Hirschsprung's Disease

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Introduction

Your child has Hirschsprung's disease. This fact sheet was written to answer some of the questions you may have. We hope the information will help you better understand the disease and its effects on your child.

Hirschsprung's disease is named after the 19th century doctor, Harald Hirschsprung, who first identified it in 1886. Prior to 1948, the basis of the disease was not understood, and there was no surgical procedure to correct it. However, since that time, many children have been treated successfully and are living full and productive lives.

What is Hirschsprung's disease?

Hirschsprung's disease, or congenital intestinal aganglionosis, is a lack of nerve cell bodies (ganglion cells) in a segment of the bowel. This interferes with the coordinated squeezing action called peristalsis, which normally moves intestinal contents forward.

In the newborn period, diagnosis may be considered when an infant does not pass meconium (first stools of a newborn) within 24 hours of birth, repeatedly vomits material stained yellow or green with bile, or shows signs of abdominal distention (enterocolitis), or a history of constipation in an older infant or child.

How many children have Hirschsprung's disease?

Hirschsprung's disease occurs in one of every 5,000 births. In the majority of children, the disease is limited to the rectum or the rectosigmoid – often termed long segment disease. In very unusual cases, the entire colon and part of the small intestine may be affected – termed total colonic aganglionosis. There are more males than females with Hirschsprung's disease. However, in cases of long segment disease, the ratio of males to females is about equal.

What causes Hirschsprung's disease?

Form the fifth to the twelfth week of pregnancy, nerve cells migrate into the intestines. For some unknown reason in a baby with Hirschsprung's disease, the nerve cells fail to migrate in a segment of the bowel. There is a genetic factor involved, although most parents do not know of any relatives with Hirschsprung's disease. Approximately 10% of children with Hirschsprung's disease have a genetic-based condition such as Down's syndrome. Genetic factors are better understood in long segment disease than in short segment disease.

Nothing you did while pregnant brought on the disease. For example, painting a room, taking an aspirin, or sitting in a whirlpool did not cause your child to have Hirschsprung's disease. It happened regardless of anything you did or did not do.

How is the diagnosis made?

The diagnosis of Hirschsprung's disease will be confirmed by one or more tests, usually a barium enema and rectal biopsy. In cooperative adults or older children, manometry (a balloon study of internal anal sphincter pressure and relaxation) is helpful.

Will my child require surgery?

Yes. In some infants, a temporary colostomy or ileostomy may be performed. These are surgically created openings of the colon (large intestine) or ileum (lower part of the small intestine) to the body wall. The opening is called a stoma and is pink or red in color. A bag specially designed for collecting waste (appliance) is attached to the skin by an adhesive substance. Sometimes when an ostomy bag is not used, ointments and gauze placed around the stoma with a diaper covering are used as an alternative method.

Before your child is discharged from the hospital, an experienced professional such as an enterostomal therapist or ostomy nurse will work with you to teach you the proper techniques of the appliance and about general ostomy care. Once you have successfully changed the appliance or dressing a few times, you will find yourself comfortable with the process. The colostomy or ileostomy will not bother your baby. In fact, it will allow your child to pass gas and stool in comfort.

When your child reaches the weight, age, or condition desired by your surgeon, surgery will take place and the colostomy or ileostomy will be closed in one or two stages.

An increasing number of surgeons are now choosing to do the reconstructive operation in one stage without a stoma. This decision will be made by your surgeon, based on the extent of the abnormal bowel, the degree of dilation of the

normal bowel, the presence of enterocolitis, and his or her preference and training.

What kind of surgery will my child have?

For short segment Hirschsprung's disease, there are three common reconstructive operations: the Swenson, the Soave, and the Duhamel. All three procedures involve removing the abnormal (aganglionic) colon and reattaching the normal colon just above the anus (a "pullthrough" procedure). For long segment disease, some surgeons leave an extended length of aganglionic bowel, and sew the healthy bowel to it in a lengthwise fashion (the Marin or Kimura "colon patch" procedures), while others use the same kind of pullthrough that would be done for short segment disease.

Many children can have their pullthrough done without the need for large incision in the abdomen. These techniques are called "laparoscopic," "transanal," or "perineal" pullthroughs. As mentioned above, your surgeon may or may not choose to do a stoma initially. These decisions are based on a number of factors. It is a good idea to discuss the options with your surgeon so that you understand the reason for the particular approach being recommended for your child.

In the absence of complications, most children stay in the hospital anywhere from one to seven days following a pullthrough.

What medical follow-up will my child need?

Follow-up visits with your surgeon are important so that progress may be monitored and possible complications identified and dealt with. Also, keeping in touch with your child's pediatrician or family physician may be helpful. Remember, Hirschsprung's disease is uncommon, so your child may very well be your primary care physician's only Hirschsprung's patient.

We also suggest that you contact a pediatric gastroenterologist, especially if there are chronic problems. These specialists are often able to prevent minor problems from becoming medical emergencies. Your surgeon, pediatrician, or family physician can refer you to a pediatric gastroenterologist if necessary.

Will my child have problems after surgery?

Your child may have loose and frequent stools at first, and many children develop severe diaper rash. Maintaining good skin care in the anal area is imperative to help prevent rashes. As the stool becomes firmer, frequency should diminish. Older children may have lingering problems with soiling or constipation, but this condition generally improves with time. A visit to your doctor to assess for a contributing physical or dietary problem is always worthwhile.

Some children require rectal dilation for awhile after surgery If this is necessary for your child, your surgeon will teach you how to accomplish this either digitally, using a lubricated gloved finger, or with a dilator.

Colds or viruses may cause loose stools and parents have to be alert because there is an increased risk of dehydration, especially in children with long segment disease. In the most serious cases, hospitalization with intravenous (IV) therapy is necessary to restore fluid balance.

A potentially serious problem that sometimes occurs post-operatively is enterocolitis. Children with this problem will develop fever, abdominal distention, and diarrhea. It is important to go immediately to the doctor if this occurs, since delay in diagnosis and treatment can result in serious complications. Other common problems that may occur post-operatively are bacterial overgrowth (excess intestinal organisms) and lactose (milk sugar) intolerance.

Persistent diarrhea, abdominal distention, abdominal pain, and constipation may be symptoms of complications. It is important to seek medical help to prevent more serious problems.

Overall, about 90% of children with Hirschsprung's have no major complications or difficulties. Of the 10% who do have problems, most eventually get better with help from their doctor and other health professionals and go on to live a perfectly normal live.

Can my child be toilet trained?

Yes. A relaxed attitude on your part helps to foster your child's success.

Is there a risk for future children?

Genetic studies show that there is an increased risk for parents of Hirschsprung children to have additional children with the disease. The risk is about 3% for parents of a boy with short segment disease, 7% for parents of a girl with short segment disease, and 12% for parents of children with long segment disease.

Female siblings are less likely to be affected. A mother with Hirschsprung's disease is more likely to pass the disease on than an affected father.

These figures may change as more Hirschsprung children become parents. For the most accurate information, we recommend a thorough discussion of your individual case with a geneticist.

At the moment, there is no way to detect Hirschsprung's disease before birth based on ultrasound or genetic studies. As further research is done, this may become a possibility in the future.

In conclusion

Problems with Hirschsprung's disease are manageable in most cases. Your child needs to be doing all the regular things that everyone else likes to do. Feeling protective is normal, but try to relax and enjoy each other!

About IFFGD

The International Foundation for Gastrointestinal Disorders (IFFGD) is a 501(c)(3) nonprofit education and research organization. We work to promote awareness, scientific advancement, and improved care for people affected by chronic digestive conditions. Our mission is to inform, assist, and support people affected by gastrointestinal disorders. Founded in 1991, we rely on donors to carry out our mission. Visit our website at: www.iffgd.org or www.aboutkidsgi.org.

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About the Publication

Glossary of Terms	
Aganglionosis:	Absence of ganglion cells (neuron cell bodies) from the muscle wall.
Anastomosis (Intestinal):	The connection between two segments of intestine.
Anus:	The opening of the rectum.
Appliance:	A bag specially designed for collecting waste, and is attached to a stoma.
Barium Enema:	A type of contrast enema for x-ray study of the lower intestinal tract.
Bile:	Secretions of the liver.
Biopsy:	Removing tissue for diagnostic examination.
Bowel:	The intestines.
Colectomy:	The removal of a segment of or the entire colon.
Colitis:	Inflammation of the colon.
Colon:	The part of the intestines extending from the cecum to the anus. Also referred to as the large intestines.
Colonoscopy:	Visual examination of the inner surface of the colon.
Colostomy:	A surgically created opening from the colon to the skin.
Congenital:	Present at birth.
Dehydration:	Excessive loss of fluids in the body.
Dilation (Rectal):	Stretching or enlarging the internal anal sphincter with a special instrument or a finger.
Distention:	An uncomfortable swelling of the abdomen, usually caused by excessive amounts of gas and fluid.
Enterocolitis:	Inflammation of the small and large intestines. In children with Hirschsprung's disease, this can be a serious problem and requires early recognition and treatment.
Enterostomal Therapist:	A nurse specializing in ostomy and wound care.
Gastroenterologist:	A doctor who specializes in the field of gastroenterology.
Ileostomy:	A surgically created opening from the ileum to the skin.
Laparoscopic:	Pertaining to the introduction of a camera through a small incision in the abdominal wall.
Luminal:	Pertaining to contents of the intestine – a mixture of food, secretions, bacteria, and cellular debris.
Manometry:	Measuring pressures in the intestine or anus.
Meconium:	Black, tarry product of the first defecation of a newborn infant.
Motility:	Bowel wall movements and the transit of intraluminal contents.
Obstruction:	In Hirschsprung's disease, an anatomic blockage caused by the absence of nerve cells in the bowel that retards transit of luminal contents.
Ostomy:	An operation in which a stoma is created. Colostomy and ileostomy are types of ostomies.
Peristalsis:	The movement of the intestine characterized by waves which move luminal contents forward.
Pullthrough:	The removal of the aganglionic colon and reattachment of the normal colon just above the anus.
Rectosigmoid:	The junction of the sigmoid colon and rectum, or pertaining to both rectum and sigmoid; the last 20% of the colon.
Resection (Intestinal):	The surgical removal of a diseased portion of the intestine.
Soiling:	The involuntary passage of stool into clothing.
Sphincter (Anal):	Ring of muscle that opens and closes the anus.
Stoma:	A surgically created opening between the intestine and the surface of the body as a colostomy or ileostomy.
Toxic Megacolon:	Acute non-obstructive dilation and inflammation of the colon.