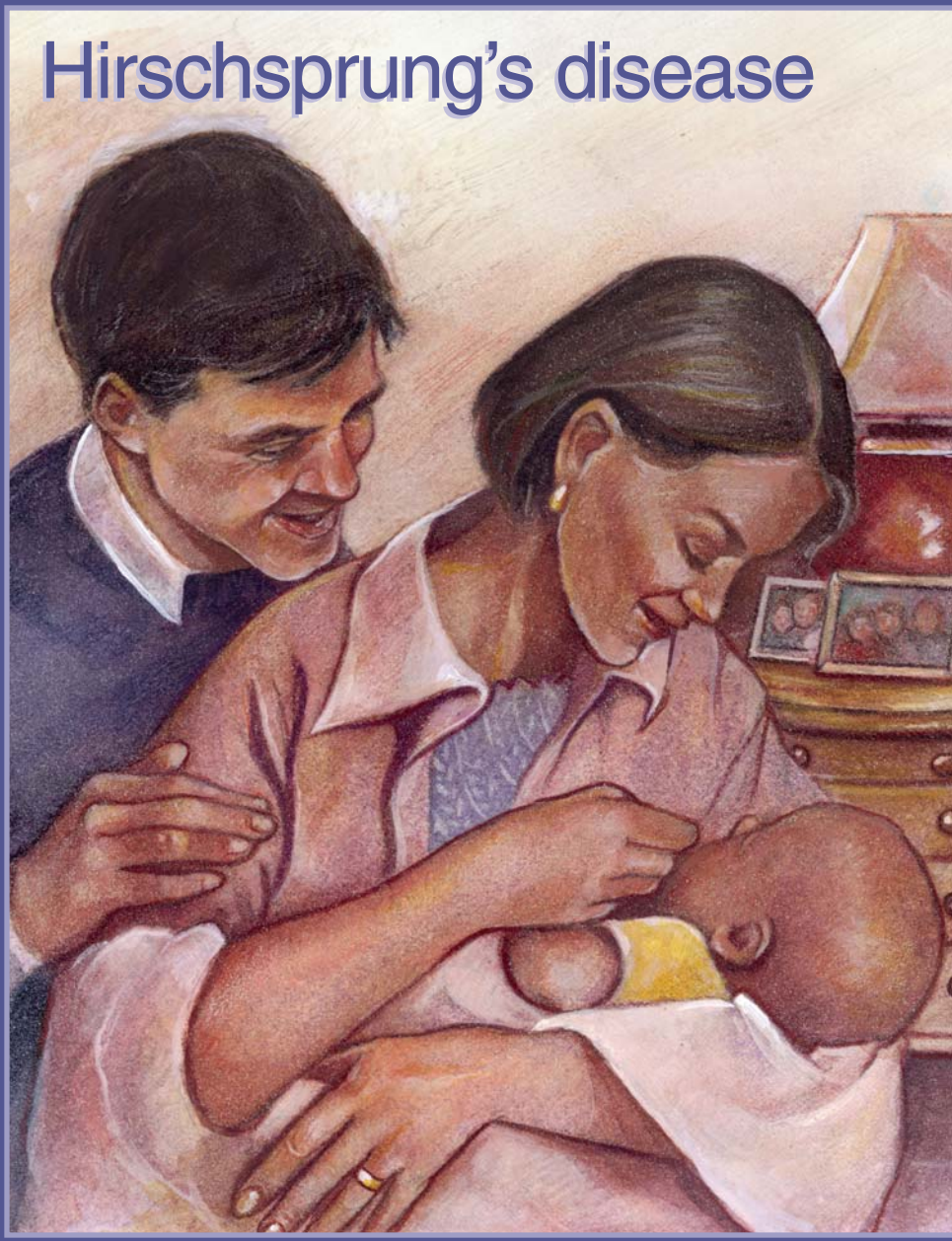
What I need to know about

Hirschsprung's disease



NIDDK
NATIONAL INSTITUTES OF HEALTH
National Digestive Diseases Information Clearinghouse

U.S. Department
of Health and
Human Services

What I need to know about Hirschsprung's disease

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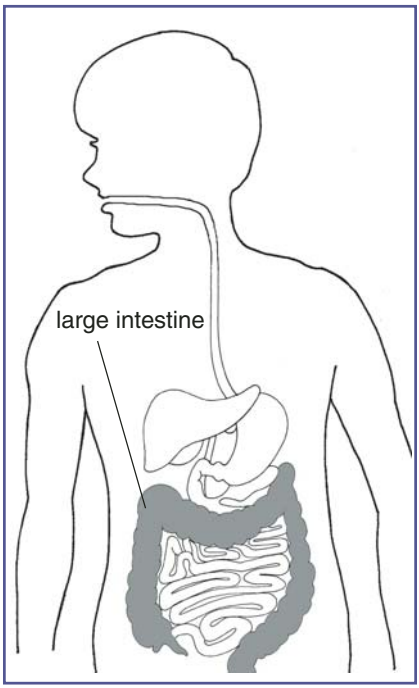
Contents

What is Hirschsprung's disease?

Hirschsprung's (HURSH-sprungz) disease, or HD, is a disease of the **large intestine.*** The large intestine is also sometimes called the colon. The word bowel can refer to the large and small intestines. HD usually occurs in children. It causes constipation, which means that bowel movements are difficult. Some children with HD can't have bowel movements at all. The stool creates a blockage in the intestine.



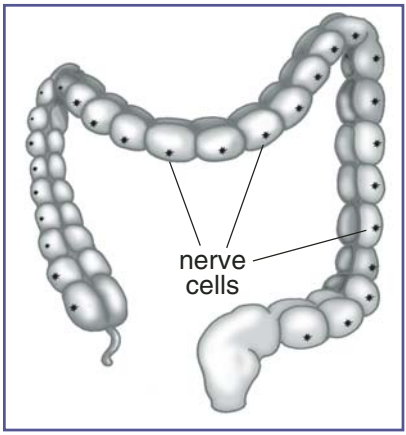
If HD is not treated, **stool** can fill up the large intestine. This can cause serious problems like infection, bursting of the colon, and even death.

Most parents feel frightened when they learn that their child has a serious disease. This booklet will help you understand HD and how you and the doctor can help your child.

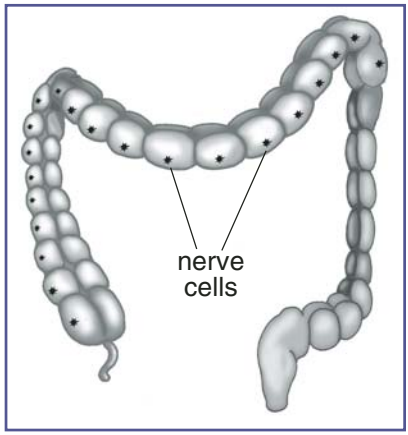
*Words in **bold** are defined on pages 19 and 20.

Why does HD cause constipation?

Normally, muscles in the intestine push stool to the **anus,** where stool leaves the body. Special **nerve cells** in the intestine, called **ganglion cells,** make the muscles push. A person with HD does not have these nerve cells in the last part of the large intestine.



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



HD large intestine: Nerve cells are missing from the last part of the intestine.

In a person with HD, the healthy muscles of the intestine push the stool until it reaches the part without the nerve cells. At this point, the stool stops moving. New stool then begins to stack up behind it.

Sometimes the ganglion cells are missing from the whole large intestine and even parts of the small intestine before it. When the diseased section reaches to or includes the small intestine, it is called long-segment disease. When the diseased section includes only part of the large intestine, it is called short-segment disease.

What causes HD?

HD develops before a child is born. Normally, nerve cells grow in the baby's intestine soon after the baby begins to grow in the womb. These nerve cells grow down from the top of the intestine all the way to the anus. With HD, the nerve cells stop growing before they reach the end.

No one knows why the nerve cells stop growing. But we do know that it's not the mother's fault. HD isn't caused by anything the mother did while she was pregnant.

Some children with HD have other health problems, such as Down's syndrome and other rare disorders.

If I have more children, will they have HD too?

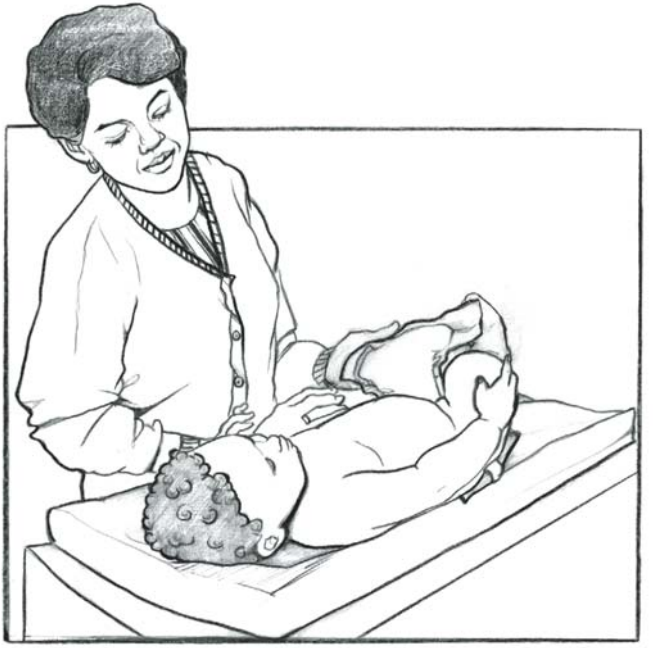
In some cases, HD is hereditary, which means mothers and fathers could pass it to their children. This can happen even if the parents don't have HD. If you have one child with HD, you could have more children with the disease. Talk to your doctor about the risk.

What are the symptoms?

Symptoms of HD usually show up in very young children. But sometimes they don't appear until the person is a teenager or an adult. The symptoms are a little different for different ages.

Symptoms in Newborns

Newborns with HD don't have their first bowel movement when they should. These babies may also throw up a green liquid called bile after eating and their **abdomens** may swell. Discomfort from gas or constipation might make them fussy. Sometimes, babies with HD develop infections in their intestines.



Symptoms in Young Children

Most children with HD have always had severe problems with constipation. Some also have more **diarrhea** than usual. Children with HD might also have **anemia,** a shortage of red blood cells, because blood is lost in the stool. Also, many babies with HD grow and develop more slowly than they should.

Symptoms in Teenagers and Adults

Like younger children, teenagers and adults with HD usually have had severe constipation all their lives. They might also have anemia.

Those with anemia look pale and tire easily.

How does the doctor find out if HD is the problem?

To find out if a person has HD, the doctor will do one or more tests:

- barium enema (BAR-ee-um EN-uh-muh) x ray
- manometry (ma-NOM-eh-tree)
- biopsy (BY-op-see)

Barium Enema X Ray

An x ray is a black-and-white picture of the inside of the body. The picture is taken with a special machine that uses a small amount of radiation. For a barium enema x ray, the doctor puts barium through the anus into the intestine before taking the picture. Barium is a liquid that makes the intestine show up better on the x ray.

In some cases, instead of barium another liquid, called Gastrografin, may be used. Gastrografin is also sometimes used in newborns to help remove a hard first stool. Gastrografin causes water to be pulled into the intestine, and the extra water softens the stool.

In places where the nerve cells are missing, the intestine looks too narrow. If a narrow large intestine shows on the x ray, the doctor knows HD might be the problem. More tests will help the doctor know for sure.

Other tests to diagnose HD are manometry and biopsy:

Manometry

The doctor inflates a small balloon inside the **rectum.** Normally, the anal muscle will relax. If it doesn't, HD may be the problem. This test is most often done in older children and adults.

Biopsy

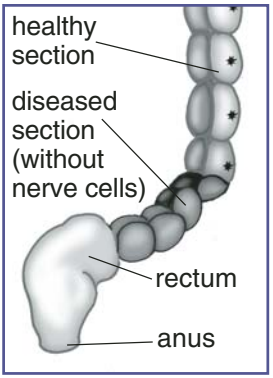
This is the most accurate test for HD. The doctor removes and looks at a tiny piece of the intestine under a microscope. If the nerve cells are missing, HD is the problem.

The doctor may do one or all of these tests. It depends on the child.

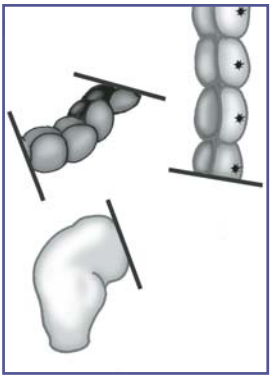
What is the treatment?

Pull-through Surgery

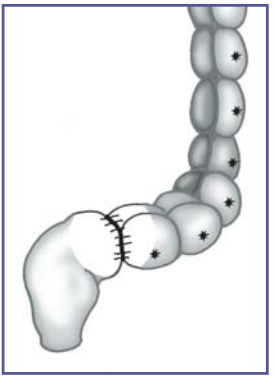
HD is treated with surgery. The surgery is called a pull-through operation. There are three common ways to do a pull-through, and they are called the Swenson, the Soave, and the Duhamel procedures. Each is done a little differently, but all involve taking out the part of the intestine that doesn't work and connecting the healthy part that's left to the anus. After pull-through surgery, the child has a working intestine.



Before surgery: The diseased section is the part of the intestine that doesn't work.



Step 1: The doctor removes the diseased section.

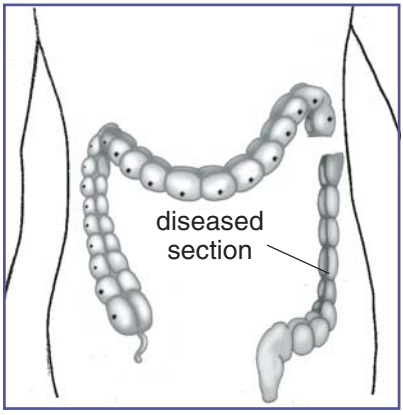


Step 2: The healthy section is attached to the rectum or anus.

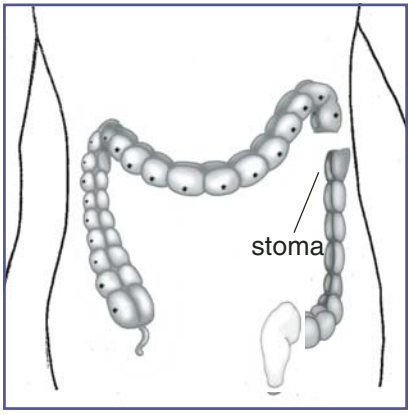
Colostomy and Ileostomy

Often, the pull-through can be done right after the diagnosis. However, children who have been very sick may first need surgery called an **ostomy.** This surgery helps the child get healthy before having the pull-through. Some doctors do an ostomy in every child before doing the pull-through.

In an ostomy, the doctor takes out the diseased part of the intestine. Then the doctor cuts a small hole in the baby's abdomen. The hole is called a **stoma.** The doctor connects the top part of the intestine to the stoma. Stool leaves the body through the stoma while the bottom part of the intestine heals. Stool goes into a bag attached to the skin around the stoma. You will need to empty this bag several times a day.



Step 1: The doctor takes out most of the diseased part of the intestine.



Step 2: The doctor attaches the healthy part of the intestine to the stoma (a hole in the abdomen).

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

Later, the doctor will do the pull-through. The doctor disconnects the intestine from the stoma and attaches it just above the anus. The stoma isn't needed any more, so the doctor either sews it up during surgery or waits about 6 weeks to make sure that the pull-through worked.

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

Ostomy

Most babies are more comfortable after having an ostomy because they can pass gas more easily and aren't constipated anymore.

Older children will be more comfortable, too, but they may have some trouble getting used to an ostomy. They will need to learn how to take care of the stoma and how to change the bag that collects stool. They may be worried about being different from their friends. Most children can lead a normal life after surgery.

Nurses at the hospital can teach you and your child how to care for a stoma and can talk to you about your worries.

Adjusting After Pull-through

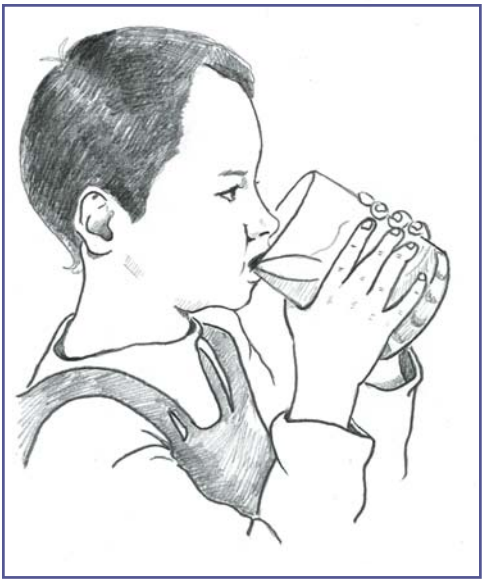
After a pull-through, 9 out of 10 children pass stool normally. Some children may have diarrhea for a while, and babies may develop a nasty diaper rash. Eventually the stool will become more solid and the child will need to go to the bathroom less often. Toilet training may be delayed, as the child learns how to use the bottom muscles only after pull-through surgery. Older children might stain their underwear for a while after the surgery. It is not their fault. They can't control this problem, but it improves with time.

Some children become constipated because 1 in 10 children with HD has difficulty moving stool through the part of the colon without nerve cells. A mild laxative may also be helpful. Ask your doctor for suggestions.

Diet and Nutrition

One job of the large intestine is to collect the water and salts the body needs. Since your child's intestine is shorter now, it absorbs less. Your child will need to drink more to make sure his body gets enough fluids.

An infant who has long-segment disease requiring an ileostomy may need special tube feedings. The shortened intestine does not allow the bloodstream enough time to absorb nutrients from food before it is pushed out of the body as stool. Tube feedings that deliver nutrients can make up for what is lost.



Drinking plenty of liquids is important after surgery for HD.

Eating high-fiber foods like cereal and bran muffins can help reduce constipation and diarrhea.

Infection

Infections can be very dangerous for a child with Hirschsprung's disease. Infection of the large and small intestines is called enterocolitis (EN-tuh-ro-ko-LY-tis). It can happen before or after surgery to treat Hirschsprung's disease. Here are some of the signs to look for:

- fever
- swollen abdomen
- vomiting
- diarrhea
- bleeding from the rectum
- sluggishness

Call your doctor immediately if your child shows any of these signs. If the problem is enterocolitis, your child may be admitted to the hospital. In the hospital, an intravenous (I.V.) line may be needed to keep body fluids up and to deliver antibiotics to fight the infection. The large intestine will be rinsed regularly with a mild salt water solution until all remaining stool has been removed. The rinse may also contain antibiotics to kill bacteria.

When the child has recovered from the infection, the doctor may advise surgery. If the child has not had the pull-through surgery yet, the doctor may prepare for it by doing a colostomy or ileostomy before the child leaves the hospital. If the child has already had a pull-through operation, the doctor may correct the obstruction with surgery.

Enterocolitis can be life threatening, so watch for the signs and call your doctor immediately if they occur.

Long-segment HD

Sometimes HD affects most or all of the large intestine, plus some of the small intestine. Children with long-segment HD can be treated with pull-through surgery, but there is a risk of complications such as infection, diarrhea, and diaper rash afterward. Parents need to pay close attention to their child's health. Also, since some, most, or all of the intestine is removed, drinking a lot of fluid is important.

Points to Remember

- HD is a disease of the large intestine.
- HD develops in children before they are born. It is not caused by anything the mother did while pregnant.
- Symptoms of HD include:
  - delayed first bowel movement in newborns;
  - swollen abdomen and vomiting;
  - constipation since birth;
  - slow growth and development; and
  - anemia.
- Children with HD may get an infection, called enterocolitis, which can cause fever and diarrhea.
- HD is a serious disease that needs to be treated right away. HD is treated with pull-through surgery or, sometimes, an ostomy.
- After treatment, most children with HD lead normal lives.

For More Information

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Glossary (by page)

Page 1

Large intestine (in-TESS-tin): A long tube that makes stool and carries it out of the body.

Stool: Solid waste from the body. The material that gets passed in a bowel movement.

Page 2

Anus (AY-nus): The opening at the end of the large intestine. Stool leaves the body through this opening.

Nerve cells: Nerves are long fibers that carry messages from the body to the brain, and back again, like telephone lines. The messages often tell a body part what to do. Nerve cells are part of nerves. In the intestine, the nerve cells tell muscles how to push the stool along.

Ganglion (GANG-li-on) cells: A type of nerve cell involved in moving stool through the large intestine. A person with HD is missing these cells from part of the large intestine.

Page 5

Abdomen (AB-duh-men): The area between the chest and the hips in the front of the body.

Page 6

Diarrhea (DY-uh-REE-ah): Loose, watery stool.

Anemia (ah-NEE-mee-ah): Not enough red blood cells in the blood.

Page 8

Rectum (REK-tum): The last section of the large intestine.

Page 10

Ostomy (OSS-tuh-mee): Surgery to connect part of the intestine to a hole in the abdomen.

Stoma (STO-mah): A hole on the outside of the body, made by surgery. Stool leaves the body through the hole, instead of through the anus.

Page 11

Ileostomy (il-ee-AW-stuh-mee): Surgery to connect the bottom of the small intestine (ileum) to a hole in the abdomen.

Colostomy (koh-LAW-stuh-mee): Surgery to connect the colon to a hole in the abdomen.

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Entercolitis (EN-tuh-ro-ko-LY-tis): Infection of the small and large intestines.

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