Identifying Hirschsprung Disease in the First Month of Life

Nationwide Children's®
When your child needs a hospital, everything matters.

Why is Constipation Critical in the First Month of Life?

Newborns typically pass meconium (thick, dark stool) within the first 24 hours of life. Failure to do so may suggest an obstruction, either functional or anatomical.

In the first month, newborn stooling patterns can vary based on feeding type. Breastfed infants are more efficient at digesting breast milk, going 7 to 10 days between bowel movements. In contrast, formula-fed infants usually pass stool every 1 to 2 days due to formula composition.

Newborns in their first month experiencing difficulty passing stool or gas, particularly when accompanied by fever and abdominal distention, should be evaluated for Hirschsprung disease.

Overview of Hirschsprung Disease

In approximately 1 in 5,000 live births, this congenital condition occurs when ganglion nerve cells are absent in the large intestine. Hirschsprung disease can affect a short segment of the colon, a longer segment, or, in rare cases, extend into parts of the small intestine. The lack of ganglion cells, combined with hypertrophic (enlarged) nerves, impair the normal movement of stool and leads to obstructions in the bowel. Hirschsprung disease accounts for about 10% of all neonatal bowel obstructions. The condition puts children at significant risk for severe enteric infections and abdominal distension, and if left untreated, can be life-threatening.

Although Hirschsprung disease develops during fetal development, it is not typically diagnosed until after birth. Around 80% of cases are identified during the first year of life. Hirschsprung disease is more common in males than females and is frequently associated with other genetic and inherited conditions, most notably Trisomy 21 (Down syndrome).

Genetic Causes of Hirschsprung Disease

Genetic factors play a significant role in the abnormal development of ganglion cells in the intestines. These factors affect the way ganglion cells migrate down the small intestine and colon and also how these cells mature. Both migration and maturation are important for normal function. Genetic abnormalities may be inherited or develop spontaneously in patients with Hirschsprung disease.

Symptoms of Hirschsprung Disease

Hirschsprung disease presents with a range of symptoms related to impaired bowel motility:

- Failure to pass the first bowel movement within 48 hours of birth
- Difficulty passing stool and gas, requiring rectal stimulation
- Explosive, foul-smelling stools
- Abdominal distention, with a rounded, firm abdomen on palpation
- Fever due to infection
- Green or brown vomit
- Feeding problems due to bowel discomfort
- Abnormal growth and weight gain

Common Complications

Enterocolitis

Children with Hirschsprung disease have a very sensitive colonic mucosal lining, making them vulnerable to enterocolitis. When stool remains static, bacterial overgrowth can occur. Rectal irrigations are required to remove stool and reduce the level of infection. If untreated, enterocolitis can lead to sepsis and rarely death.

Slowed Growth

Failure to thrive is common, with slowed growth and difficulty gaining weight due to chronic constipation, bacterial overgrowth in the bowel and impaired nutrient absorption. Careful monitoring and intervention are needed to promote proper growth and development.

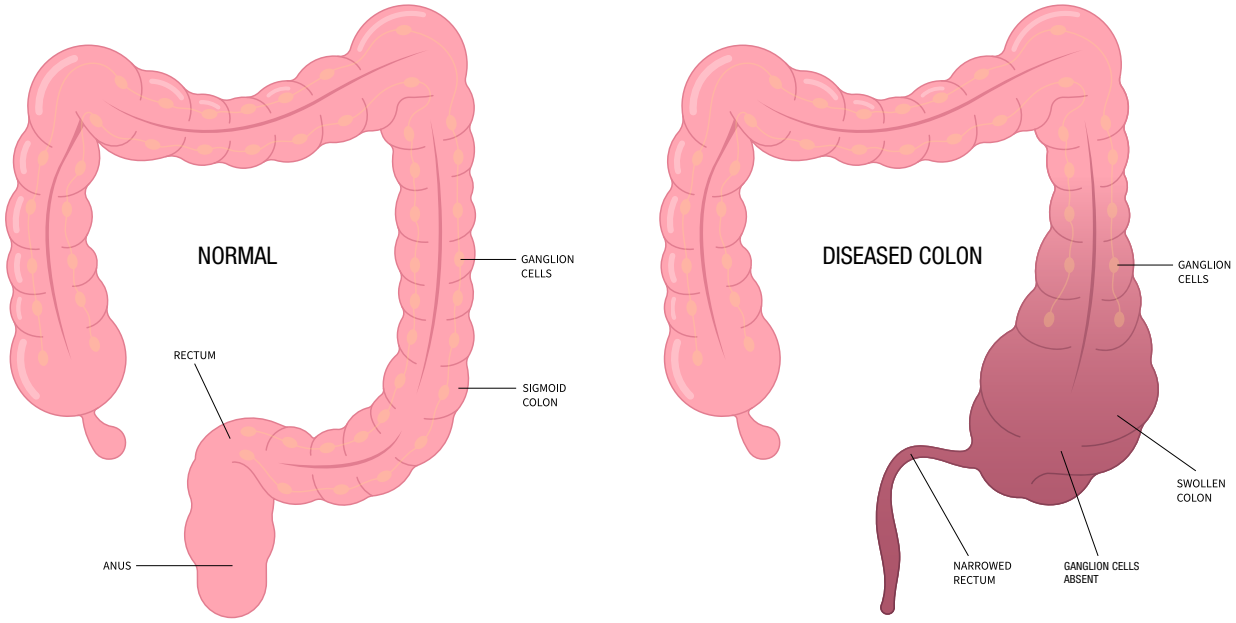
Diagnostic Testing

While contrast studies of the colon can help assess anatomical changes in the colon and raise suspicion for Hirschsprung disease, they are not sufficient to confirm the diagnosis. The definitive standard for diagnosis is either a suction rectal biopsy or a full-thickness rectal biopsy. A biopsy confirms the absence of ganglion cells and assesses the size of nerves in the affected bowel segment.

Treatment Options

Surgical removal of the affected portion of the bowel is the primary treatment. This surgery is typically performed in one procedure unless the affected segment of the bowel is unusually long. In that case, a staged approach may be necessary.

Post-surgery, patients remain at risk for enterocolitis. Ongoing medical management is essential to ensure the child regularly passes stool and gas. In the early stages, the focus is on ensuring the child is stooling regularly in their diapers. As the child reaches the potty training stage, specialists will introduce a customized medication regimen to ensure daily bowel function.



When to Refer to a Specialist

Referral to the Center for Colorectal and Pelvic Reconstruction at Nationwide Children's Hospital is recommended if Hirschsprung disease is suspected based on the clinical presentation of one or more symptoms described above.

About Our Center

The Center for Colorectal and Pelvic Reconstruction at Nationwide Children's is one of the first centers in the world to formally integrate all specialties involved in providing complete care for the colon and rectum in children. Our team includes experts in Colorectal Surgery, Gastroenterology, Gynecology, and Urology.

Each year, we care for more children with complex colorectal conditions and perform more colorectal surgeries than any other pediatric health care system in the country. This extensive experience leads to better long-term surgical and clinical outcomes.

We are also committed to advancing research, sharing best practices, and collaborating with primary care pediatricians and other providers who share our mission to deliver the best possible care for pediatric patients.

Referrals and Consultations

Online: **NationwideChildrens.org/CCPR**

Phone: **(614) 722-6200** or **(877) 722-6220** | Fax: **(614) 722-4000**

Physician Direct Connect Line for 24-hour urgent physician consultations: **(614) 355-0221** or **(877) 355-0221**.

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