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## Hirschsprung's Disease in Children and Adults

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# Hirschsprung's Disease in Children and Adults

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## At a Glance

- Hirschsprung's disease is a rare condition that people are born with.
- Symptoms usually appear in newborns, but may also appear in childhood or adulthood.
- Diagnosis is usually made within the first few months or years of life.
- Sometimes diagnosis is made in adulthood.
- Tests confirm the diagnosis.
- Treatment is surgical.
- Some symptoms may persist after surgery and need medical management.

## Introduction

Hirschsprung's disease is an illness that people are born with (congenital), in which there is a lack of nerve cells (ganglion cells) in the segments of the intestinal tract located in the colon or rectum. These abnormal segments produce obstruction that stops the movement of stool because of failure to relax during the wave-like motion (peristalsis) within the bowel. Hirschsprung's disease always starts in the anus (anal verge), but the length of the segment without ganglion cells (aganglionic) varies; most involve a short segment of bowel but some involve long segments. It is limited to the rectum and sigmoid colon in 75% of patients; involves the whole colon in 8%; and rarely involves the small bowel.

The incidence of Hirschsprung's disease varies from 1 in 5,000 to 1 in 10,000 live births. It appears to occur mostly in males with a ratio of 3:1 to 5:1. This is true particularly in those with short segments of the bowel that lack the normal nerve cells. However, the incidence in both sexes seems to be the same in those with long segments of the bowel affected.

The average age at the time of diagnosis has been decreasing over the years. In infants with Hirschsprung's disease about one-half are diagnosed in the first 3 months of life and more than three-quarters are diagnosed by 4 years of age. Occasionally, the diagnosis of Hirschsprung's disease is not made until adulthood. One study of 29 patients within an age range of 11 to 73 years reported the mean age at the time of diagnosis was 26 years. Most of these adults reported symptoms from an early age, but others reported the onset during adulthood. Most frequently, adults report constipation as the predominant symptom, and most evacuate the bowel only with enemas.

## Glossary

**Anus:** The opening of the rectum

**Barium enema:** A type of contrast enema for x-ray study of the lower intestinal tract

**Biopsy:** Removing tissue for diagnostic examination

**Bowel:** The intestines

**Colon:** The large intestine

**Enterocolitis:** Inflammation of the small and large intestines

**Fecal incontinence:** Involuntary leakage of liquid or solid stool

**Ganglion cells:** A group of nerve cells

**Intestines:** Also known as the gut or bowels, consisting of the small and large intestines

**Manometry:** A test that measures pressure or contractions in the intestinal tract

**Peristalsis:** The movement of the intestine characterized by waves which move contents forward

**Pull-through:** Surgical removal of the abnormal bowel and reattachment of the normal bowel just above the anus

**Rectum:** The lower end of the large intestine, leading to the anus

The symptoms vary with the age of the individual and the extent of the disease. In the newborn period, vomiting stained with bile, abdominal swelling, and failure to pass meconium (the first stools of a newborn within 24–48 hours after birth), or abnormal stool frequency are common. Complete intestinal obstruction and perforation of the colon where it meets the small intestine (cecum) or the appendix may occur. If the diagnosis is not established in the newborn,

the infant may present with mild constipation that may be followed with acute obstruction, frequent episodes of fecal impaction, or with the development of acute, and even life threatening, inflammation of the large and small intestines (enterocolitis). The latter develops in 15–50% of cases, and may be the presenting feature of Hirschsprung's disease in up to 12% of patients. It remains the main cause of death, and its mortality rate can reach 20–50%. From infancy until adulthood, mild to severe constipation may be the only symptom, so Hirschsprung's disease must be distinguished from functional constipation. Because clinical features do not allow a complete distinction between these problems, the diagnosis of Hirschsprung's disease must always be considered in any child, adolescent, or adult with severe constipation that does not respond to usual treatments.

## Diagnosis

Once the diagnosis is suspected, confirmation of the disorder is necessary. The final diagnosis needs to be based on the demonstrated lack of the nerve cells (ganglion cells) that control intestinal muscle contractions with the use of rectal biopsies.

*Biopsies* – Confirming the absence of ganglion cells in the diseased segment is a crucial step in the diagnosis of Hirschsprung's disease. There are different techniques to obtain small amounts of tissue from the rectum, and the test can usually be performed in an outpatient setting, although at times they may need to be performed in the operating room. Accuracy is excellent if the specimen is adequate, and if there is a trained pathologist, a physician who interprets and diagnoses the changes caused by disease in tissues and organs. When ganglion cells are present, the diagnosis of Hirschsprung's disease is excluded.

Because obtaining biopsies involves risks, other less invasive techniques such as anorectal manometry or a barium enema can be used to select those patients that require a biopsy.

*Anorectal manometry* – During anorectal manometry, a flexible balloon is introduced into the rectum, and the pressures are measured. When the balloon is inflated in normal individuals, the sphincter relaxes, mimicking the effect of stool. If the patient has Hirschsprung's disease, there is no relaxation of the sphincter muscle after the distention of the balloon. After the newborn period, manometry has been shown to accurately exclude or diagnose Hirschsprung's disease in 90–100% of the patients. Therefore, in this age group, anorectal manometry is the diagnostic study of choice to exclude Hirschsprung's disease; however, it is not widely available and is performed only in specialized centers. If the study is abnormal the diagnosis needs to be confirmed by biopsy. In newborns and premature infants the diagnostic accuracy is less (70–90%).

*Barium Enema* – The barium enema, a procedure in which barium is instilled in the rectum, and x-ray pictures are taken, while not diagnostic, can be strongly suggestive and

supportive. Barium enema is widely available. Single contrast barium enemas are used, and the colon is not prepared. In patients with a total lack of colonic ganglion cells the entire colon may appear normal. A barium enema may be less helpful in the newborn because a visible transition zone, the area between the normal colon and the part that lacks ganglion cells, is often not present. However, even if there is a normal barium enema, further work up may be indicated in those patients whose symptoms continue to not respond to treatment.

## Treatment

The treatment of Hirschsprung's disease is surgical. Initial medical management is important, however, in stabilizing the patient before surgical therapy is undertaken. This includes the correction of any fluid and electrolyte imbalances, antibiotic therapy if enterocolitis is present, decompression of the colon, and gentle rectal irrigations using soft rectal tubes until the time of surgery.

The basic principle for the definitive surgical therapy is removal of the segment of bowel that lacks ganglion cells followed by a pull-through of the bowel that does contain ganglion cells down to the anus. There are different procedures that have been used, but the three most common ones are: Swenson pull-through, Duhamel pull-through, and Soave pull-through. Recently the surgery is being performed in the newborn period using minimally invasive surgical techniques, like laparoscopy.

It is difficult to compare the results obtained with the three techniques, because the incidence of complications may be closely related to the skill of the individual surgeon, to the institution, or to the year of the study. Nevertheless, the long-term outcome of these procedures appears to be similar. Surgery for Hirschsprung's disease generally results in a satisfactory outcome. There are however, some patients that continue to have long-term difficulties. A recent review of 45 patients showed that 51% had some type of bowel dysfunction, and 38% fecal soiling. The most common symptoms are constipation, diarrhea, and sometimes periodic infections in the colon.

Satisfactory school performance is usually achieved and nearly all patients in one study appeared to be well adjusted. In another long-term study of 19 adolescents it was found that 32% had significant impairment of continence, but no more psychological or psychosocial dysfunction when compared with healthy controls. Fecal incontinence was associated with poorer psychosocial functioning, and parental criticism and psychosocial functioning was significantly correlated with the degree of fecal incontinence.

## Common Problems Found after Surgical Treatment of Hirschsprung's Disease

*Obstructive symptoms* – Of the postoperative symptoms occurring in children that have undergone surgical treatment

for Hirschsprung's disease, recurrent obstruction (manifested as constipation, abdominal distention, and difficulty having bowel movements) is one of the most common and difficult to manage. Treatment will depend upon the identified cause.

Obstructive symptoms may be related either to an anatomic problem, or to functional alterations. For example, there may be narrowing of the anus (anal stenosis), which can usually be managed with dilatation (stretching the opening with a special instrument), or there may be strictures, or narrowing, in the pulled-through bowel, which may require surgical correction.

The first consideration must be to determine whether the patient has an area remaining that lacks ganglion cells after an inadequate initial operation. Rarely, this may also be acquired or secondary after a successful initial operation.

Persistent internal anal sphincter (IAS) dysfunction, where the sphincter fails to relax, is one of the most common causes for obstructive symptoms. Persistent chronic obstruction from the IAS may also lead to recurrent enterocolitis, or bacterial overgrowth. The injection of Botox has become the treatment of choice for these patients. Frequent injections are necessary, and eventually a physician may recommend that the sphincter be cut. [A possible complication is incontinence.] However many children have long-term improvement after Botox alone.

If symptoms persist after treatment, or are not related to dysfunction of the sphincter, they may be due to generalized motility problems. In those patients a colonic manometry may be needed to pinpoint the extent and location of the motility problems. While controversial, some authors have also suggested that neuronal intestinal dysplasia (NID) type B, which is an abnormality of the intestinal nerves, may be associated with the presence of obstructive symptoms. Therefore in children with obstructive symptoms it may be necessary to obtain a full thickness rectal biopsy to exclude the diagnosis. In those children conservative management is indicated. However, if symptoms are severe, and clearly associated with the abnormal segment, surgical removal of the diseased portion of the intestines may be necessary.

Occasionally repeat surgery is indicated. This may involve removing the areas lacking ganglion cells, cutting the sphincter, redoing the pull-through surgery, or antegrade colonic enemas (ACE). The ACE involves a surgical procedure that creates a continent conduit from the skin to the colon at the cecum through which fluid used to wash out the bowel can then be easily self-administered.

*Fecal Incontinence* – Fecal incontinence is another frequent occurrence. It has been described in up to 30–80% of patients. In some it may be significant with constant

leaking, and in some less severe. The treatment of the fecal incontinence is complex. It includes treatment of constipation, biofeedback, and at times enemas, or more recently, antegrade colonic enemas.

*Enterocolitis* – Enterocolitis, infection or inflammation of the intestines, continues to be the major cause of complications in Hirschsprung's disease. It occurs after surgical treatment in 2–33% of patients, and can be life-threatening. It generally appears suddenly, with rapid progression, shock, and weakness. It may occur many years after surgery. Symptoms include abdominal distention, explosive diarrhea, vomiting, fever, lethargy, and rectal bleeding. The occurrence of explosive diarrhea in any patient with Hirschsprung's disease should suggest the diagnosis, even in the absence of other symptoms. The presence of postoperative enterocolitis needs to be recognized promptly, as the child can present initially with mild symptoms that are followed by a rapidly worsening course that may be fatal.

Diagnosis is facilitated by clinical exam, abdominal x-rays, and at times endoscopic examination. Non-operative management in the hospital is usually necessary.

## Conclusions

Since the first operative curative technique for Hirschsprung's disease was described in 1948 by Swenson, progress in diagnostic methods and surgical techniques have allowed the survival and successful treatment of most children with Hirschsprung's disease. In spite of these advances, postoperative problems continue to occur. Recently, research has produced a better understanding of the disease, knowledge that will undoubtedly lead to further refinements of the surgical techniques and better treatment of these patients.

*Note: This article is written for the American Neurogastroenterology and Motility Society (ANMS) and the International Foundation for Functional Gastrointestinal Disorders (IFFGD).*

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