

Hirschsprung's Disease

(Congenital Megacolon; Colonic Aganglionosis)

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En Español (Spanish Version)

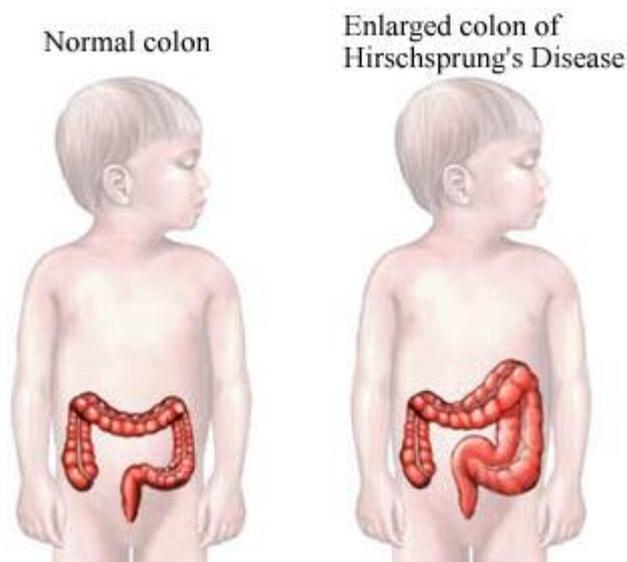
Definition

Hirschsprung's disease is a rare congenital disorder. It affects about 1 in 5,000 US newborns. The disease results in an obstruction of the bowel and prevents normal bowel movements. It occurs most often by itself but can also be part of a syndrome.

Causes

Hirschsprung's disease is caused by the absence of certain nerve cells. These cells called ganglia are in the wall of the bowel. Normally, these nerve cells help relax the bowel wall. The relaxation allows fecal matter to move through the colon. In children with this disease, a part of the colon stays contracted. This makes the bowel contents build up before the obstruction. The condition usually affects the last 1-2 feet of the colon that ends with the rectum.

Hirschsprung's Disease



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The absence of ganglia is due to a genetic defect. In some cases, Hirschsprung is hereditary. This means parents could pass it to their children. This can happen even if the parents don't have the disease. If you have one child with the disease, you could have more children with the disease. Talk to your doctor about the risk and consider genetic counseling.

Risk Factors

A risk factor is something that increases your chance of getting a disease or condition. Risk factors for Hirschsprung's include:

- Family members with the disease
- More common in males
- May be associated with other congenital defects

Symptoms

Hirschsprung's is usually diagnosed in infancy, but can also be found later. Symptoms can differ with age.

- **In newborns:**
 - Failure to pass meconium within the first 48 hours of life (Meconium is a dark sticky substance that is the first bowel movement.)
 - Vomiting after eating
 - Abdominal distention
- **In young children:**
 - Severe constipation
 - Diarrhea
 - Anemia
 - Growth delay
- **In teenagers:**
 - Severe constipation for most of their lives
 - Anemia

Diagnosis

Most cases of Hirschsprung's are diagnosed in infancy, although some may not be diagnosed until adolescence or early adulthood.

Tests for diagnosis may include:

- Barium enema —injection of fluid into the rectum that makes the colon show up on an x-ray so the doctor can see abnormal areas in the colon
- Biopsy —removal of a sample of bowel tissue to check for ganglia (or the absence of ganglia)
- Anorectal manometry—measurement of the pressures of the internal and external sphincter with a rectal balloon

Treatment

The primary treatment for Hirschsprung's is surgery to remove the affected portion of the colon. There are three potential phases to the surgery, but all three phases may not be needed. Your doctor will discuss the best methods for you or your child's condition.

The three phases are:

- Colostomy—This involves surgically creating an opening into the colon, which is brought to the abdominal surface. Stool contents are excreted through this opening and into a bag.
- Pull-through operation—The affected area of the colon is removed, then the healthy colon is brought down to the rectum and joined to the rectal wall.

- Closure of the colostomy—The colostomy opening is closed, and bowel function gradually returns to normal.

If your child is diagnosed with Hirschsprung's, follow your doctor's instructions.

Outcome

Symptoms are eliminated in 90% of children after surgical treatment. A better outcome is associated with early treatment, and shorter bowel segment involvement.

Complications

Complications may include:

- Perforation of the intestine
- Enterocolitis
- Short gut syndrome

Prevention

There are no guidelines for the prevention of Hirschsprung's.

RESOURCES:

International Foundation for Functional Gastrointestinal Disorders
<http://www.aboutkidsgi.org/>

National Digestive Diseases Information Clearinghouse
<http://digestive.niddk.nih.gov/>

CANADIAN RESOURCES:

Health Canada
<http://www.hc-sc.gc.ca/>

SickKids
<http://www.sickkids.ca/index.html/>

REFERENCES:

Behrman RE, Kliegman RM, Jenson HB, eds. *Nelson Textbook of Pediatrics*. 18th ed. Philadelphia, PA: WB Saunders Company; 2007.

DynaMed Editorial Team. Hirschsprung disease. EBSCO DynaMed website. Available at: <http://www.ebscohost.com/dynamed/what.php>. Updated June 23, 2010. Accessed November 17, 2010.

Kasper DL, Harrison TR. *Harrison's Principles of Internal Medicine*. 14th ed. New York, NY: McGraw-Hill; 1998.

Med Help International. Hirschsprung's disease. Med Help International website. Available at: http://www.medhelp.org/HealthTopics/Hirschsprung's_Disease.html. Accessed July 22, 2009.

National Digestive Diseases Information Clearinghouse. Hirschsprung's disease. National Digestive Diseases Information Clearinghouse website. Available at: <http://digestive.n...>. Accessed July 22, 2009.

Townsend CM, Sabiston D. *Sabiston Textbook of Surgery*. 17th ed. Philadelphia, PA: Saunders; 2004.

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