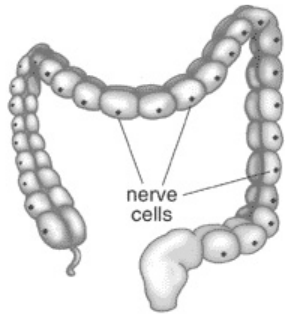




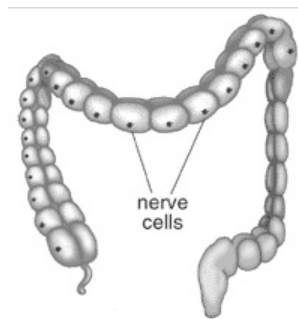
## What is Hirschsprung's disease?

Hirschsprung's disease is a congenital (present at birth) disease of the large intestine in which there is an absence of special nerve cells — called ganglion cells — that affect the muscles in the intestine. In healthy people, these muscles contract to push stool to the anus, where it leaves the body. People with Hirschsprung's disease, however, cannot move their bowels normally, leading to constipation. If Hirschsprung's disease is not treated, stool can back up in the large intestine, which can lead to serious problems such as infection and bursting of the intestine.

**This is a healthy large intestine. Nerve cells are located throughout the intestine.**



**This is an intestine with Hirschsprung's disease. Nerve cells are missing from the last part of the intestine.**



## How common is Hirschsprung's disease?

This disease occurs in approximately 1 in 5,000 births. It occurs more often in infants and children, but can occur in adults. It affects boys more often than girls.

## What causes Hirschsprung's disease?

Hirschsprung's disease develops when the baby is in the womb. It occurs when the ganglion (nerve) cells fail to grow through the entire intestine. It is not known why these nerve cells stop growing.

## What are the symptoms of Hirschsprung's disease?

Symptoms vary slightly depending on the age of onset. The most common symptom in newborns is difficulty having the first bowel movement after birth (meconium). A problem may develop when an infant switches from breast milk or formula to solid foods. Serious constipation is the first sign of Hirschsprung's disease in older children and adults.

## How is Hirschsprung's disease diagnosed?

The first step in diagnosing Hirschsprung's disease is to conduct a complete history and physical exam. Generally, the first diagnostic test performed is an un-prepped barium enema, in which a solution called

barium is used to make the intestine show up more clearly on an X-ray. This test can only suggest that Hirschsprung's might be the problem. A biopsy, in which a small piece of the intestine is removed for testing, can be used to make the definitive diagnosis. This test is most accurate because it shows whether or not there are any ganglion cells in that area of the intestine.

A biopsy can be obtained in smaller infants using a suction device. This can be performed in the doctor's office or at the bedside. For older children and adults, or those from whom an adequate specimen cannot be obtained at the bedside, a formal full-thickness biopsy must be performed in the operating room.

## How is Hirschsprung's disease treated?

Options for treating Hirschsprung's disease include:

- **Ostomy** — During this procedure, the doctor removes part of the diseased intestine. The doctor then cuts a small hole in the abdomen, called a stoma, through which stool can leave the body. A special ostomy bag is placed around the stoma to collect the stool. This procedure is temporary, and is often used until the child is well enough for surgery. In some cases, the child might respond to conservative treatment with enemas to stimulate bowel movements.

This approach makes the ostomy procedure unnecessary while the child is getting well enough for surgery.

- **Surgery** — The definitive surgery for Hirschsprung's disease involves removing the part of the large intestine without ganglion cells and replacing it with part of the large intestine that has the normal number of cells. This will allow proper bowel movements. This surgery is called pull-through surgery.

The standard surgical approach usually entails the formation of a stoma and then the subsequent one or two operations to perform the pull-through.

A newer alternative is treating the child conservatively and subsequently performing a one-stage pull-through procedure with laparoscopic assistance. For this procedure, the level at which the normal functioning bowel occurs is determined at the time of the operation with the assistance of gastrointestinal (GI) pathologists, doctors who studies cells and tissues to identify diseases and help with diagnoses.

## What happens after surgery?

After surgery, the child is treated with IV antibiotics while waiting for the return of bowel function. This usually occurs three to five days after surgery.