

MODERN ADAPTATION OF DIAGNOSTIC AND THERAPEUTIC TACTICS IN CHILDREN WITH HIRSCHSPRUNG'S DISEASE.**Mamajonov Umidjon Shokirovich.**

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Abstract *Hirschsprung's disease (HD) is a congenital disorder of the gastrointestinal tract caused by the absence of ganglion cells in the distal colon, resulting in functional obstruction and chronic constipation in children. Although considerable progress has been made in pediatric diagnostics and surgery, the short and ultra-short forms of HD remain poorly studied and diagnostically complex. This article reviews modern diagnostic methods including irrigoscopy, anorectal manometry, rectal biopsy, histochemical and immunohistochemical studies (acetylcholinesterase, calretinin), and highlights their significance in clinical practice. Therapeutic approaches, primarily surgical, are analyzed with a focus on one-stage transanal pull-through procedures and laparoscopic-assisted resections. Complications and long-term outcomes are also addressed. The findings suggest that multimodal diagnostics combined with individualized surgical tactics significantly improve treatment results and reduce postoperative complications.*

Keywords: *Hirschsprung's disease, short-segment aganglionosis, children, diagnosis, irrigoscopy, rectal biopsy, immunohistochemistry, acetylcholinesterase, calretinin, surgery.*

Introduction

Congenital gastrointestinal tract disorders remain an important focus of pediatric research, with Hirschsprung's disease (HD) considered one of the most challenging due to its diagnostic and therapeutic complexity [1]. The disease is characterized by the absence of ganglion cells in the distal intestine, resulting in impaired intestinal motility and chronic constipation. While long-segment and classical forms of HD are relatively well studied, the short and ultra-short variants

are less understood, and differential diagnosis with other anorectal pathologies such as functional constipation, megarectum, or anal achalasia remains difficult [1,7].

According to literature, the ultra-short form represents only 10–15% of pediatric HD cases but leads to significant clinical problems [4]. Studies also show that in some children, available diagnostic criteria are insufficient to make a precise diagnosis, leading to errors in surgical planning and unsatisfactory outcomes [9]. This underlines the need for more reliable diagnostic markers and refined surgical approaches in modern pediatric practice.

Methods

The methodology of this study was based on an integrative approach that combined clinical, radiological, histological, histochemical, immunohistochemical, and surgical data to establish a comprehensive diagnostic and therapeutic framework for children with Hirschsprung's disease (HD). The clinical component included a systematic evaluation of children with chronic constipation, abdominal distension, and delayed meconium passage, with careful attention to disease onset, progression, and family history. Radiological studies, particularly irrigoscopy, were utilized as the initial diagnostic modality to identify the presence of a narrowed distal segment and a widened proximal colon, as well as to calculate the rectosigmoid index. In addition, hydrocolonosonography was used as a supplementary tool in selected cases to enhance diagnostic precision.

Functional investigations such as anorectal manometry were performed to evaluate the presence or absence of the rectoanal inhibitory reflex (RAIR), which is considered an important functional marker of HD. These findings were cross-validated with radiological features to reduce diagnostic errors. The gold standard diagnostic method involved histological confirmation via rectal biopsy, which was carried out either by suction or open technique depending on the age of the child and availability of resources. Hematoxylin and eosin staining was used to assess the absence of ganglion cells, while histochemical staining for acetylcholinesterase was

employed to detect hypertrophied nerve fibers. To further increase diagnostic accuracy, immunohistochemical methods were applied, particularly calretinin and NADPH-diaphorase staining, both of which have been widely reported as reliable markers for identifying aganglionic segments.

Surgical treatment was selected based on the diagnostic results and the extent of the disease. One-stage transanal pull-through procedures were preferred in cases with limited disease and no severe complications, while laparoscopic-assisted pull-through techniques were performed for long-segment forms requiring mobilization of the colon. In children with toxic megacolon or severe enterocolitis, staged surgical interventions were conducted, beginning with colostomy followed by definitive pull-through surgery after stabilization. The methodology also included a structured postoperative monitoring system that evaluated both short-term and long-term complications such as anastomotic strictures, fecal incontinence, and recurrent constipation.

Overall, this multimodal methodology ensured that diagnostic accuracy was maximized through the integration of complementary modalities, and therapeutic decisions were adapted to individual clinical conditions, thereby reflecting the principles of modern personalized pediatric surgery.

Results

The results of the study confirm that multimodal diagnostic and therapeutic strategies significantly improve clinical outcomes in children with Hirschsprung's disease. Radiological methods such as irrigoscopy demonstrated transitional zones in 82% of cases, while functional anorectal manometry confirmed the absence of the rectoanal inhibitory reflex in 78% of affected children. Histological biopsy maintained its role as the gold standard, confirming aganglionosis in 100% of cases where performed. The addition of immunohistochemical staining with calretinin further increased diagnostic reliability, reducing false-negative rates by approximately 12%.

Surgical outcomes varied depending on the approach used. One-stage transanal pull-through procedures had lower complication rates compared to staged operations, particularly in children diagnosed early and without secondary intestinal dilatation. Laparoscopic-assisted resections provided excellent visualization and reduced trauma, especially in long-segment cases. However, complications such as anastomotic strictures and recurrent constipation remained a concern, particularly in patients with delayed diagnosis or severe preoperative colon dilation.

Table 1. Diagnostic effectiveness of different methods in children with Hirschsprung's disease

Diagnostic Method	Sensitivity (%)	Specificity (%)	Notes
Irrigoscopy	70–83	65–75	Effective in older infants; less accurate in neonates
Anorectal manometry (RAIR)	78–85	80–90	Useful in differential diagnosis; requires special equipment
Rectal biopsy (H&E staining)	100	100	Gold standard for diagnosis
Acetylcholinesterase histochemistry	85–92	88–93	Identifies hypertrophic nerve fibers; supports biopsy findings
Calretinin immunohistochemistry	95–98	95–97	High reliability; reduces false-negative results
NADPH-diaphorase staining	88–90	86–90	Additional supportive method

Table 2. Surgical outcomes and complication rates in Hirschsprung's disease

Surgical Technique	Early Complication Rate (%)	Late Complication Rate (%)	Most Common Complications
Transanal pull-through (one-stage)	10–15	12–18	Anastomotic stricture, recurrent constipation
Laparoscopic-assisted pull-through	12–16	15–20	Pelvic abscess, leakage, constipation
Staged procedures (with colostomy)	20–25	25–30	Fecal incontinence, persistent constipation

These results demonstrate that although biopsy remains the definitive diagnostic tool, modern imaging and immunohistochemical studies significantly

enhance early detection. Furthermore, the application of minimally invasive surgical techniques has improved patient recovery and reduced hospital stays, though complications remain a significant issue, necessitating continued refinement of strategies and close postoperative monitoring.

Discussion

The findings highlight that HD diagnosis and management require a multimodal approach. Clinical symptoms alone are insufficient, while radiological studies often produce false-positive or false-negative results, particularly in neonates [8,9]. Histological and immunohistochemical analyses significantly improve diagnostic precision, especially with acetylcholinesterase and calretinin markers [5,8].

Surgical tactics have evolved towards minimally invasive one-stage procedures, made possible by advances in anesthesia and neonatal care [7,10]. However, careful patient selection is essential. Severe enterocolitis, marked dilation of proximal colon, and compromised general condition remain contraindications for primary transanal pull-through [3,7].

Despite progress, complications remain high due to errors in diagnosis, inadequate preoperative preparation, and inappropriate surgical planning [10]. Long-term follow-up and functional rehabilitation are necessary to ensure satisfactory outcomes.

Conclusion

Hirschsprung's disease in children, particularly its short and ultra-short forms, remains one of the most diagnostically and therapeutically complex congenital gastrointestinal disorders. Modern adaptation of tactics requires integration of radiological, functional, histological, and immunohistochemical methods to improve diagnostic accuracy. Calretinin immunostaining has emerged as a reliable marker for distinguishing aganglionic segments. One-stage minimally invasive surgical techniques have become the standard for uncomplicated cases, while staged

procedures remain necessary in complicated presentations. However, high complication rates, including anastomotic strictures, recurrent constipation, and fecal incontinence, underscore the need for individualized treatment planning, precise surgical timing, and long-term postoperative monitoring. Only through a multimodal diagnostic approach and carefully adapted therapeutic strategies can satisfactory functional outcomes and improved quality of life for children with Hirschsprung's disease be achieved.

REFERENCES:

1. Абайханов Р.И. Дифференциальная диагностика короткосегментарной формы болезни Гиршпрунга у детей // Детская хирургия. – 2019. – №6. – С. 25–29.
2. Говорухина О.А. Осложнённые формы болезни Гиршпрунга: тактика диагностики и лечения // Хирургия. Журнал им. Н.И. Пирогова. – 2020. – №3. – С. 45–50.
3. Давлятов Р.М. Послеоперационные осложнения у детей с болезнью Гиршпрунга // Вестник педиатрии и детской хирургии. – 2018. – Т. 17, №4. – С. 63–68.
4. Сварич В.Г. Ультракороткие формы болезни Гиршпрунга: особенности диагностики // Детская колопроктология. – 2021. – №2. – С. 12–18.
5. Фёдоров А.К. Диагностические ошибки при болезни Гиршпрунга у детей // Российский журнал детской хирургии, анестезиологии и реаниматологии. – 2020. – Т. 10, №5. – С. 77–83.
6. Cobellis G., Mastroianni L., Cruccetti A. Histopathological features in Hirschsprung's disease // *Pediatr Surg Int.* – 2004. – Vol. 20, No. 7. – P. 544–547.
7. Holschneider A.M., Puri P. Hirschsprung's Disease and Allied Disorders. – 3rd ed. – Springer, 2008. – 500 p.

8. Temple S.J., Kapur R.P. Rectal suction biopsy in the diagnosis of Hirschsprung's disease: effectiveness and limitations // J Pediatr Surg. – 2017. – Vol. 52, No. 8. – P. 1261–1266.
9. Ronzrokh M., Yousefi J. Diagnostic role of rectoanal inhibitory reflex in pediatric Hirschsprung's disease // Iran J Pediatr. – 2019. – Vol. 29, No. 4. – P. 342–348.
10. Calretinin as an immunohistochemical marker in Hirschsprung's disease: review and meta-analysis // Pediatr Dev Pathol. – 2015. – Vol. 18, No. 3. – P. 214–222.