GI Kids
Help & Hope for Children with Digestive Disorders
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Hirschsprung's Disease

What is Hirschsprung's disease?

Hirschsprung's disease (HD) is a rare but serious cause of severe constipation and/or intestinal blockage in infants. Rarely, older children and adolescents who have severe constipation are diagnosed with HD.

Infants with this condition are missing a type of nerve cell called ganglion cells in part of their intestine (usually the colon, or large intestine). When these cells are absent, the intestine cannot move its contents through. As a result, the infant can have severe constipation or symptoms of lower intestinal blockage (vomiting, swollen belly).

How common is HD? Who is affected?

Overall, HD is rare, occurring in 1:5000 infants. It usually presents in newborns or early infancy, but rarely it may present in late childhood and adolescence.

Boys are more commonly affected than girls. HD may affect multiple members in a family. In fact, 10%–33% of individuals with HD also have other family members with HD.

The exact causes of HD are unknown, but genetic mutations likely contribute. HD is common in individuals with genetic conditions like Down syndrome, Waardenburg-Shah syndrome, Smith-Lemli-Opitz syndrome, neurofibromatosis, and multiple endocrine neoplasia type 2.

What are the symptoms of HD?

HD may present in any of the following ways:

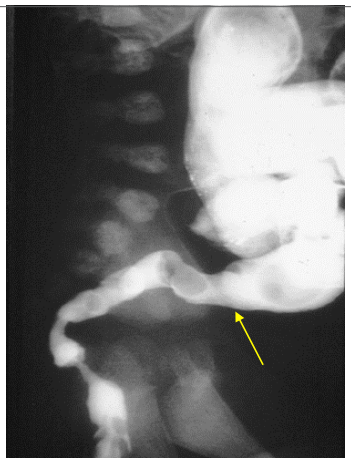
- A newborn who does not pass meconium (a black, sticky stool) in the first 48 hours of life may be showing the first sign of possible HD.
- Severe constipation in newborns or infants. Infants who have difficulty having spontaneous bowel movements and frequently require rectal stimulation to pass stool, for example, using suppositories and enema, may be suspected of having HD.
- Lower intestinal obstruction symptoms. A newborn or infant may show symptoms of lower intestinal obstruction such as severe constipation, inability to pass gas (obstipation), abdominal swelling (distention), bile in the vomit, abdominal pain, feeding difficulties, and weight loss. This is a surgical emergency.
- Older children and teenagers may experience "difficult-to-treat", or refractory, chronic constipation. They frequently require significant amounts of oral and rectal laxative treatment to pass stool. They may have chronic vomiting, chronic abdominal swelling, poor appetite, and malnutrition.
- Rarely, HD can present acutely with severe symptoms of vomiting, diarrhea, abdominal swelling and pain, fevers, and an ill-appearing child. This is called Hirschsprung's

associated enterocolitis (HAEC). This is a medical emergency and requires prompt and intensive care by several specialists.

How is HD diagnosed?

Your child's doctor may suspect HD based on the child's medical history and physical examination; further testing by a specialist is required to confirm the diagnosis. A correct diagnosis is important to guide proper treatment and prevent complications from HD. Diagnosis may involve a series of tests, including the following:

- A contrast enema study - This is a contrast-based radiographic (X-ray) test to outline the shape of the intestines and can help rule out other causes of constipation. HD usually, but not always, shows a specific pattern on X-ray, where the lowest part of the large intestine is narrow, and above the narrow area is a wider area. This appearance is sometimes referred to as a "funnel shape."



BARIUM ENEMA (X-RAY) IN A HIRSCHSPRUNG'S DISEASE PATIENT

The arrow highlights the transition from dilated (widened) intestine to the narrowed intestine which is missing nerve cells.

- Anorectal manometry - Some centers offer studies, called manometry, to observe the intestine's ability to move contents through. In an anorectal manometry test, a balloon is inserted into the rectum. The balloon is then inflated. In people without HD, the body believes the balloon is a bowel movement, and so the anus relaxes to let the balloon out. In a patient with HD, the anus is unable to relax because of the missing intestinal nerve cells.
- Rectal biopsy to look for ganglion cells - Sometimes a biopsy can help to identify whether the nerve cells are present or not. A physician will obtain a small amount of rectal tissue by either inserting a small tube (called suction biopsy) or by making a small incision (open surgical full-thickness biopsy) to look for ganglion cells. Both techniques are safe and reliable when performed by experts. The biopsied tissue is then analyzed under a microscope to look for the features of HD. In HD, ganglion cells are absent, and there is more of a protein called acetylcholinesterase.

How is HD treated?

HD is treated through surgery. There are several different surgical techniques, for example Swenson, Duhamel-Martin, and Soave endorectal pull-through operation. For all of these techniques, the goal is to remove the part of the colon that is missing the ganglion cells (aganglionic colon) and use the neighboring normal (ganglionic) colon to create a new rectum.

In the past, a temporary colostomy was performed to allow time for the infant to grow and for the wider region of colon to reduce in size, making the operation easier. Now, however, because of earlier diagnosis of HD and improved surgical techniques, there is rarely a need for a colostomy. Surgeries to treat HD are performed fairly easily even in young infants.

What can I expect after surgery for HD?

Overall, children have favorable outcomes after surgery for HD. Most children recover well, pass 1–3 bowel movements per day, and grow normally.

However, about 15% of children may continue to experience issues like persistent constipation, difficulties in toilet training, fecal incontinence, narrowing of the connecting piece of colon, and repeated episodes of enterocolitis. These children need ongoing follow-up and an individualized treatment plan directed by an interdisciplinary bowel management program in a specialized center.

Occasionally, a repeat operation may be required for persistent constipation symptoms not responding to medical treatment.

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Locate a Pediatric Gastroenterologist

IMPORTANT REMINDER: *This information from the North American Society for Pediatric Gastroenterology, Hepatology and Nutrition (NASPGHAN) is intended only to provide general educational information as a definitive basis for diagnosis or treatment in any particular case. It is very important that you consult your doctor about your specific condition.*

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