

## Patient Care > Hirschsprung's Disease (Colonic Aganglionosis)

### Introduction

The clinical presentation of absence of nerve cells (congenital aganglionosis) in the intestine was classically described by **Harold Hirschsprung of Copenhagen in 1886 and bears his name**. This occurs once in approximately 5000 live births and more in boys than girls. There may be a hereditary influence with a risk of ~5% of a second child having the disorder.

### Embryology/Pathology

**During intrauterine development, the nerves to the intestine do not develop properly.** Normal coordinated intestinal motion, or "peristalsis," depends on nerve cells embedded within the intestinal wall. Without these nerve cells, the bowel cannot propel the foodstuff or waste products. Instead of normal peristalsis, the segment of bowel without nerves often remains constricted and unmoving, resulting in a functional obstruction. The bowel behind this segment may become very dilated, but despite its abnormal appearance, this dilated bowel is the unaffected area.

**In Hirschsprung's Disease, nerve cells are usually absent just in the lower part of the large intestine** (from the rectum upward). Occasionally however, nerves may be absent (or abnormal) in the entire large intestine, or rarely the entire intestine.

### Symptoms/Signs

**The majority of patients with Hirschsprung's Disease are diagnosed in the neonatal period.** Since it is unusual for a full term and premature neonate not to have a stool within 24 hours and 48 hours, respectively, the first sign may be the failure to pass a first stool without assistance. This is followed by abdominal distension and vomiting.

**Sometimes the disease is subtle, and is not suspected until later.** In older children, there is almost always a history of never having had a normal bowel movement. Most of these children have a markedly protuberant abdomen. Advanced disease may result in watery diarrhea (as only liquid can get by the obstructed stool), colitis (bacterial overgrowth and inflammation of the colon), and even life-threatening sepsis (blood borne infection).

### Diagnosis

**The definitive diagnosis of Hirschsprung's Disease is made by taking small pieces of the lining of the bowel called a biopsy.** This is usually performed in the newborn by a suction biopsy instrument. This does not require

anesthesia and is usually done at the bedside in the nursery.

**In older patients**, a deeper biopsy is needed and is usually done in the operating room under general anesthesia.

The tissue is examined by the pathologist who determines whether nerve cells are present or absent.

**Prior to the biopsy**, your baby's doctors may order an abdominal x-ray and a contrast enema (an x-ray study performed with contrast placed into the rectum). Occasionally, anorectal manometry is performed which measures the pressures within the anorectal area.

## Treatment

**The basic principle in the treatment of Hirschsprung's Disease is to remove or bypass the segment of intestine without nerve cells and replace it with normal bowel.** In the stable newborn, after confirmation of the diagnosis, an endoscopic pull-through procedure using four to five small incisions may be performed. The operation is viewed by the surgeon on a television monitor. If significant bleeding occurs or if for anatomic reasons the operation cannot be performed laparoscopically then an incision is made and the procedure is performed "open".

**Occasionally, a baby or child is so sick that a definitive operation cannot be initially done.** In this case a colostomy, bringing a piece of the bowel to the surface of the skin, is performed. This relieves the obstruction and allows the patient to recover after which the pull-through procedure may be done safely.

If the pull-through procedure is difficult to perform, your surgeon may elect to do a colostomy with the pull-through to protect the operation diverting the stool. The colostomy is then closed in four to six weeks.

## Post-operative and Long-term Complications

**All operations under general anesthesia incur the risks of the anesthesia, bleeding, infection, and post-operative bowel obstruction.** Endosurgery has few complications and will be discussed with you by the surgeon. In addition, for the pull-through procedure, there may be blood supply insufficiency of the pull-through segment of intestine necessitating reoperation. As the new segment of intestine is sutured or sewn to the anal tissue, scarring and narrowing may occur which may require dilatations or operative revision of the connection. The majority (75-80%) of the patients do quite well eventually with normal bowel movements and no soiling. However, about 5% may continue to have severe constipation or incontinence which is usually treated by medical means of stool softeners, laxatives, enemas, and dilations.

**What is most important for all patients having had a pull-through procedure is to avoid constipation.** Constipation may lead to enterocolitis or an infection of the intestine which is quite serious and may be life-threatening. If

an infant or child becomes constipated, has small, watery stools with abdominal distension, and appears lethargic or has a fever, it is imperative that you contact us or your pediatrician. The best is, of course, to avoid constipation. This is particularly true for children when their diet changes (like on vacation, starting school, etc). You should always have glycerine suppositories and Pediatric Fleet® enemas with you when traveling.

**Patients with Down's Syndrome and neurologic impairment will have a higher incidence of constipation and incontinence and can only be treated with medical management and a great deal of patience.**

*Disclaimer: Your child's condition is unique. The information contained on this web site is not intended to substitute for advice from a doctor or nurse. If you are unsure about any aspect of your patient's care, please contact us at 303-839-6001, or talk to your pediatrician.*

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