

# What I need to know about Hirschsprung Disease



U.S. Department  
of Health and  
Human Services

NATIONAL INSTITUTES OF HEALTH

**NIDDK** NATIONAL INSTITUTE OF  
DIABETES AND DIGESTIVE  
AND KIDNEY DISEASES

National Digestive Diseases  
Information Clearinghouse



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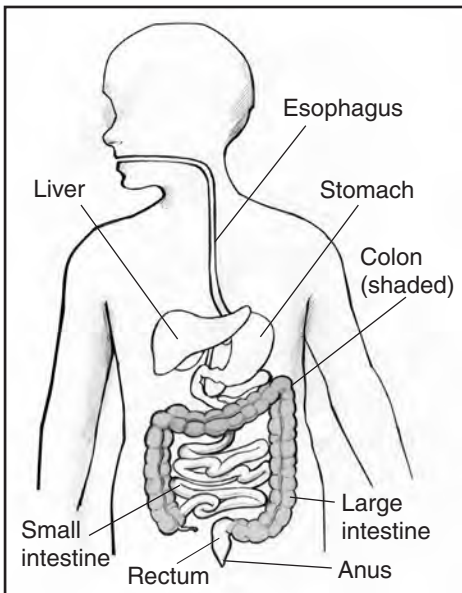
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## Contents

What is Hirschsprung disease (HD)?.....	1
What are the large intestine, colon, rectum, and anus? .....	2
Why does HD cause constipation?.....	2
What causes HD?.....	3
What are the symptoms of HD? .....	4
How is HD diagnosed? .....	6
How is HD treated? .....	7
What will my child's life be like after surgery? .....	10
If I have more children, will they also have HD? .....	13
Points to Remember .....	14
Hope through Research .....	15
Pronunciation Guide.....	16
For More Information .....	17
Acknowledgments .....	18

## What is Hirschsprung disease (HD)?

**Hirschsprung\*** disease (HD) is a disease of the large **intestine** that causes severe **constipation** or intestinal **obstruction**. Constipation means stool moves through the intestines slower than usual. Bowel movements occur less often than normal and stools are difficult to pass. Some children with HD can't pass stool at all, which can result in the complete blockage of the intestines, a condition called intestinal obstruction. People with HD are born with it and are usually diagnosed when they are infants. Less severe cases are sometimes diagnosed when a child is older. An HD diagnosis in an adult is rare.



HD is a disease of the large intestine that causes severe constipation or intestinal obstruction.

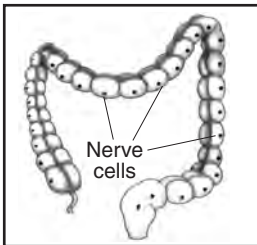
\*See page 16 for tips on how to say the words in **bold** type.

## What are the large intestine, colon, rectum, and anus?

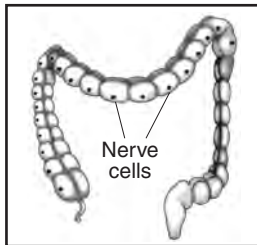
The large intestine, which includes the colon and **rectum**, is the last part of the digestive tract. The large intestine's main job is to absorb water and hold stool. The rectum connects the colon to the anus. Stool passes out of the body through the **anus**. At birth, the large intestine is about 2 feet long. An adult's large intestine is about 5 feet long.

## Why does HD cause constipation?

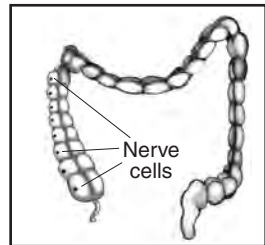
People with HD have constipation because they lack nerve cells in a part or all of the large intestine. The nerve cells signal muscles in the large intestine to push stool toward the anus. Without a signal to push stool along, stool will remain in the large intestine.



Healthy large intestine. Nerve cells are found throughout the large intestine.



Short-segment HD. Nerve cells are missing from the last segment of the large intestine.



Long-segment HD. Nerve cells are missing from most or all of the large intestine and sometimes the last part of the small intestine.

How severe HD is depends on how much of the large intestine is affected. Short-segment HD means only the last part of the large intestine lacks nerve cells. Long-segment HD means most or all of the large intestine, and sometimes the last part of the small intestine, lacks nerve cells.

In a person with HD, stool moves through the large intestine until it reaches the part lacking nerve cells. At that point, the stool moves slowly or stops, causing an intestinal obstruction.

## **What causes HD?**

Before birth, a child's nerve cells normally grow along the intestines in the direction of the anus. With HD, the nerve cells stop growing too soon. Why the nerve cells stop growing is unclear. Some HD is inherited, meaning it is passed from parent to child through genes. HD is not caused by anything a mother did while pregnant.

## What are the symptoms of HD?

The main symptoms of HD are constipation or intestinal obstruction, usually appearing shortly after birth. Constipation in infants and children is common and usually comes and goes, but if your child has had ongoing constipation since birth, HD may be the problem.

### Symptoms in Newborns

Newborns with HD almost always fail to have their first bowel movement within 48 hours after birth. Other symptoms include

- green or brown vomit
- explosive stools after a doctor inserts a finger into the rectum
- swelling of the belly, also known as the **abdomen**
- lots of gas
- bloody **diarrhea**





## Symptoms in Toddlers and Older Children

Symptoms of HD in toddlers and older children include

- not being able to pass stools without **laxatives** or **enemas**. A laxative is medicine that loosens stool and increases bowel movements. An enema is performed by flushing water, or sometimes a mild soap solution, into the anus using a special wash bottle.
- swelling of the abdomen.
- lots of gas.
- bloody diarrhea.
- slow growth or development.
- lack of energy because of a shortage of red blood cells, called **anemia**.



People with anemia tire easily because of a shortage of red blood cells.

## How is HD diagnosed?

HD is diagnosed based on symptoms and test results.

A doctor will perform a physical exam and ask questions about your child's bowel movements. HD is much less likely if parents can identify a time when their child's bowel habits were normal.

If HD is suspected, the doctor will do one or more tests.

### **X rays**

An x ray is a black-and-white picture of the inside of the body. To make the large intestine show up better, the doctor may fill it with **barium** liquid. Barium liquid is inserted into the large intestine through the anus.

If HD is the problem, the last segment of the large intestine will look narrower than normal. Just before this narrow segment, the intestine will look bulged. The bulging is caused by blocked stool stretching the intestine.

### **Manometry**

During **manometry**, the doctor inflates a small balloon inside the rectum. Normally, the rectal muscles will relax. If the muscles don't relax, HD may be the problem. This test is most often done in older children and adults.

# Biopsy

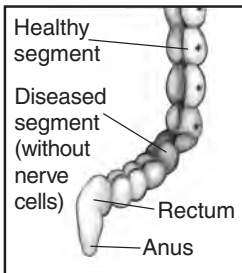
**Biopsy** is the most accurate test for HD. The doctor removes a tiny piece of the large intestine and looks at it with a microscope. If nerve cells are missing, HD is the problem.

## How is HD treated?

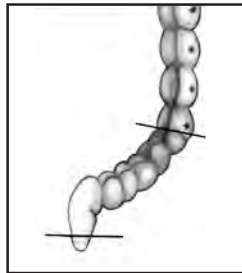
### Pull-through Procedure

HD is treated with surgery called a pull-through procedure. A surgeon removes the segment of the large intestine lacking nerve cells and connects the healthy segment to the anus. The pull-through procedure is usually done soon after diagnosis.

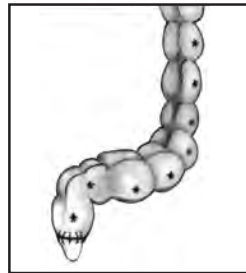
#### Pull-through Procedure



Before pull-through surgery: The diseased segment doesn't push stool.



Step 1: The diseased segment is removed.

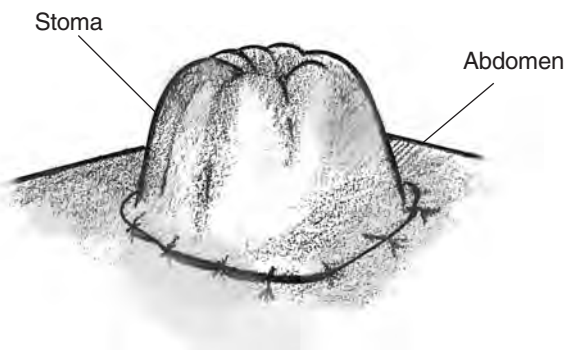


Step 2: The healthy segment is attached to the remaining rectum.

## Ostomy surgery

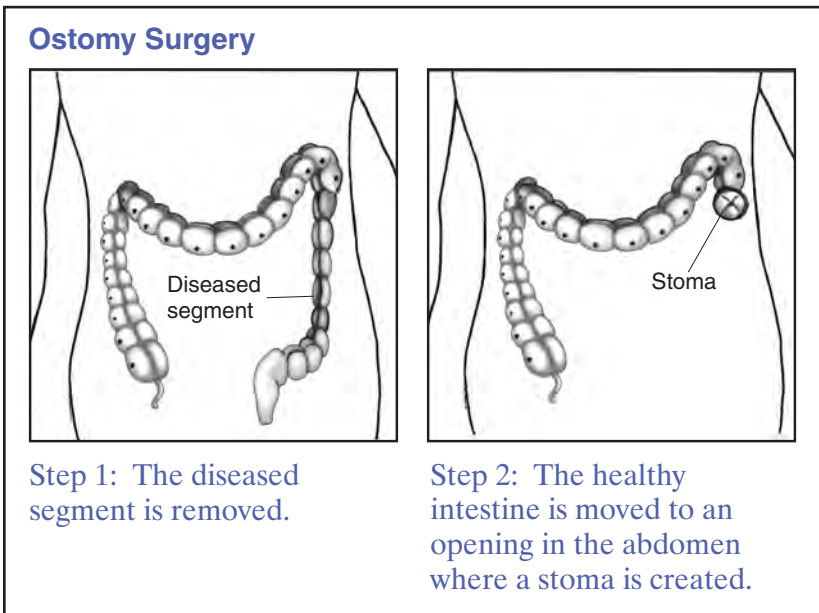
An **ostomy** allows stool to leave the body through an opening in the abdomen. Although most children with HD do not need an ostomy, a child who has been very sick from HD may need an ostomy to get better before the pull-through procedure.

For ostomy surgery, the surgeon first takes out the diseased segment of the large intestine. The end of the healthy intestine is moved to an opening in the abdomen where a **stoma** is created. A stoma is created by rolling the intestine's end back on itself, like a shirt cuff, and stitching it to the abdominal wall. An ostomy pouch is attached to the stoma and worn outside the body to collect stool. The pouch will need to be emptied several times each day.



If the surgeon removes the entire large intestine and connects the small intestine to the stoma, the surgery is called an **ileostomy**. If the surgeon leaves part of the large intestine and connects it to the stoma, the surgery is called a **colostomy**.

Later, during the pull-through procedure, the surgeon removes the stoma and closes the abdomen with stitches.



# What will my child's life be like after surgery?

## After Ostomy Surgery

Infants will feel better after ostomy surgery because they will be able to easily pass gas and stool.

Older children will feel better, too, but they must adjust to living with an ostomy. They will need to learn how to take care of the stoma and how to change the ostomy pouch. With a few changes, children with ostomies can lead normal lives.

However, they may worry about being different from their friends. A special nurse called an ostomy nurse can answer questions and show how to care for an ostomy.



An ostomy nurse can answer questions and show how to care for an ostomy.

## After the Pull-through Procedure

Most children pass stool normally after the pull-through procedure. Children may have diarrhea for awhile, and infants and toddlers may develop diaper rash, which is treatable with diaper creams. Over time, stool will become more solid and the child will go to the bathroom less often. Toilet training may take longer. Children often must learn how to use the muscles of the anus after surgery. Some children may leak stool for awhile, but most will learn to have better bowel control as they get older.

## Diet and Nutrition

After the pull-through procedure, children with long-segment HD need to drink more fluids. Now that the large intestine is shorter, or entirely gone, it is less able to absorb fluids the body needs. Drinking more helps make up for the loss.



After the pull-through procedure, children with long-segment HD need to drink more fluids.

Some infants may need tube feedings for awhile. A feeding tube allows infant formula or milk to be pumped directly into the stomach or small intestine. The feeding tube is passed through the nose or through an incision in the abdomen.

Eating high-fiber foods can help reduce constipation and diarrhea. Fiber helps form stool, making bowel movements easier. High-fiber foods include whole-grain breads, vegetables, and fruits. Some children may need laxatives to treat ongoing constipation. Consult a doctor before giving a laxative to your child.

## **Infection**

People with HD can suffer from an infection of the intestines, called **enterocolitis**, before or after surgery. Symptoms include

- fever
- swollen abdomen
- vomiting
- diarrhea
- bleeding from the rectum
- lack of energy

Call the doctor right away if your child shows any of these signs.



Children with enterocolitis need to go to the hospital. An **intravenous (IV)** tube is inserted into a vein to give fluids and antibiotics. The large intestine is rinsed regularly with a mild saltwater solution until all stool has been removed. The solution may also contain antibiotics to kill bacteria. A temporary ostomy may be needed to help the intestine heal.

Sometimes infection is a sign of a problem with the pull-through procedure. More surgery may be needed to correct the problem and prevent future infections.

## If I have more children, will they also have HD?

If you have a child with HD, your chance of having more children with HD is greater. Talk with your doctor about the risk.



## Points to Remember

- Hirschsprung disease (HD) is a disease of the large intestine that causes severe constipation or intestinal obstruction. People with HD are born with it.
- The large intestine, which includes the colon and rectum, is the last part of the digestive tract.
- The cause of HD is unclear. HD is not caused by anything a mother did while pregnant.
- The main symptoms of HD are constipation or intestinal obstruction, usually appearing shortly after birth.
- Newborns with HD almost always fail to have their first bowel movement within 48 hours after birth.
- HD is diagnosed based on symptoms and test results.
- HD is treated with surgery called a pull-through procedure.
- A child who has been very sick from HD may need an ostomy to get better before the pull-through procedure.
- Most children pass stool normally after the pull-through procedure.

- People with HD can suffer from an infection of the intestines, called enterocolitis, before or after surgery.
- If you have a child with HD, your chance of having more children with HD is greater.

## Hope through Research

The National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK) conducts and supports basic and clinical research into many digestive disorders, including HD. Scientists are studying the genetics involved in HD to improve diagnosis, treatment, and genetic counseling. They are also looking at therapies for treating enterocolitis, a major complication of HD.

Participants in clinical trials can play a more active role in their own health care, gain access to new research treatments before they are widely available, and help others by contributing to medical research. For information about current studies, visit *www.ClinicalTrials.gov*.

## Pronunciation Guide

**abdomen** (AB-doh-men)

**anemia** (uh-NEE-mee-uh)

**anus** (AY-nuhss)

**barium** (BA-ree-uhm)

**biopsy** (BY-op-see)

**colostomy** (koh-LOSS-toh-mee)

**constipation** (KON-stih-PAY-shuhn)

**diarrhea** (DY-uh-REE-uh)

**enemas** (EN-uh-muhz)

**enterocolitis** (EN-tur-oh-koh-LY-tiss)

**Hirschsprung** (HURSH-spruhng)

**ileostomy** (IL-ee-OSS-tuh-mee)

**intestine** (in-TESS-tin)

**intravenous** (IN-truh-VEE-nuhss)

**laxatives** (LAK-suh-tivz)

**manometry** (muh-NOM-uh-tree)

**obstruction** (ob-STRUHK-shuhn)

**ostomy** (OSS-tuh-mee)

**rectum** (REK-tuhm)

**stoma** (STOH-muh)

## For More Information

### **International Foundation for Functional Gastrointestinal Disorders**

P.O. Box 170864

Milwaukee, WI 53217-8076

Phone: 1-888-964-2001 or 414-964-1799

Fax: 414-964-7176

Email: [iffgd@iffgd.org](mailto:iffgd@iffgd.org)

Internet: [www.iffgd.org](http://www.iffgd.org)

### **Pull-thru Network, Inc.**

2312 Savoy Street

Hoover, AL 35226-1528

Phone: 205-978-2930

Email: [PTNmail@charter.net](mailto:PTNmail@charter.net)

Internet: [www.pullthrurnetwork.org](http://www.pullthrurnetwork.org)

### **United Ostomy Associations of America, Inc.**

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Fairview, TN 37062-0066

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Fax: 615-799-5915

Email: [info@uoaa.org](mailto:info@uoaa.org)

Internet: [www.uoaa.org](http://www.uoaa.org)

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The National Digestive Diseases Information Clearinghouse (NDDIC) is a service of the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK). The NIDDK is part of the National Institutes of Health of the U.S. Department of Health and Human Services. Established in 1980, the Clearinghouse provides information about digestive diseases to people with digestive disorders and to their families, health care professionals, and the public. The NDDIC answers inquiries, develops and distributes publications, and works closely with professional and patient organizations and Government agencies to coordinate resources about digestive diseases.

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