Hirschsprung Disease (aganglionosis)

Patient and family information, brought to you by the Education Committee of APSA

Overview - "What is it?"

Hirschsprung disease is a developmental disorder of the nerves of the intestine. The intestine normally contains nerves in its wall allowing the intestine to move contents forward for digestion and removal (bowel movement). The specific nerves of intestinal movement are called "ganglion cells". Ganglion nerve cells develop from special cells called neural crest cells that travel down the intestine while the baby develops in the mother. They are thought to travel from the upper to the lower intestinal tract. In Hirschsprung disease, this process is disturbed, and the nerves do not make it down to the end of the intestine, the colon and anus. Under the microscope, there are no ganglion cells in the wall of the affected intestine. 80-85% of children with Hirschsprung disease have the rectum and last part of colon (large intestine) affected. Sometimes, a longer segment of colon, the entire colon, or even part of the small intestine may have absent ganglion cells. If ganglion cells are not present in a part of the intestine, that intestine does not move contents forward causing constipation. The affected area acts like a blockage or obstruction. The baby can have a swollen belly, vomiting and inability to have bowel movements (stools) on their own.

- Hirschsprung disease occurs in about 1:5,000 children. It occurs more often in males vs females (ratio 4:1). Genetics can be a factor. It is typically diagnosed in newborns and infants, but can be diagnosed in adult years.
- Hirschsprung disease can be associated with other disorders such as: Trisomy 21, MEN2A, Waardenburg-Shah syndrome, Mowat-Wilson Syndrome, Goldberg-Shprintzen syndrome, central hypoventilation syndrome and Smith-Lemil-Opitz malformation syndrome.
- Types of Hirschsprung disease:
    - *Short segment:* 80% of Hirschprung disease patients have only a small part of their colon without ganglion cells, and most commonly the connection of colon and rectum (rectosigmoid junction) is involved.
    - *Long segment:* When the affected colon is at least 1/3 of the colon length.

Total colon aganglionosis: 5-10% of cases. The entire colon lacks ganglion cells. Rarely, the colon and a large portion of the small intestine has no ganglion cells, leaving only 10-40 cm of normally innervated small bowel.

Signs and Symptoms - "What symptoms will my child have?"

Early signs:

- ***Obstruction*:** Intestinal blockage in a newborn baby: 50-90% of children present during the neonatal period with a swollen belly, bilious vomiting when feedings are attempted and no passage of meconium (stool) for the first 48 hours of life.
- ***Enterocolitis*:** 10% of children with Hirschsprung disease present with an intestinal infection (enterocolitis). Since the stool is not evacuated effectively out of the body through bowel movements, bacteria multiplies in the intestines. This results in fever, abdominal swelling, diarrhea or no stools, and even spread of bacteria into the blood. Enterocolitis maybe seen in infants or older children.
- ***Perforation*:** Because the distal bowel cannot relax and decompress, the normal intestine cannot empty its contents. The normal intestine becomes very distended and can rupture (perforate). There will be air and stool leaking into the abdominal cavity and this can be diagnosed on X-ray. The surgeon will recommend emergent surgery.

Later signs/symptoms:

- ***Chronic constipation*:** Some children with Hirschsprung disease present later in childhood with chronic constipation. This is most common in breastfed infants that typically develop constipation around the time of weaning. Constipation in older children can lead and present as failure to thrive (not gaining weight appropriately), abdominal distension and dependence on enemas, suppositories or "rectal stimulation" to pass stool. Unlike older children with behavioral associated constipation and stool holding, children with undiagnosed Hirschsprung associated constipation generally do not soil their underwear.

Diagnosis - "What tests are done to find out what my child has?"

Diagnosis of Hirschsprung disease is based on the patient's history, X-ray studies and a rectal biopsy.

Abdominal X-ray: Plain radiographs may show dilated intestines or other findings suggestive of obstruction

Contrast enema: In this study, a tube is gently placed in the rectum. Liquid dye (contrast) is injected through the tube into the rectum and colon. The radiologist takes pictures and looks

for narrowing of intestine (area where the nerves are not present) along with a distended/dilated intestine before the abnormal segment. This region where there is a difference in caliber of intestine (narrow abnormal segment and dilated normal segment) is called the "transition" zone. The study approximates the length of colon without ganglion cells. Though this test can suggest and be consistent with Hirschsprung's disease, a definitive diagnosis cannot be made without a tissue sample, or biopsy.

Rectal biopsy: is a procedure taking pieces of the inside of the rectum to be sent to the pathologist, who is a doctor who looks at the tissue under a microscope. Patients are diagnosed with Hirschsprung disease if there is a lack of ganglion cells in the tissue. There are two biopsy techniques used:

- ***Suction Rectal Biopsy*:** can be performed at the bedside without general anesthesia. A small instrument is inserted in the anal opening, pushed in about 2 cm and pieces of the intestinal lining are sampled (biopsy).
- ***Incisional Rectal Biopsy*:** a larger segment of rectal tissue is obtained (full thickness rectal biopsy). This is typically performed on older children and done under anesthesia.

Anorectal Manometry: This test is usually performed by a pediatric gastroenterologist (specialist). In this study, the relaxation of the sphincter muscle will be measured. Lack of relaxation is suggestive of Hirschsprung disease.

Conditions that mimic Hirschsprung Disease:

- In infants: meconium plug syndrome, meconium ileus
- In older children: regular constipation

Treatment - "What will be done to make my child better?"

The treatment of Hirschsprung disease is surgery, but there are several preoperative interventions considered prior to surgery.

Medical Treatment: If the child has evidence of enterocolitis (infection), the first priority is fluids and antibiotics. If the baby is vomiting and/or the belly is distended, a tube is placed into the nose down into the stomach to evacuate air and contents. Children with enterocolitis, or those in whom immediate surgery cannot be done for other reasons, should undergo bowel decompression by rectal irrigations. Irrigations are performed by gently placing a rubber tube into the bottom and flushing with water while carefully advancing the tube. This helps remove stool and bacteria from the intestine.

- Total or near total aganglionosis: In some cases there is not enough functional intestine to support nutrition and growth. These children must be fed from birth through the vein with total parenteral nutrition (TPN). The surgeon may recommend a stoma in the area where there are ganglion cells. These children are considered to have intestinal failure.

They require detailed care from physicians, usually pediatric gastroenterologists, with experience caring for intestinal failure patients.

Surgery: Surgery to remove the affected intestine is the definitive treatment of Hirschsprung disease. However, depending on many factors, surgery may be postponed (for baby to grow as one example), performed in one procedure, or performed in two staged fashion. The most common methods are below:

- Single-staged procedure: A "pull-through procedure" is the definitive procedure for Hirschsprung disease. The main goals of this procedure are to remove the abnormal segment of bowel and connect the intestine with nerves to the anus. There are several operations to accomplish these goals (names are Soave, Swenson and Duhamel). Which type of pull-through is best for your child depends on many factors and will be discussed by your surgeon.
- Two-staged procedure: A stoma or ostomy may be necessary in certain situations before the pull-through procedure. A stoma is a surgery that sews the end of the colon to the abdominal wall, bypassing the affected colon allowing stool to drain. For children with severe enterocolitis (infection), perforation (a hole in the bowel from high pressure), poor nutrition, or an older child with extensively dilated proximal bowel, a temporary stoma may be recommended because definitive surgery would have higher risks of failing/complications. After the stoma, then the pull-through procedure is done at a later date based on safety.
- Laparoscopic pull-through surgery: In laparoscopic surgery, several small cuts (incisions) are made. Through one of the cuts, a video camera is placed. The surgery itself is done using small instruments placed through the other incisions. The usual number of incisions (cuts) for laparoscopic surgery vary.
- Open pull-through surgery (laparotomy) uses a larger incision, either vertical or transverse to perform the operation.

Preoperative preparation: If the operation is scheduled (elective), a bath or shower is recommended the night before or the morning prior to surgery. The child will not eat anything for 6-8 hrs before surgery. You will be instructed on the details by the surgeon. Intravenous antibiotics will be given in the operating room.

Postoperative care: Once the child has stool out of the bottom or stoma, diet is resumed. Activity is normal. Medications for pain will be given.

- Surgeons may recommend thick creams around the bottom to protect the surrounding skin from bowel movements.
- After a pull-through surgery, nothing should be placed into the rectum for 2-3 weeks – including medications and temperature probes.

Risks of surgery: The surgeon will observe for the following possible complications:

- Wound infection

- Bleeding
- Anastomotic leak: The new connection between the normal colon to the anus leaks or falls apart.
- Stricture (scarred narrowing of the where the new connection was sewn)
- Bowel obstruction: Can be due to scar tissue from surgery, a twist in the pull-through, or the new connection where the bowel is pulled through can scar tightly and cause a blockage. Symptoms of obstruction are abdominal distension, nausea, vomiting.
- Fistulas: Connections between bowel and bladder, or bowel and vagina, have been reported to develop after operation. These are rare problems.

Benefits of surgery: Ability to have normal bowel movements.

Long term complications:

- *Stricture* or narrowing at the bowel connection site or anastomosis. Child may need procedures to dilate the stricture
- *Enterocolitis* – even after the affected bowel is removed, there remains risk of infection with constipation.

Home Care - "What do I need to do once my child goes home?"

Diet: Your child should be able to resume a normal diet without restrictions.

Activity: Your child should avoid strenuous activity and heavy lifting for the first week after laparoscopic surgery, 4-6 weeks after open surgery

Wound care: Surgical incisions should be kept clean and dry for a few days after surgery. Most of the time, the stitches used in children are absorbable and do not require removal. Your surgeon will give you specific guidance regarding wound care, including when your child can shower or bathe.

Medicines: Medicines for pain such as acetaminophen (Tylenol®) or ibuprofen (Motrin® or Advil®). Stronger pain medications like a narcotic may be needed to help with pain for a few days after surgery. Stool softeners and laxatives are needed to help regular stooling after surgery, especially if narcotics are still needed for pain.

What to call the doctor for: Call your doctor for worsening belly pain, fever, vomiting, diarrhea, worsening constipation, problems with urination or if the wounds are red or draining fluid.

Follow-up care: Your child should follow up with his or her surgeon 2-3 weeks after surgery to ensure proper postoperative healing.

- The surgeon will check the anastomosis (rectal stitches) at about two weeks after the pull-through. The surgeon will ask questions to make sure your child is not showing

signs of enterocolitis after surgery. Some parents will need to be taught how to dilate the anastomosis to prevent narrowing.
- Barrier cream: This is needed for some time, as diaper rash can be severe.

Long Term Outcomes - "Are there future conditions to worry about?"

Occasional problems related to the bowel are fairly common in the first few years of life. The surgeon will follow the child until after the toilet training process, longer if needed.

Ongoing obstructive symptoms: There are a range of obstructive symptoms can occur after a pull-through.

- *Mechanical obstruction*
    - Stricture: If the anastomosis scar tightens and narrows, sometimes it can be dilated. Surgery may be needed.
    - Twist: If a twist in the pull-through is identified, surgery will be needed to correct it.
- *Persistent aganglionosis*: This is usually due to pathology interpretation in evaluating the tissue under a microscope, or lack of blood flow/inflammation of previously normal bowel causing nerves to die. A repeat biopsy may show that not all the colon without ganglion cells were removed during pull-through surgery.
    - Treatment: Surgery will be needed to remove the affected bowel and place the pull-through at the correct level.
- *Motility Disorder*: Children with Hirschsprung disease have a higher incidence of the normal intestine not functioning appropriately and moving contents through the intestinal tract. This leads to the bowel not working normally even though there is no obstruction, and even if there are normal ganglion cells in the rectum after pull-through operation.
    - Treatment: These children may benefit from evaluation by a pediatric gastroenterologist and recommendations for bowel management.
- *Internal sphincter achalasia*: All children with Hirschsprung disease have an absent rectal inhibitory reflex, (the anal sphincter may not relax normally when trying to use the restroom), but it is unclear why some develop obstructive symptoms from this, and some do not. Sometimes the sphincter is tight, the bowel dilates and the patient has obstructive symptoms.
    - Treatment: Most children outgrow the problem by age five years old; however, the surgeon may recommend Botox injection of the sphincter. Botox relaxes the sphincter but is only good for a few months. This diagnoses and treats the problem and is generally successful.

- *Functional megacolon:* Stool-holding behavior is a common cause of constipation after pull-through operation. Passing hard, painful stools can start a cycle of withholding stool behavior.
    - Treatment: A bowel management program may be recommended.
- *Soiling*: The surgeon may recommend studies to have a clear understanding of the cause of the soiling. Physical exam including rectal exam, X-rays, contrast enema and manometry may be recommended. The surgeon will determine the quality of the sphincter and degree of normal rectal sensation.
    - Treatment will be based on whether the soiling is due to a sphincter problem or rectal sensation problem, slow transit through the bowel with overflow of stool, stool holding behavior with overflow, or hyperperistalsis (rapid transit of stool due to abnormal motility).
- *Enterocolitis*: Enterocolitis may present both before and after surgical correction of Hirschsprung disease. It is the most common cause of death in children with Hirschsprung disease, and more common in younger children, those with long segment disease and children with Down's syndrome. Symptoms include fever, abdominal distension and diarrhea. X-rays will look abnormal.
    - Treatment: involves a tube in the nose down into the stomach (NG tube), antibiotics, intravenous fluids and decompression of the colon and rectum, usually by irrigations. Enterocolitis can be minimized by the routine use of irrigations. Some surgeons also prescribe a regular dose of an antibiotic. If any of the above symptoms occur, the surgeon or a physician should be contacted immediately.

Resolution over first five years: Obstructive symptoms, soiling and enterocolitis (in the absence of obstruction), usually resolve in the first five years of life. Studies of teenagers suggest social satisfaction and quality of life normalize by the late teen years.

Ongoing incontinence, enterocolitis and dehydration are more common in children with long-segment disease, Down's syndrome or other causes of neurological impairment. Their long-term results are less satisfactory.

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