Let's Talk About...

Hirschsprung's Disease

What is Hirschsprung's disease?

Hirschsprung's [HERSH-prungs] **disease** (HD) is when the nerve cells in a child's intestines (bowels) do not form the right way. This usually happens in one of the last parts of the intestines called the **rectum**. The nerve cells are called **ganglion** [GANG-lee-on] cells. They help to move stool (poop) forward and relax the rectum so stool can pass from the body. If the child does not have ganglion cells in the lower part of the colon or rectum, stool backs up in the the colon. This can cause severe constipation, which can cause an infection or even create a hole (perforation) in the intestines.

The part of the intestines that does not have nerves could be short (just a part of the colon or rectum) or could include the entire colon and part of the small intestine. All children with HD need surgery to take out the parts of the intestines that do not work.

How often does HD occur?

HD occurs in 1 out of every 5,000 live births. It happens more often in boys than in girls. It is usually the only birth defect. It may occur in children with others syndromes, like Down syndrome.

Sometimes HD is inherited, and sometimes it is not.

Newborns can have the following signs of HD:

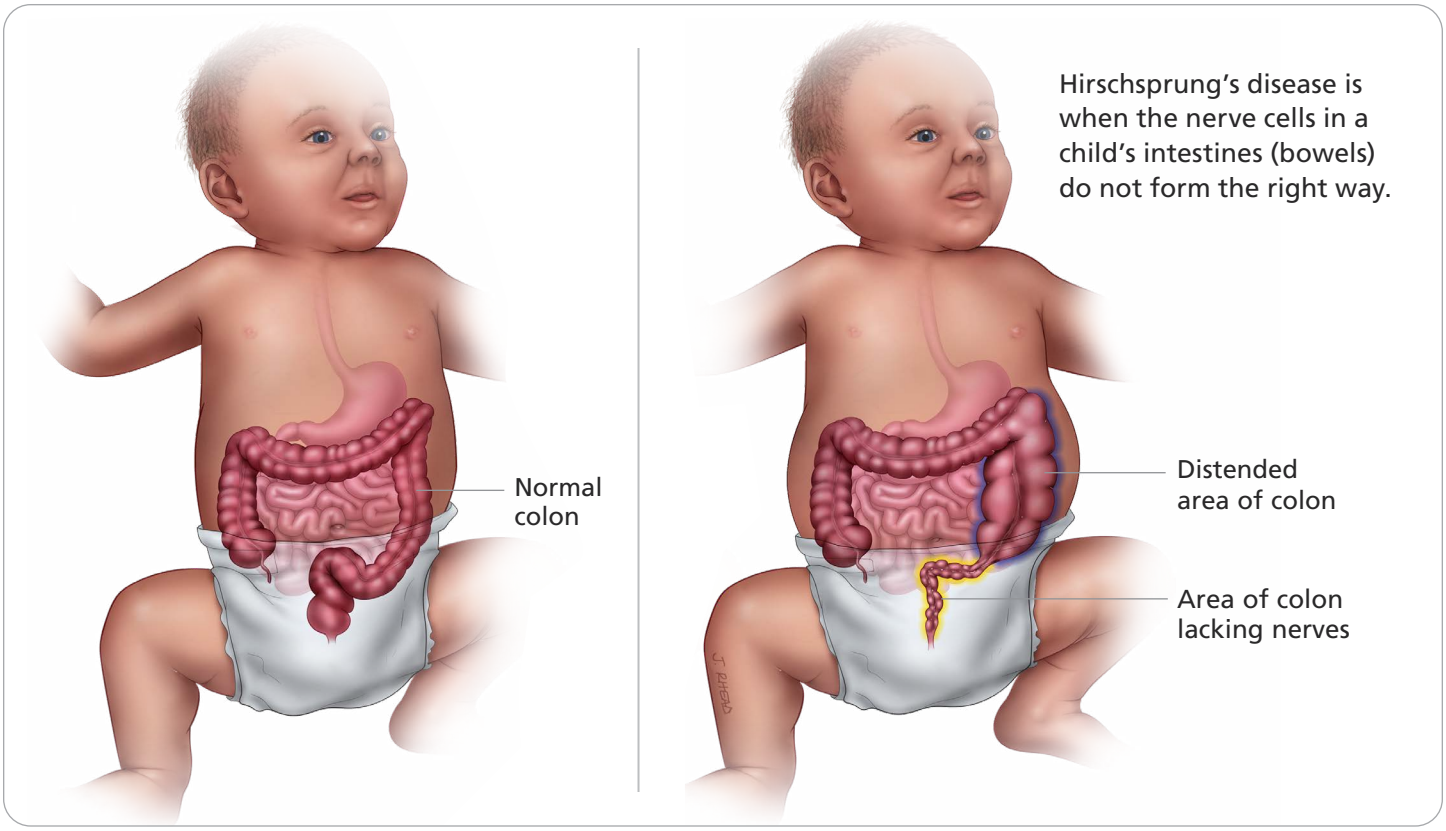
- Does not pass a stool (meconium) in the first two days of life.
- Belly gets bigger and fuller (abdominal distention).
- Refuses to eat.
- Vomits, has a fever with abdominal distension and/or failure to pass meconium.

Infants can have the following signs of HD:

- Does not grow normally.
- Does not have a good appetite.
- Belly gets bigger and fuller.
- Has diarrhea and vomiting.
- Has an infection in the intestines (enterocolitis) and may have explosive, watery diarrhea, fever, vomiting, and look very sick.

Children can have the following signs of HD:

- Ribbon-like stools that smell bad.
- Belly gets bigger and fuller.
- Does not eat enough.
- Does not grow enough.
- The doctor can feel a mass of stool during examination.



How does the doctor know my child has HD?

All children with HD have constipation, and there may be other signs. The doctor will examine your child and order some tests, including:

- **Abdominal x-ray.** This is an x-ray of the belly and can show if the intestines are blocked.
- **Contrast enema.** This is a special enema with contrast dye. The dye makes the intestines easier to see on x-ray. HD can cause the colon to be narrower or larger than normal at certain points.
- **Rectal biopsy.** This test must be performed to diagnose HD. The doctor will take a small bit of tissue from the rectum to be examined with a microscope to see whether or not it has nerve cells. In infants, a **suction rectal biopsy** may be done right in the child's hospital room or in the doctor's office. This does not hurt, because children cannot feel that part of the intestines.

  It is possible the child may need another biopsy in the operating room. This procedure tells the doctor whether the child has HD or not.
- **Anorectal manometry.** This test may be done in older children. It tells the doctor if there are normal reflexes in the rectum and anus (the last part of the intestines).

How is HD treated?

HD is treated with surgery.

- **Laparoscopic** [lap-ah-ruh-SKOP-ik] (minimally invasive) **surgery**, is done with tiny tools and a camera through one or more small incisions (cuts).
- **Open surgery** is done through a large incision in the belly.

During surgery, the surgeon removes the part of the intestine without ganglion cells. Then, the normal intestine is attached to the anus (called a **pull-through procedure**). The surgeon may do this in **one-stage or two-stage** surgery.

One-stage surgery is commonly used for newborns. The intestine without ganglion cells is removed and the normal intestine is attached to the anus.

Two-stage surgery is done to give your child a temporary **colostomy**. In stage 1, the surgeon makes a small hole in the belly and attaches part of the colon to it. This is called a **stoma** [STOW-mah]. A small bag is attached to the stoma so stool can leave the body empty into the bag. The child can eat and grow while the bowel heals and returns to normal size. In the second stage of surgery, the part of the intestine without ganglion cells is removed. The stoma is closed with stitches and the normal intestine is attached to the anus.

The surgeon will carefully consider whether your child needs one-stage or two-stage surgery. The choice is complicated. If the surgeon chooses one-stage surgery, you may have to give your child rectal irrigations for a while before surgery. Your surgeon will let you know which surgery is best for your child's individual needs.

If your child has any of the signs of entercolitis, tell your child's doctor immediately. Your child may need to have rectal irrigations to rinse stool and bacteria out of the colon, or be admitted to the hospital for IV fluids and antibiotics.

What kind of care will my child need after surgery?

If your child has the two stage procedure, the ostomy nurses at the hospital will teach you how to take care of the stoma and change the colostomy bag that collects the stool. You may need to:

- Protect your child's skin from loose stool with creams and powders.
- Change your child's diet or have them drink more water.

Your doctor and nurses will help you learn all the care your child will need.

What problems might my child have after surgery?

Diarrhea. After pull-through surgery, most children have diarrhea for a while. This may give a baby a diaper rash. You can help by using special creams and powders on your child's skin. After a while, your childwill have fewer bowel movements and their stools will get more solid.

Anus is too tight. Some children may have trouble with bowel movements because the area where normal intestine was sewn to the anus is tight. You may need to learn how to do anal dilations to help increase the anus size and prevent scar tissue from forming.

Toilet training issues. Some children toilet train later than usual. This is normal.

Need more water. Because the colon is shorter, your child may need to drink more liquids to help the colon absorb water and salts from the stool.

Tube feedings. Your child may need special tube feedings if a larger portion (or all) of the colon does not have nerve cells.

Infection. Your child may get a serious infection called **enterocolitis** [en-ter-coh-LIE-tiss]. This is when stool and bacteria back up in the colon. **Entercolitis is serious, and could be life threatening.**

Signs of entercolitis include:

- Fever
- Swollen belly
- Bleeding from the rectum
- Vomiting
- Diarrhea
- Sluggishness

What can I expect for my child's future?

Most chidren with HD have a small amount of colon that does not have ganglion nerves. This is called **short-segment disease.** These children usually have good bowel control over time. Immediately after surgery, these children have frequent stools (5 to 10 times a day). By 1 year after surgery, the child has 1 to 4 stools per day.

Enterocolitis can happen after surgery, but usually within 1 to 2 years after surgery.

Most (3 in every 4) children have problems being able to hold stool properly (fecal [FEE-kuhl] incontinence) or constipation. These can both be treated with a special diet, laxatives, or enemas.

A small number of children (1 in every 10) have bad constipation or fecal incontinence for a long period of time. These children may need a bowel management program. Children who have nerve issues or Down syndrome have more problems with fecal incontinence and severe constipation.

Notes