Hirschsprung’s Disease

In normal fetal development, bundles of nerve cells (Ganglion cells) form in the muscles of the intestine forming last at the end of the colon (rectum). In digestion, these ganglion cells help move the food through the colon via a wavelike motion called peristalsis.

If a person has Hirschsprung’s disease, these nerve cells stop forming at a point in the colon (large intestine). The colon cannot push the food past this point, creating a blockage or obstruction. The portion lacking ganglion cells always includes the rectum and extends upstream. This can be limited to the rectum, but can also include the entire colon.
Who Gets Hirschsprung’s Disease?

Hirschsprung’s disease is something a child is born with. The exact cause of Hirschsprung’s is unknown, but it is sometimes hereditary (inherited from the parents). It occurs in about 1 in 5000 births, and is more commonly found in boys than in girls.

Symptoms

**Typical presenting symptoms in a newborn baby:**
- Difficulty or straining with a bowel movement
- Failure to pass first stool (meconium) shortly after birth (within 1 or 2 days of birth)
- Infrequent, but explosive stools
- Poor feeding
- Poor weight gain
- Vomiting (sometimes a green/brown substance)
- Fever
- Gradual swelling of belly

**Typical presenting symptoms in older children**
- Constipation that gradually gets worse over time
- Small, watery and/or foul smelling stools
- Explosive bowel movements
- Loss of appetite
- Failure to gain weight
- Gradual swelling of the belly
- Fatigue
- Fever

How is Hirschsprung’s disease diagnosed?

There are three ways Hirschsprung’s Disease can be diagnosed:

1) **Contrast Enema with x-ray**
   A liquid contrast is gently administered via a soft rubber tube into the rectum (this is an enema). The contrast coats the inside of the colon and rectum so it shows up on an x-ray. The doctors can then look at the x-ray to see if they can identify any abnormalities that could suggest Hirschsprung’s Disease.

2) **Rectal Biopsy**
   The surgeon will remove a tiny piece of rectal tissue to learn whether the ganglion cells that control the bowel muscles are present in the tissue. This can often be performed in the clinic or at the bedside in the hospital with minimal discomfort.

3) **Rectal Manometry**
   This involves placing a small balloon in the rectum which can record pressure changes and muscle movements, helping to identify an area where there is a lack of muscle movements in the colon. This is most helpful in older patients.
How is Hirschsprung’s disease treated?

If the tests show the ganglion cells are missing at a certain point within the colon, it is usually recommended that your baby or child have surgery. The goal of the surgery is the remove the portion of the colon that doesn’t have nerve bundles, and then to connect the healthy areas of the colon together. This procedure is called a pull through (or primary repair). Some babies and children will require more than one surgery to correct Hirschsprung’s Disease (a staged repair) which could involve the creation of a colostomy.

A colostomy is an operation that connects the colon to the surface of the abdomen. Stool will drain into a collection bag that sticks to a colostomy site. In most cases a pull through procedure can later be performed, and the colostomy is closed. After a short period of time, your baby/child will begin stooling normally form the rectum.

Rectal irrigations (flushing out the colon from the bottom) is sometimes used to help relieve blockage of the colon and to keep stool from getting backed up. Rectal irrigations are sometimes necessary before and/or after surgery.

What can I expect after Surgery?

Your baby will have an IV (a very small tube that goes into a vein) for pain medicine, antibiotics and fluids to prevent dehydration. There will also be a tube in his or her nose that will go into the stomach, to help air/gas escape, and help keep the stomach empty. Once your baby/child has started passing gas on his or her own, the tube in the nose can be removed. He or she can begin drinking clear liquids, and possibly taking pain medications by mouth. If clear liquids are tolerated without vomiting, he or she can move on to formula, or regular foods.

Once your baby/child is able to eat enough, is having bowel movements, and isn’t running a fever, he or she can go home. Throughout your stay in the hospital, many people (nurses, doctors, case managers, etc.) will help you learn how to take care of your baby or child after surgery at home. We encourage you to ask questions and be as hands on as possible while in the hospital.
Caring for my child at home after surgery

After surgery, strenuous activity will need to be avoided. Quiet, supervised activity is best until the follow up appointment with the surgeon. At this appointment, appropriate activity levels can be discussed. Baths may also need to be avoided after surgery. This is something your child’s surgeon will discuss with you before going home.

At your follow up appointment in the clinic, your child’s surgeon may make the decision to begin anal dilations. After surgery, as the body heals itself, it creates scar tissue. This scar tissue can create a narrowing or stricture of the area the stool is meant to pass through. The anal dilations can help prevent this from happening. You will be taught how to perform these daily dilations for your child. A dilator is a small tubular instrument that comes in advancing sizes to gradually stretch the anus to normal size, so stool can easily pass through.

Nearly all children do very well; however, 10-20% of children may have continued problems with bowel movements, so continued follow-up with your surgeon is very important. Some kids may struggle with constipation, while others may have “accidents” in their underwear because they cannot hold their stool in. There are options for treatment if your child struggles with either of these problems, so it is important to bring either problem to the surgeon’s attention if they arise. Enterocolitis is an infection that is caused by a blocked colon. This is a common sometimes severe issue that can confront children with Hirschsprung’s Disease, see below to know what to watch for.

After your child has surgery, you should expect normal growth and development. You can follow up with your child’s pediatrician for regular checkups and other illnesses. If your child becomes ill with vomiting or diarrhea, be sure to remind your pediatrician that your child has Hirschsprung’s disease. (Remember, even though your child has had surgery, enterocolitis is still a possibility).

Call your surgeon for:

- Fever
- Irritability or lethargy
- Refusing to eat
- Abdominal distension
- Explosive, foul smelling stools
- Vomiting (especially yellow/green)

VERY IMPORTANT:

If your child has any episode of watery, explosive diarrhea with or without a fever, you MUST call the surgeon. This is a sign of Hirschsprung’s enterocolitis. Hirschsprung’s enterocolitis is a dangerous condition that can affect children with Hirschsprung’s disease before and/or after surgery. It becomes less common about a year after surgery. Once diagnosed, antibiotics and/or rectal irrigations may be used to treat the issue.
Further information/Support groups:

Hirschsprung’s Disease-Solving the Puzzle
By: Nicole B. Murphy (book)

http://www.pediatricsurgerymd.org (APSA family site)

http://www.pullthrunetwork.org/ (Support group for families)

http://www.mayoclinic.com/health/hirschsprungs-disease/DS00825

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