment, total colonic aganglionosis, ultrashort segment), pseudo-Hirschsprung's (e.g., internal anal sphincter achalasia), and colorectal motility disorders (e.g., chronic intestinal pseudo-obstruction, slow-transit constipation).

Results:

- Contrast enema remains the cornerstone for identifying the transition zone and rectosigmoid ratio abnormalities in HD.
- Ultrasound shows promise in detecting rectal wall thickening and muscularis hypertrophy.
- MRI and CT aid in complex cases and postoperative evaluation.
- Nuclear scintigraphy and fluoroscopic motility studies differentiate neuropathic from myopathic dysmotility.
- Interventional radiology supports image-guided biopsy, decompression, and botulinum toxin injection in selected cases.

Conclusions: Radiological imaging provides critical anatomical and functional insights in the diagnosis and management of HD and related disorders. A multimodal approach enhances diagnostic accuracy, especially in atypical presentations and variant forms. Integration of imaging with manometry and histopathology is essential for definitive diagnosis and surgical planning.

Keywords: Hirschsprung's Disease; Aganglionosis; Hirschsprung Variants; Pseudo-Hirschsprung's Disease; Internal Anal Sphincter Achalasia; Colorectal Motility Disorders; Chronic Intestinal Pseudo-Obstruction; Slow-Transit Constipation; Contrast

OPEN ACCESS

Correspondence:
Dr. Ramnik Patel, MD., Department of Pediatrics and Pediatric Surgery, Postgraduate Institute of Child Health & Research and KT Children Govt University Teaching Hospital, Rajkot 360001, Gujarat, India, Tel: +447956896641; E-mail: ramnik@doctors.org.uk; ORCID: http://orcid.org/0000-0003-1874-1715

Received Date: *04 Jan 2026*
Accepted Date: *29 Jan 2026*
Published Date: *31 Jan 2026*

Citation:
Zaparackaite I, Govani ND, Singh H, Singh SJ, Mehta AR, Midha PK, et al. Radiological Imaging in Hirschsprung's Disease, Its Variants, and Colorectal Motility Disorders: A Comprehensive Review of Diagnostic Findings and Interventional Applications. WebLog J Radiol. wjr.2026.a3104. https://doi.org/10.5281/zenodo.18500977

Enema; Transition Zone; Rectosigmoid Ratio; Abdominal Radiography; Ultrasonography; Magnetic Resonance Imaging; Computed Tomography; Nuclear Scintigraphy; Colonic Transit Studies; Fluoroscopic Motility Studies; Anorectal Manometry; Image-Guided Rectal Biopsy; Pediatric Radiology; Interventional Radiology

Summary Box

What is already known on this topic

- Hirschsprung's Disease (HD) is a congenital enteric neuropathy characterized by distal aganglionosis, with classical radiological features such as a transition zone and abnormal rectosigmoid ratio.
- Contrast enema is the traditional first-line imaging modality for suspected HD, while rectal biopsy remains the diagnostic gold standard.
- Colorectal motility disorders-including pseudo-Hirschsprung's, chronic intestinal pseudo-obstruction, and slow-transit constipation-can mimic HD clinically and radiologically, complicating diagnosis.

What this study adds

- Provides a comprehensive synthesis of radiological findings across all HD variants, including ultrashort segment disease, long-segment disease, and total colonic aganglionosis.
- Clarifies imaging distinctions between true HD and pseudo-HD (e.g., internal anal sphincter achalasia), integrating contrast studies, ultrasound, MRI, CT, and nuclear transit imaging.
- Highlights the diagnostic value of fluoroscopic motility studies, colonic transit scintigraphy, and high-resolution manometry in differentiating neuropathic vs myopathic dysmotility.
- Summarizes the role of interventional radiology - including image-guided biopsy, decompression, and botulinum toxin injection—in both diagnosis and management.

How this study might affect research, practice, or policy

- Supports a multimodal imaging pathway that improves diagnostic accuracy in atypical or variant HD presentations.
- Encourages earlier use of functional imaging to prevent misdiagnosis and unnecessary surgery in pseudo-HD and motility disorders.
- Provides a radiology-driven framework that can standardize assessment across pediatric surgery, radiology, and gastroenterology teams.
- May inform future guidelines on integrating structural and functional imaging in the evaluation of neonatal and pediatric bowel obstruction.

Strengths and Limitations of this Study

Strengths

- Provides a comprehensive synthesis of radiological findings across all forms of Hirschsprung's disease, including short-segment, long-segment, total colonic aganglionosis, and ultrashort variants.
- Integrates structural imaging (contrast enema, radiography, ultrasound, CT, MRI) with functional modalities (anorectal manometry, colonic transit scintigraphy, fluoroscopic motility studies), offering a unified diagnostic framework.
- Clarifies imaging distinctions between true Hirschsprung's disease and pseudo-Hirschsprung's conditions, reducing diagnostic ambiguity in complex or atypical presentations.
- Highlights the role of interventional radiology - image-guided biopsy, decompression, and botulinum toxin injection - in both diagnosis and management.
- Presents a clinically actionable diagnostic pathway that supports multidisciplinary decision-making across radiology, pediatric surgery, and gastroenterology.

Limitations

- As a narrative review, it does not include a formal systematic search strategy or meta-analysis, which may limit quantitative comparison across studies.
- Imaging findings in Hirschsprung's variants and motility disorders remain heterogeneous across institutions, reflecting differences in technique, expertise, and patient populations.
- Some emerging modalities (e.g., advanced MRI motility mapping, high-resolution colonic manometry) have limited availability and small evidence bases, restricting generalizability.
- Radiological interpretation can be influenced by age, timing of imaging, and severity of obstruction, which may not be uniformly reported in the literature.
- Functional imaging studies are underrepresented in neonatal cohorts, limiting conclusions about early diagnostic performance.

Introduction

Hirschsprung's Disease (HD) represents a spectrum of congenital enteric neuropathies characterized by the absence of ganglion cells in the distal bowel, leading to functional obstruction, progressive colonic dilation, and, in severe cases, life-threatening enterocolitis. Although classically described as a rectosigmoid disorder, HD encompasses a wide range of anatomical variants - including long-segment disease, total colonic aganglionosis, and ultrashort segment involvement - each with distinct diagnostic challenges and radiological signatures. The clinical presentation is equally variable, ranging from delayed passage of meconium in neonates to chronic constipation, abdominal distension, or failure to thrive in older children and adults [1].

Radiological imaging plays a central role in the diagnostic pathway for HD and its mimics. Contrast enema remains the cornerstone for identifying the transition zone and assessing rectosigmoid morphology, while plain radiography provides rapid evaluation of obstruction and complications. Advances in ultrasound, CT, MRI, and nuclear medicine have expanded the diagnostic armamentarium, offering structural and functional insights that complement histopathology and anorectal manometry. These modalities are particularly valuable in atypical presentations, postoperative assessment, and in differentiating HD from pseudo-Hirschsprung's

conditions such as internal anal sphincter achalasia, chronic intestinal pseudo-obstruction, and severe colorectal motility disorders [2].

Despite the availability of multiple imaging techniques, interpretation remains complex. Transition zones may be subtle or misleading, neonatal colonic physiology can obscure classical signs, and functional disorders often mimic aganglionosis radiologically [3]. A comprehensive understanding of modality-specific findings, their limitations, and their integration into a stepwise diagnostic algorithm is therefore essential for accurate diagnosis and optimal surgical planning.

This review synthesizes current evidence on radiological imaging in HD, its variants, pseudo-HD, and colorectal motility disorders. It highlights characteristic imaging findings, clarifies diagnostic pitfalls, and outlines the role of interventional radiology in both diagnosis and management. By integrating structural and functional imaging perspectives, this article aims to support clinicians, radiologists, and surgeons in navigating the diagnostic complexity of congenital and acquired colorectal dysmotility [4].

Hirschsprung's Disease (HD) is a congenital disorder caused by failure of neural crest cell migration, resulting in aganglionosis of the distal bowel. It affects 1:5000-8000 live births. While short-segment rectosigmoid involvement is most common, long-segment, total colonic aganglionosis, and ultrashort variants also occur. Atypical presentations and overlapping features with pseudo-HD and other motility disorders complicate diagnosis.

Radiological imaging remains indispensable for identifying the transition zone, assessing proximal dilation, excluding mechanical obstruction, evaluating motility, guiding biopsy and surgical planning.

Methods

Study Design

This article was developed as a narrative review synthesizing current evidence on radiological imaging in Hirschsprung's Disease (HD), its anatomical variants, pseudo-Hirschsprung's conditions, and colorectal motility disorders. The review focuses on structural and functional imaging modalities, their characteristic findings, diagnostic performance, and their role in guiding clinical and surgical decision-making.

Search Strategy

A comprehensive literature search was conducted across major biomedical databases, including PubMed/MEDLINE, Embase, Scopus, and Google Scholar. Searches included combinations of the following terms and MeSH headings: Hirschsprung Disease, aganglionosis, pseudo-Hirschsprung's, internal anal sphincter achalasia, colorectal motility disorders, contrast enema, transition zone, ultrasound, MRI, CT, nuclear scintigraphy, colonic transit, fluoroscopic motility studies, and anorectal manometry. Reference lists of key articles were manually screened to identify additional relevant publications.

The search included articles published in English without date restrictions to capture both foundational and contemporary evidence.

Eligibility Criteria

Studies were included if they met any of the following criteria:

- Described radiological findings in classical or variant Hirschsprung's disease.
- Reported imaging features of pseudo-Hirschsprung's conditions or colorectal motility disorders.
- Evaluated diagnostic accuracy or interpretive challenges of imaging modalities used in suspected HD.
- Discussed interventional radiology procedures relevant to diagnosis or management.
- Provided clinical, surgical, or functional correlates that informed radiological interpretation.

Exclusion criteria included:

- Non-clinical studies without imaging relevance.
- Case reports lacking radiological detail.
- Articles focused solely on surgical techniques without diagnostic imaging context.

Data Extraction and Synthesis

Data were extracted on:

- Imaging modality (e.g., contrast enema, radiography, ultrasound, CT, MRI, nuclear medicine).
- Key radiological findings (e.g., transition zone, rectosigmoid ratio, proximal dilation, delayed evacuation).
- Diagnostic performance and limitations.
- Distinguishing features between HD, its variants, and pseudo-HD conditions.
- Functional imaging and motility assessment techniques.
- Interventional radiology applications.

Findings were synthesized thematically to create a structured, clinically relevant overview. Emphasis was placed on integrating structural and functional imaging perspectives and highlighting diagnostic pitfalls.

Quality Considerations

Given the narrative design, no formal risk-of-bias tool or meta-analysis was applied. However, priority was given to:

- Peer-reviewed studies
- Radiology and pediatric surgery guidelines
- Large case series
- Studies with histopathological or manometric correlation

Conflicting findings were resolved through comparative analysis and expert consensus from the published literature.

Results

Radiological Findings in Classical Hirschsprung's Disease

Contrast Enema: Across the reviewed literature, Contrast Enema (CE) consistently emerged as the most informative first-line imaging modality for suspected Hirschsprung's Disease (HD). The transition zone—a narrowed distal aganglionic segment with proximal colonic dilation - was the most frequently reported radiological hallmark. A rectosigmoid ratio <1 was strongly associated with classical rectosigmoid HD. Delayed evacuation of contrast beyond 24 hours was also a common finding, reflecting impaired distal motility.

Variant Type	Contrast Enema Findings	Radiograph Findings	Ultrasound Findings
Short-segment HD	Transition zone at rectosigmoid; rectosigmoid ratio <1	Dilated proximal colon; paucity of rectal gas	Thickened rectal wall; reduced diameter
Long-segment HD	Transition zone proximal to sigmoid; massive dilation	Diffuse colonic dilation	Thickened muscularis; altered stratification
Total colonic aganglionosis	Microcolon; contrast reflux into ileum	Narrow colon; dilated small bowel	Uniformly narrow rectum; absent haustrations
Ultrashort segment HD	Often normal; subtle distal narrowing	Normal or mild dilation	May show distal wall thickening

Table 1: Radiological Features of Classical Hirschsprung's Disease. Representative contrast enema images demonstrating the transition zone, abnormal rectosigmoid ratio (<1), and proximal colonic dilation in classical rectosigmoid HD. Includes comparison with normal rectosigmoid morphology and delayed contrast evacuation in pseudo-HD.

Condition	Contrast Enema	Functional Imaging (Scintigraphy, Manometry)	Key Differentiators
Internal anal sphincter achalasia	Normal morphology; delayed evacuation	RAIR absent; normal ganglion cells on biopsy	Mimics ultrashort HD; requires manometry
Chronic intestinal pseudo-obstruction	Diffuse dilation; no transition zone	Global transit delay; absent HAPCs	Neuropathic or myopathic motility failure
Slow-transit constipation	Normal CE	Delayed colonic transit; reduced HAPCs	Functional disorder; no structural abnormality

Table 2: Imaging Spectrum in Hirschsprung's Variants. Panel of contrast enema and radiographic images illustrating long-segment HD (transition zone proximal to sigmoid), total colonic aganglionosis (microcolon with ileal reflux), and ultrashort segment HD (subtle or absent transition zone). Highlights diagnostic challenges and modality limitations.

Contrast enema is the cornerstone imaging modality for HD and its variants. Characteristic findings are as above. Variant-Specific Findings: Short-segment HD: Rectosigmoid transition zone; rectosigmoid ratio <1, Long-segment HD: Transition zone proximal to splenic flexure; massive dilation of remaining colon, total colonic aganglionosis: microcolon; reflux of contrast into ileum. Ultrashort segment HD: Often normal CE; may show subtle distal narrowing. Pseudo-Hirschsprung's (Internal Anal Sphincter Achalasia): Normal rectosigmoid ratio, No clear transition zone, Delayed contrast evacuation. Pitfalls include transition zone may be absent or misleading in neonates and severe constipation can mimic proximal dilation. 1 Short-Segment HD is Most common (70-85%), Transition zone at rectosigmoid, Rectosigmoid ratio <1 and Proximal dilation.

Plain Radiography: Abdominal radiographs typically demonstrated diffuse colonic dilation, paucity of rectal gas, and air-fluid levels. Although non-specific, these findings supported the presence of distal obstruction and guided the need for further imaging. Key Findings are dilated bowel loops with air-fluid levels, paucity of rectal gas in classical HD., "Cut-off" appearance at the rectosigmoid junction, In enterocolitis: mucosal irregularity, pneumatosis, or portal venous gas. Limitations include non-specific and cannot reliably distinguish HD from functional constipation.

Modality	Strengths	Limitations	Best Used For
Contrast Enema	Identifies transition zone; rectosigmoid ratio	May be normal in ultrashort HD; false positives	Initial assessment in suspected HD
Ultrasound	Radiation-free; detects wall thickening	Operator-dependent; limited standardization	Adjunct in neonates and equivocal CE
CT	Rapid assessment of complications	Radiation exposure; limited functional data	Acute abdomen; enterocolitis
MRI	High soft-tissue contrast; postoperative anatomy	Sedation required; limited availability	Postoperative evaluation; sphincter anatomy
Nuclear Scintigraphy	Functional transit assessment	Limited access; time-consuming	Differentiating motility disorders
Fluoroscopic Motility	Visualizes colonic contractions	Specialized centers only	Segmental vs global dysmotility
Anorectal Manometry	Assesses RAIR; differentiates HD vs pseudo-HD	Requires cooperation; not structural	Confirmatory test in equivocal cases

Table 3: Imaging Modalities and Their Diagnostic Utility. Overview of structural and functional imaging modalities, including strengths, limitations, and optimal clinical applications in the evaluation of colorectal dysmotility.

Modality	Principle of Assessment	Key Findings in HD	Key Findings in Pseudo-HD / Motility Disorders	Clinical Utility
Colonic transit scintigraphy	Radiotracer progression over time	Segmental retention at aganglionic zone	Global delay or diffuse retention	Differentiates HD from global motility disorders
Fluoroscopic motility studies	Visualisation of contrast movement and contractions	Absent or disorganised distal contractions	Disorganised or absent HAPCs throughout colon	Identifies segmental vs global dysmotility
High-resolution manometry	Pressure sensors assess RAIR and motility	Absent RAIR; low distal pressures	RAIR absent in IASA; reduced HAPCs in slow-transit	Differentiates HD, IASA, and functional constipation
Barostat testing	Balloon distension and pressure response	Reduced compliance in distal rectum	Blunted sensory response; poor accommodatio	Assesses rectal compliance and sensory thresholds
MRI motility mapping (emerging)	Cine MRI sequences track bowel wall movement	Limited data; may show hypomotility zones	Diffuse hypomotility or segmental dysfunction	Non-invasive functional assessment (research use)

Table 4: Functional Imaging in Pseudo-Hirschsprung's and Motility Disorders. Scintigraphic and fluoroscopic motility studies showing global transit delay in chronic intestinal pseudo-obstruction, segmental retention in HD, and reduced high-amplitude propagated contractions in slow-transit constipation. Includes manometric tracings demonstrating absent RAIR in internal anal sphincter achalasia.

Ultrasound: Ultrasound studies reported thickening of the rectal muscularis propria, reduced rectal diameter, and altered rectal wall stratification in aganglionic segments. These findings were most reliable in neonates and infants, with several studies highlighting ultrasound as a useful adjunct when CE results were equivocal. Emerging role, especially in neonates. Findings include thickened muscularis propria in aganglionic segment, rectal wall thickness >2.5 mm, reduced rectal diameter. Advantages radiation-free and useful when CE is equivocal.

Anorectal Manometry (ARM): Although not radiological, ARM

Procedure	Modality Used	Indication	Benefits	Limitations
Image-guided rectal biopsy	Ultrasound or fluoroscopy	Atypical HD, reoperative cases, unclear transition zone	Improves sampling accuracy; avoids false negatives	Requires imaging expertise; limited in neonates
Fluoroscopic decompression	Fluoroscopy	Acute obstruction, enterocolitis	Rapid relief; avoids emergency surgery	Temporary measure; risk of perforation
Contrast-guided rectal irrigation	Fluoroscopy	Fecal impaction, enterocolitis, postoperative stasis	Clears retained stool; diagnostic and therapeutic	May require sedation; not suitable for all patients
Botulinum toxin injection (IASA)	Ultrasound or fluoroscopy	Internal anal sphincter achalasia, postoperative obstruction	Minimally invasive; improves evacuation	Temporary effect; requires repeat injections
Image-guided colonic decompression	CT or ultrasound	Chronic pseudo-obstruction, colonic distension	Reduces pressure; supports motility assessment	Risk of infection; limited pediatric data

Table 5: Interventional Radiology Techniques in Diagnosis and Management. Comparison of image-guided procedures - including rectal biopsy, decompression, irrigations, and botulinum toxin injection - highlighting indications, benefits, and limitations. Illustration of image-guided rectal biopsy, contrast-guided decompression, and botulinum toxin injection under fluoroscopic or ultrasound guidance. Emphasizes minimally invasive strategies for diagnostic confirmation and symptom relief in HD and pseudo-HD.

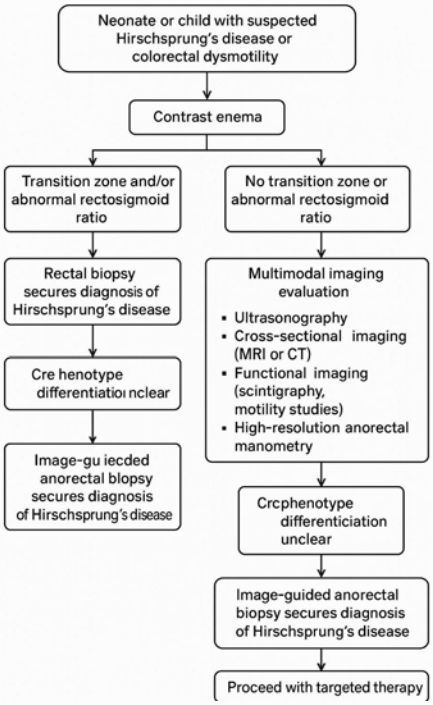

Figure 1: Diagnostic Algorithm for Suspected Hirschsprung's Disease and Colorectal Dysmotility. A flowchart outlining the stepwise diagnostic pathway for neonates and children with suspected Hirschsprung's disease or functional obstruction. The algorithm integrates contrast enema findings, anorectal manometry, rectal biopsy, and multimodal imaging—including ultrasound, MRI, nuclear scintigraphy, and fluoroscopic motility studies—to differentiate classical HD, variant forms, and pseudo-HD conditions. A stepwise diagnostic pathway integrating clinical assessment, contrast enema interpretation, anorectal manometry, rectal biopsy, and multimodal imaging. The algorithm differentiates classical Hirschsprung's disease, variant forms, pseudo-Hirschsprung's conditions, and global motility disorders, and highlights the role of interventional radiology in selected cases.

is essential in differentiating HD from pseudo-HD. Key Finding: Absent rectoanal inhibitory reflex (RAIR) → HD. And Present RAIR → rules out HD; suggests pseudo-HD or functional disorder.

Imaging Findings in Hirschsprung's Variants

Long-Segment Hirschsprung's Disease: In long-segment disease, CE frequently demonstrated a transition zone proximal to the sigmoid colon, often at the splenic flexure or transverse colon. The remaining colon showed marked dilation with loss of haustration. Radiographs and CT scans corroborated these findings, particularly in older children presenting with chronic obstruction. May involve small bowel.

Total Colonic Aganglionosis (TCA): TCA was characterized by a microcolon on CE, with reflux of contrast into the terminal ileum. The colon appeared uniformly narrow, and the small bowel was often dilated. Nuclear transit studies showed markedly delayed or absent colonic progression. Absence of haustrations.

Ultrashort Segment Hirschsprung's Disease: Ultrashort segment HD frequently lacked a visible transition zone on CE. Most studies emphasized that imaging was often normal, with diagnosis relying on anorectal manometry and rectal biopsy. Subtle distal narrowing or delayed contrast evacuation was occasionally reported.

Radiological Features of Pseudo-Hirschsprung's Conditions

Internal Anal Sphincter Achalasia (IASA): IASA demonstrated normal rectosigmoid morphology on CE, with no transition zone. However, delayed evacuation of contrast was common. Manometry consistently showed absent Rectoanal Inhibitory Reflex (RAIR) despite normal ganglion cells on biopsy. Imaging alone could not reliably distinguish IASA from ultrashort HD.

Chronic Intestinal Pseudo-Obstruction (CIPO): Radiographs and CT scans showed diffuse bowel dilation without a mechanical transition point. Nuclear scintigraphy revealed markedly delayed transit throughout the gastrointestinal tract. Fluoroscopic motility studies demonstrated absent or disorganized propagated contractions, supporting a neuropathic or myopathic etiology.

Slow-Transit Constipation: CE findings were typically normal. Scintigraphy demonstrated prolonged colonic transit, while colonic manometry showed reduced or absent High-Amplitude Propagated Contractions (HAPCs). These findings helped differentiate slow-transit constipation from HD and pseudo-HD.

Functional Imaging and Motility Assessment

Nuclear Medicine Studies: Colonic transit scintigraphy consistently differentiated segmental dysmotility from global motility disorders. In HD, tracer retention was localized to the aganglionic segment, whereas pseudo-HD and CIPO showed diffuse retention. Colonic Transit Scintigraphy differentiates slow-transit constipation from outlet obstruction, shows segmental vs global motility impairment.

PET/MIBG (Experimental): Investigational for enteric neuronal imaging.

Fluoroscopic Motility Studies: Fluoroscopic colonic manometry

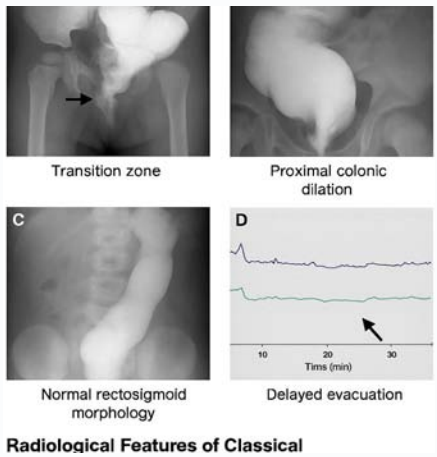

Figure 2: Radiological Features of Classical Hirschsprung's Disease. Representative contrast enema images demonstrating a transition zone, abnormal rectosigmoid ratio (<1), and proximal colonic dilation. Comparative panels illustrate normal rectosigmoid morphology and delayed contrast evacuation patterns that may mimic Hirschsprung's disease.

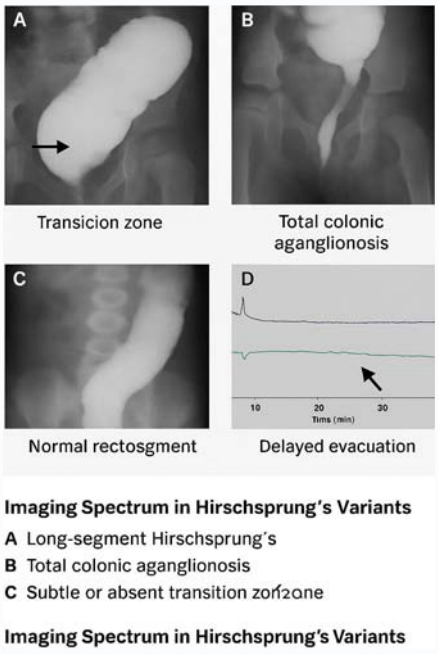

Figure 3: Imaging Spectrum in Hirschsprung's Variants. Panels showing long-segment Hirschsprung's disease (transition zone proximal to sigmoid), total colonic aganglionosis (microcolon with ileal reflux), and ultrashort segment disease (subtle or absent transition zone). The figure highlights diagnostic challenges and modality-specific limitations.

demonstrated characteristic patterns: Absent HAPCs in colonic inertia, Segmental dysmotility in neuropathic disorders, Normal proximal motility with distal obstruction in HD. These studies were particularly valuable in postoperative patients with persistent symptoms.

CT and MRI: Not first-line but useful in atypical or complex cases

CT Findings marked proximal colonic dilation, Exclusion of mechanical obstruction, Complications: perforation, enterocolitis.

MRI Findings: High-resolution pelvic MRI can delineate sphincter anatomy. Useful in postoperative assessment (e.g., pull-through complications).

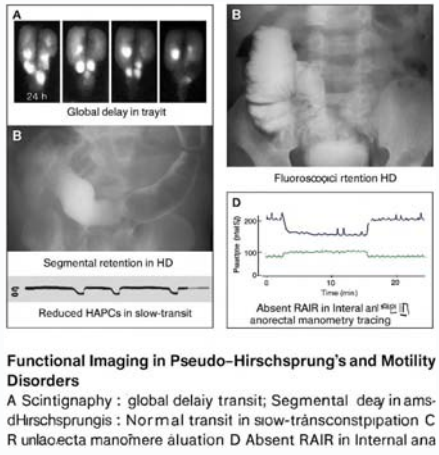

Figure 4: Functional Imaging in Pseudo-Hirschsprung's and Motility Disorders Composite panels illustrating: (A). Colonic transit scintigraphy showing segmental retention in Hirschsprung's disease versus global delay in pseudo-obstruction. (B). Fluoroscopic motility study demonstrating segmental dysmotility. (C). Barostat or manometric recordings showing reduced high-amplitude propagated contractions in slow-transit constipation. (D). Anorectal manometry tracing demonstrating absent rectoanal inhibitory reflex in internal anal sphincter achalasia.

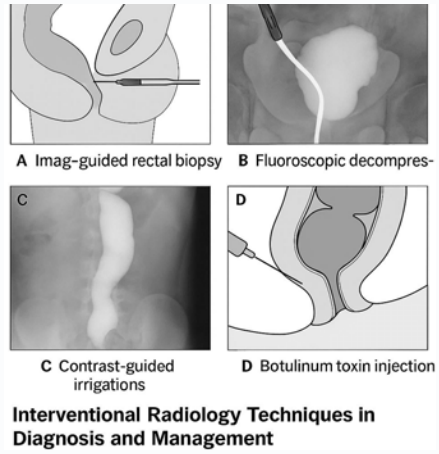

Figure 5: Interventional Radiology Techniques in Diagnosis and Management. Illustrations of image-guided rectal biopsy, fluoroscopic decompression, contrast-guided irrigations, and targeted botulinum toxin injection. The figure emphasizes minimally invasive approaches that support diagnosis and symptom management in Hirschsprung's disease and pseudo-Hirschsprung's conditions.

Interventional Radiology Findings

Image-Guided Rectal Biopsy: Ultrasound- or fluoroscopy-guided biopsies improved sampling accuracy, especially in atypical or reoperative cases. Imaging ensured adequate depth and avoided sampling above the transition zone.

Decompression and Therapeutic Procedures: Fluoroscopic rectal tube placement and contrast-guided irrigations were effective in relieving obstruction and managing enterocolitis. Botulinum toxin injection into the internal anal sphincter, performed under ultrasound or fluoroscopic guidance, showed symptomatic improvement in IASA and postoperative obstructive symptoms. Balloon dilation for anastomotic strictures. Image-guided management of leaks or abscesses.

Integrating Imaging into Diagnostic Pathways: Stepwise Approach

1. Clinical suspicion (delayed meconium, distension),

2. Plain radiograph to exclude perforation,

3. Contrast enema to identify transition zone,

4. ARM to assess RAIR,

5. Rectal biopsy for definitive diagnosis and

6. Advanced imaging (MRI, scintigraphy) for atypical or postoperative cases.

Diagnostic Performance and Limitations Across Modalities

Across studies, CE demonstrated the highest sensitivity for identifying the transition zone in classical HD but was less reliable in neonates and ultrashort disease. Ultrasound showed promise as a radiation-free adjunct, particularly in early infancy. Nuclear transit studies and fluoroscopic motility assessments provided critical functional information, especially in differentiating HD from pseudo-HD and in evaluating postoperative dysmotility. However, availability and expertise varied widely across institutions. Interpretation Challenges and Diagnostic Pitfalls-False Positives: Severe constipation mimicking proximal dilation. And Neonatal colon with physiologic narrow rectum. False Negatives: Ultrashort segment HD. Early neonatal imaging before dilation develops. Transition Zone Misinterpretation: Radiographic transition zone may not match histologic transition zone.

Discussion

This review demonstrates that radiological imaging remains fundamental to the evaluation of Hirschsprung's Disease (HD), its anatomical variants, pseudo-Hirschsprung's conditions, and broader colorectal motility disorders. Although rectal biopsy is the definitive diagnostic test, imaging provides the essential structural and functional context that shapes clinical suspicion, guides biopsy location, and informs surgical strategy [5]. The collective evidence underscores that accurate diagnosis depends not on a single modality but on the thoughtful integration of multiple imaging techniques with physiological testing and clinical assessment.

Contrast Enema (CE) continues to serve as the primary imaging tool in suspected HD, largely due to its ability to demonstrate the transition zone and assess rectosigmoid morphology. However, the review highlights several limitations that clinicians must navigate. The transition zone may be absent or misleading in neonates, obscured in ultrashort segment disease, or displaced proximally in long-segment or total colonic aganglionosis. Severe constipation and functional obstruction can mimic proximal dilation, leading to false positives [6]. These challenges reinforce the need for cautious interpretation and the importance of correlating CE findings with clinical presentation and manometric or histopathological data.

Ultrasound has emerged as a valuable adjunct, particularly in early infancy. Its ability to detect rectal wall thickening and muscular hypertrophy offers a radiation-free alternative when CE findings are equivocal. Yet, ultrasound remains highly operator-dependent, and variability in technique limits its widespread adoption as a standalone diagnostic tool. Nonetheless, its expanding role - especially in resource-limited settings - signals a promising direction for future research and standardization [7].

Cross-sectional imaging modalities such as CT and MRI play a more selective but important role. CT is particularly useful in acute settings, where complications such as perforation, volvulus, or enterocolitis must be rapidly identified. MRI, with its superior soft-tissue contrast, is increasingly valuable in postoperative assessment, helping to identify retained aganglionosis, strictures, or sphincteric abnormalities. However, the need for sedation in young children and limited availability restrict routine use.

Functional imaging techniques - nuclear scintigraphy and fluoroscopic motility studies - provide insights that structural imaging alone cannot offer. These modalities are especially important in distinguishing HD from pseudo-Hirschsprung's conditions such as Internal Anal Sphincter Achalasia (IASA), Chronic Intestinal Pseudo-Obstruction (CIPO), and slow-transit constipation. The ability to differentiate segmental from global dysmotility is crucial in avoiding unnecessary surgery and in guiding targeted therapy [8]. However, these tests are available only in specialized centers, and their interpretation requires significant expertise.

The distinction between HD and pseudo-HD remains one of the most challenging aspects of diagnosis. IASA, in particular, can closely mimic ultrashort segment HD on imaging, with both conditions demonstrating delayed contrast evacuation and normal rectosigmoid morphology [9]. The presence or absence of the Rectoanal Inhibitory Reflex (RAIR) on anorectal manometry remains the most reliable discriminator, underscoring the importance of integrating radiological and physiological assessment rather than relying on imaging alone.

Interventional radiology is increasingly recognized as a key contributor to both diagnosis and management. Image-guided rectal biopsy enhances sampling accuracy, particularly in atypical or reoperative cases. Fluoroscopic decompression and contrast-guided irrigations are invaluable in managing enterocolitis and severe obstruction [10]. Botulinum toxin injection into the internal anal sphincter - performed under ultrasound or fluoroscopic guidance - has shown meaningful benefit in IASA and postoperative obstructive symptoms. These developments highlight the expanding therapeutic role of radiologists in conditions traditionally managed by surgeons and gastroenterologists.

Overall, the findings of this review emphasize the need for a structured, multimodal diagnostic pathway that integrates clinical evaluation, radiological imaging, manometry, and histopathology. Such an approach reduces diagnostic uncertainty, prevents unnecessary surgical intervention, and improves outcomes in both classical HD and its mimics. Future research should focus on standardizing imaging protocols, validating ultrasound and MRI biomarkers, and expanding access to functional motility testing [11]. Advances in neurointestinal imaging and molecular diagnostics may further refine the evaluation of enteric neuropathies and transform the diagnostic landscape [12].

Radiological imaging remains central to the diagnosis and management of Hirschsprung's disease, its variants, and colorectal motility disorders [13]. Contrast enema continues to be the most informative modality, but multimodal imaging - including ultrasound, MRI, scintigraphy, and fluoroscopic motility studies - enhances diagnostic accuracy, especially in atypical presentations. Understanding the nuances of imaging findings, their limitations, and their integration with clinical and histopathological data is essential for accurate diagnosis and optimal surgical planning [14-15].

Clinical Implications

Radiological imaging plays a decisive role in the early recognition, accurate diagnosis, and effective management of Hirschsprung's Disease (HD), its variants, pseudo-Hirschsprung's conditions, and colorectal motility disorders. The findings of this review highlight several key implications for clinical practice.

First, contrast enema should remain the initial imaging modality in neonates and children with suspected HD, but clinicians must interpret its findings within the broader clinical context. Awareness of its limitations - particularly in ultrashort segment disease, long-segment involvement, and early neonatal imaging - can prevent misdiagnosis and reduce unnecessary delays in definitive testing.

Second, multimodal imaging improves diagnostic accuracy. Incorporating ultrasound, MRI, CT, nuclear scintigraphy, and fluoroscopic motility studies allows clinicians to distinguish true aganglionosis from functional obstruction, identify variant forms of HD, and evaluate postoperative symptoms. This integrated approach is especially important in atypical presentations and in older children where classical radiological signs may be absent.

Third, functional imaging and manometry are essential complements to structural imaging. Differentiating HD from pseudo-Hirschsprung's conditions such as internal anal sphincter achalasia or chronic intestinal pseudo-obstruction requires physiological assessment. Reliance on imaging alone risks both over- and under-diagnosis.

Fourth, interventional radiology expands therapeutic options. Image-guided rectal biopsy enhances diagnostic precision, while fluoroscopic decompression, contrast-guided irrigations, and targeted botulinum toxin injections provide minimally invasive management strategies for acute and chronic symptoms. These interventions reduce the need for more invasive procedures and support individualized care.

Finally, the review underscores the importance of multidisciplinary collaboration. Optimal outcomes depend on coordinated input from pediatric surgeons, radiologists, gastroenterologists, pathologists, and motility specialists. Standardized imaging protocols and shared diagnostic pathways can streamline evaluation, reduce variability, and improve patient outcomes.

Together, these implications support a shift toward structured, multimodal, and functionally informed diagnostic pathways that enhance accuracy, reduce diagnostic uncertainty, and guide tailored management across the spectrum of congenital and acquired colorectal dysmotility.

Future Research Directions

Despite significant advances in radiological and functional assessment of Hirschsprung's Disease (HD), its variants, and colorectal motility disorders, several important gaps remain. Future research should focus on developing standardized, reproducible imaging protocols that reduce variability across institutions and improve diagnostic consistency. Harmonization of contrast enema techniques, ultrasound criteria, and MRI sequences would allow more reliable comparison of findings and facilitate multicentre collaboration.

Emerging imaging biomarkers warrant further investigation. Ultrasound-based metrics, such as rectal wall thickness and muscularis propria hypertrophy, show promise but require validation in larger, age-stratified cohorts. Similarly, advanced MRI techniques, including diffusion-weighted imaging, cine MRI, and motility mapping, may offer non-invasive functional assessment of the enteric nervous system. Prospective studies are needed to determine their diagnostic accuracy, feasibility in infants, and utility in postoperative evaluation.

Functional imaging remains underutilized due to limited availability and expertise. Expanding access to colonic transit scintigraphy, fluoroscopic motility studies, and high-resolution colonic manometry could transform diagnostic pathways, particularly for differentiating HD from pseudo-HD and global motility disorders. Research should also explore novel nuclear tracers or molecular imaging techniques capable of visualizing enteric neuronal integrity or neurotransmitter activity.

The role of interventional radiology in diagnosis and therapy is another promising area. Image-guided rectal biopsy techniques could be refined to improve sampling accuracy, especially in atypical or reoperative cases. Further evaluation of botulinum toxin injection for internal anal sphincter achalasia and postoperative obstructive symptoms.

Conclusion

Radiological imaging remains indispensable in the evaluation of Hirschsprung's Disease (HD), its anatomical variants, pseudo-Hirschsprung's conditions, and colorectal motility disorders. While rectal biopsy provides the definitive diagnosis, imaging offers the essential structural and functional context that guides clinical suspicion, directs biopsy strategy, and informs operative planning. Contrast enema continues to serve as the cornerstone of initial assessment, yet its limitations - particularly in neonates, ultrashort segment disease, and long-segment or total colonic aganglionosis - underscore the need for complementary modalities.

Ultrasound, MRI, CT, nuclear scintigraphy, and fluoroscopic motility studies each contribute unique insights that enhance diagnostic accuracy, especially in atypical presentations or when distinguishing HD from pseudo-HD and other motility disorders. Functional imaging, in particular, plays a critical role in differentiating segmental aganglionosis from global dysmotility, preventing unnecessary surgery and enabling targeted management.

Interventional radiology has expanded the diagnostic and therapeutic landscape, offering image-guided biopsy, decompression techniques, and targeted interventions such as botulinum toxin injection. These minimally invasive approaches support both acute management and long-term symptom control.

Taken together, the evidence reinforces the importance of a multimodal, multidisciplinary diagnostic pathway that integrates radiological imaging with manometry, histopathology, and clinical evaluation. Such an approach reduces diagnostic uncertainty, improves patient selection for surgery, and enhances outcomes across the spectrum of congenital and acquired colorectal dysmotility. Continued research into advanced imaging biomarkers, standardized protocols, and functional assessment tools will further refine diagnostic precision and support earlier, more accurate identification of these complex conditions.

Hirschsprung's Disease (HD) and its variants represent a spectrum of congenital enteric neuropathies characterized by aganglionosis of the distal bowel. Radiological imaging plays a central

role in the diagnostic pathway, complementing clinical evaluation and histopathology. This review synthesizes the radiological features of classical and variant HD, pseudo-HD, and colorectal motility disorders, emphasizing contrast enema, plain radiography, CT, MRI, ultrasound, nuclear medicine studies, and fluoroscopic motility assessments. Interpretation pitfalls, transition zone variability, and the role of interventional radiology are discussed. Evidence from recent literature highlights the importance of multimodal imaging in differentiating true aganglionosis from functional obstruction and in guiding surgical planning.

References

1. Govani DJ, Trambadia, RA, Chhaniara RA, Mirani ZR, Chhaniara AP, Midha PK, et al. Pre-Operative Diagnosis of Infantile Triad of Waugh's Syndrome Associated with-Hypoganglionosis-Key Radiological Findings. Health Child Pediatr SMP. 2023: 1; 1-9.
2. Amaral FTS, Costa LLS. Updates on the diagnostic approach to Hirschsprung's disease: A literature review. Res Soc Dev. 2024; 13(12). doi:10.33448/rsd-v13i12.47684.
3. Georgy MNL, Guirguis MS, Niazi GEM, Zaky MM. Role of ultrasound in the diagnosis of Hirschsprung disease in infants. QJM. 2024. doi:10.1093/qjmed/hcae175.933.
4. Hirschsprung disease. Nat Rev Dis Primers. 2023.
5. Recent advances in paediatric motility disorders. Paediatr Child Health. 2024.
6. Rao SSC. Advances in anorectal manometry and motility testing. Neurogastroenterol Motil. 2023.
7. Modern histopathological approaches to diagnosing Hirschsprung disease. Pathology. 2020; 52(6).
8. Diagnostic accuracy of contrast enema in Hirschsprung disease. J Pediatr. 2021; 232: 123-130.
9. 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(1): 77-82. doi:10.1136/jcp.2004.016030.
10. Modern immunohistochemical markers for aganglionosis. Am J Clin Pathol. 2022.
11. Ambartsumyan L, Smith C, Kapur RP. Diagnosis of Hirschsprung Disease. Pediatric and Developmental Pathology. 2019; 23(1): 8-22. doi:10.1177/1093526619892351.
12. Trambadia RA, Anthony FM, Chhaniara RA, Govani DR, Correia RC, Patel RV. Hirschsprung- Associated recurrent enterocolitis in a case of trisomy 21 an atrioventricular septal defect. Front Infect Diseases Microbiol. 2022; 2: 110-13.
13. Patel RV, Elmalik K, Bouhadiba N, Shawis R. Hirschsprung's disease associated with Mowat-Wilson syndrome. BMJ Case Rep. 2014. doi:10.1136/bcr-2013-203262.
14. Patel RV, Lawther S, McCallion WA. Discordant monozygotic total colonic Hirschsprung's disease presenting with neonatal isolated ileal perforation. BMJ Case Rep. 2013. doi: 10.1136/bcr-2013-200743.
15. Zaparackaite I, Singh H, Singh SJ, Swamy KB, Midha PK, Patel RV. Neonatal Haddad Syndrome-Ondine curse with Hirschsprung's disease. Clinical Surgery Journal Submission.