CONDITION

Hirschsprung disease

Learn how aganglionic megacolon, also called Hirschsprung disease, affects bowel function, including symptoms, diagnosis, surgery, complications, and follow-up care.

WHAT IT IS
Congenital absence of bowel nerve cells

BODY SYSTEM
Digestive

MAIN SYMPTOMS
Delayed meconium, severe constipation, abdominal swelling, vomiting

WHAT CAUSES IT
Sometimes genetic, often unknown

HOW IT IS DIAGNOSED
Rectal biopsy

MAIN TREATMENT
Surgery

CAN IT BE CURED?
Sometimes - surgery works, symptoms may persist

IS IT AN EMERGENCY?
Sometimes - fever with a swollen abdomen

LONG-TERM OUTLOOK
Many symptoms improve, but ongoing follow-up is often needed

Overview

Aganglionic megacolon is more commonly called Hirschsprung disease . It is a condition present from birth in which nerve cells called ganglion cells are missing from part of the large intestine. These cells normally coordinate muscle movements that push stool through the bowel.

Without ganglion cells, the affected bowel remains tightly contracted. Stool and gas cannot pass through it normally. The bowel above the narrowed area may become stretched and enlarged, which is why the condition is sometimes called megacolon.

The affected segment usually begins at the rectum, the final part of the large intestine. In many children, only the rectum and nearby sigmoid colon are involved. Some have a longer affected segment, and a small number have disease involving the entire colon or part of the small

intestine.

Most cases are identified in newborns or young children. However, milder or short-segment disease may not be recognized until later childhood or, rarely, adulthood.

Hirschsprung disease matters because it can cause severe constipation, intestinal blockage, and a serious bowel infection called Hirschsprung-associated enterocolitis . Surgery can restore a pathway for stool to leave the body, but ongoing follow-up is often needed for bowel symptoms, nutrition, and quality of life.

Symptoms

Symptoms depend on the child's age, the length of affected bowel, and whether a complication has developed.

A common early sign is failure to pass meconium , the thick first stool, within the first 48 hours after birth. Not every baby who passes meconium late has Hirschsprung disease, and some babies with the condition pass stool during this period.

Other symptoms in newborns may include:

Older babies and children may have long-term constipation, abdominal swelling, poor appetite, slow growth, or difficulty gaining weight. Stools may be small, hard, or infrequent. Some children have episodes of stool leakage because liquid stool passes around retained stool. Unlike common childhood constipation, symptoms may have been present since infancy and may respond poorly to usual treatments.

Some people with a short affected segment are diagnosed later. They may report severe constipation throughout life, frequent abdominal swelling, or dependence on enemas or other bowel treatments. These symptoms have many possible causes, so testing is needed before Hirschsprung disease can be confirmed.

Hirschsprung-associated enterocolitis can occur before or after surgery. Symptoms may include fever, increasing abdominal swelling, vomiting, lethargy, poor feeding, and explosive, foul-smelling, or watery diarrhea.

Enterocolitis can worsen quickly. Seek urgent medical assessment if these symptoms develop. Green vomit, marked sleepiness, breathing difficulty, signs of dehydration, a rapidly enlarging abdomen, or a child who appears seriously unwell should be treated as an emergency. Families whose child has Hirschsprung disease should follow the emergency plan provided by their surgical team.

- A swollen or firm abdomen

- Trouble feeding
- Vomiting, which may be green when it contains bile
- Few or no bowel movements
- Difficulty gaining weight
- Sudden passage of gas and stool after a rectal examination

Causes and risk factors

Hirschsprung disease develops before birth. During early fetal development, nerve-forming cells normally move through the digestive tract. In this condition, they do not reach the end of the bowel. The reason this process stops is not always known.

Genetic changes contribute to some cases. Several genes involved in nerve development have been associated with the condition, including the RET gene. The genetics are complex: having a related genetic change does not always mean that a person will develop Hirschsprung disease.

The condition may occur by itself or as part of a genetic syndrome. It is associated with Down syndrome and several less common conditions affecting nerve development. Some children also have congenital differences involving the heart, urinary system, or other parts of the digestive tract.

Factors linked with a higher likelihood include:

Parents do not cause Hirschsprung disease through diet, medicines, or actions during pregnancy. When there is a family history, extensive bowel involvement, or a suspected syndrome, genetic counseling may help families understand testing and the chance of the condition occurring in another relative.

- A close relative with Hirschsprung disease
- Being male, especially in the more common short-segment form
- Down syndrome or certain other genetic conditions
- Having other congenital differences

Diagnosis

Diagnosis begins with a medical history and physical examination. The clinician will ask when symptoms started, when the baby first passed meconium, how often stool passes, and

whether there has been vomiting, abdominal swelling, poor growth, or enterocolitis.

An abdominal examination can identify swelling or tenderness. A careful rectal examination may find a tight rectum or trigger the release of stool and gas. This finding can support suspicion of Hirschsprung disease, but it does not confirm the diagnosis.

Several tests may be used:

The biopsy should be reviewed by a pathologist experienced in pediatric bowel disease. If results are uncertain, another sample or additional staining may be necessary.

During suspected enterocolitis or severe obstruction, stabilizing the patient takes priority. Some diagnostic procedures may be delayed until the acute illness is controlled.

- Abdominal X-ray: This may show widened bowel loops, retained stool, or signs of obstruction. The findings are not specific to Hirschsprung disease.
- Contrast enema: Contrast material is placed into the rectum while X-ray images are taken. The study may show a narrow affected segment and a wider bowel above it. In young babies, this transition may not yet be obvious.
- Anorectal manometry: A small balloon and pressure sensor test how the rectum and anal muscles respond to stretching. In Hirschsprung disease, a normal relaxation response called the rectoanal inhibitory reflex is usually absent.
- Rectal biopsy: This is the test used to confirm the diagnosis. A small tissue sample is examined for ganglion cells. A suction biopsy can often be performed in infants. An older child or a person with an unclear result may need a larger, full-thickness biopsy.

Treatment and management

Surgery is the main treatment. The goal is to remove or bypass bowel without ganglion cells and connect healthy, functioning bowel to the anus. This is called a pull-through procedure .

Several pull-through techniques are available. The choice depends on the length of affected bowel, the surgeon's experience, the child's health, and whether complications are present. Many children can have a one-stage operation. The procedure may be performed through the anus, through the abdomen using small incisions, or with a combination of approaches.

Some patients need treatment in stages. A temporary ostomy may be created, bringing healthy bowel to an opening in the abdominal wall so stool can empty into a bag. An ostomy may be used when a baby is very unwell, the bowel is severely enlarged, enterocolitis or perforation is present, or the affected segment is extensive. It may later be closed during or after pull-through surgery.

Before surgery, rectal irrigations may be used to remove trapped stool and gas. Caregivers should receive hands-on instruction because the correct tube, fluid, volume, and technique depend on the patient and clinical situation.

Enterocolitis generally requires prompt medical treatment. Care may include rectal irrigations, fluids, bowel rest, antibiotics like amoxicillin or doxycycline , and hospital monitoring. Severe illness may require intensive support or emergency surgery. Families should not try to manage suspected enterocolitis only with ordinary constipation treatments.

After surgery, pain control, hydration, wound care, and gradual return to feeding are important. The surgical team may recommend temporary anal dilations if narrowing develops or if the specific operation requires them. These should be performed only as instructed.

Some children continue to have constipation or trouble emptying after technically successful surgery. Management may include dietary changes, laxatives, enemas, structured bowel-management programs, pelvic-floor therapy, or selected procedures. Botulinum toxin injection may be considered when the internal anal sphincter does not relax appropriately. Treatment should be guided by specialists because similar symptoms can result from different surgical, muscular, or motility problems.

Complications

Before treatment, Hirschsprung disease can cause complete bowel obstruction, severe bowel enlargement, poor nutrition, dehydration, or intestinal perforation. Enterocolitis is one of the most important complications and may occur even after the affected bowel has been removed.

Possible problems after surgery include:

Many bowel symptoms can be improved once their cause is identified. Evaluation may include an examination under anesthesia, contrast imaging, repeat biopsy, or tests of bowel and anal muscle function.

Bowel-control difficulties can affect school, travel, sports, sleep, and self-esteem. Age-appropriate education, access to toilets, a written school plan, and psychological support may be useful parts of care.

- Recurrent enterocolitis
- Constipation or difficulty emptying the bowel
- Frequent stools, urgency, or stool leakage

- Irritated skin around the anus
- Narrowing at the surgical connection
- An abnormal twist, pouch, or blockage in the reconstructed bowel
- Remaining bowel without ganglion cells
- Difficulty relaxing the anal sphincter
- Growth or nutrition concerns

Follow up

Regular follow-up with a pediatric surgeon is important after diagnosis and surgery. A pediatric gastroenterologist, dietitian, continence nurse, genetic counselor, or bowel-management team may also be involved.

Routine visits may review stool frequency and consistency, abdominal swelling, continence, skin care, diet, growth, medicines, and any episodes of enterocolitis. Persistent symptoms should not automatically be assumed to be an expected result of surgery. They may need structured investigation.

Contact the care team promptly for worsening constipation, repeated abdominal swelling, poor intake, vomiting, new stool leakage, painful bowel movements, or slowed growth. These concerns usually require timely clinical review but are not always emergencies.

Fever with a swollen abdomen, explosive diarrhea, green vomiting, unusual sleepiness, or signs of serious illness needs urgent or emergency assessment because of the possibility of enterocolitis or obstruction.

Follow-up should continue through adolescence. Young people benefit from learning the name of their operation, warning symptoms, current bowel plan, and how to obtain adult care. Some continue to need support for constipation, continence, sexual health, or emotional well-being after leaving pediatric services.

Sources

- National Institute of Diabetes and Digestive and Kidney Diseases. Hirschsprung Disease: Definition and Facts; Symptoms and Causes; Diagnosis; Treatment . National Institutes of Health.

- MedlinePlus Genetics. Hirschsprung Disease . National Library of Medicine, National Institutes of Health.
- Kyrklund K, Sloots CEJ, de Blaauw I, et al. ERNICA guidelines for the management of rectosigmoid Hirschsprung's disease. Orphanet Journal of Rare Diseases . 2020;15:164.
- Gosain A, Frykman PK, Cowles RA, et al. Guidelines for the diagnosis and management of Hirschsprung-associated enterocolitis. Pediatric Surgery International . 2017;33(5):517-521.
- North American Society for Pediatric Gastroenterology, Hepatology and Nutrition. Evaluation and management of the postsurgical patient with Hirschsprung disease: a position paper. Journal of Pediatric Gastroenterology and Nutrition . 2023.