Hirschsprung's Disease

Aganglionosis

Hirschsprung's disease is name for Harald Hirschsprung, a Danish pediatrician who was the first to describe this condition in 1886. Also, it is known as “aganglionosis” because one of the main characteristics in all these patients is that they do not have ganglion cells in the rectum.

What is Hirschsprung's disease?

Hirschsprung's disease is the congenital absences of ganglion cells in the rectum and the lack of these cells causes an obstruction of the large bowel. These babies may present at birth a delay to pass meconium (a dark green poop in newborns) for more 24 hours or have abnormal bowel movements associated with abdominal distention and vomiting.

Is Hirschsprung's disease alike in all patients?

Hirschsprung's disease is not alike in all patients. Although congenital aganglionosis always affects the rectum, which is the final part of the large intestine, it can be extended to longer segments or even to the entire large intestine.

For a better description, please see the anatomical illustration of a normal colon below, and then compare to illustrations of some forms of Hirschsprung's disease.

How do I know if my child could have Hirschsprung's disease?

In a simple way, children with Hirschsprung's disease do not have normal bowel movements, most of them since the neonatal period. A wide-range of common problems can sometimes makes it difficult to achieve a timely diagnosis. Some children can improve using laxatives or suppositories and others may require enemas. Moderate to severe cases may present with abdominal distention, fever, vomiting, and dehydration, in a life threating condition named Proliferative Obstructive Colitis or Enterocolitis.

How is Hirschsprung’s disease confirmed?

Currently, Hirschsprung’s disease can be confirmed with an adequate rectal biopsy that should be studied by an experienced pathologist. During this procedure we take a small fragment of the rectum, obtaining a result in 1 or 2 days.

How do we know the length of the affected colon?

Once we have confirmed the aganglionosis of the rectum, it becomes necessary to perform a radiological study called a “Contrast Enema.” This study should be performed by an experienced pediatric radiologist to achieve our goal: demonstrate the narrowed proximal segment and the zone where the dilated segment arise, this area is called the “Transitional Zone.”

Are the symptoms of Hirschsprung's disease similar to those of other medical problems?

Yes, Hirschsprung's disease has symptoms that are similar to constipation, allergies to some components of formula lactose intolerance, enterocolitis, and intestinal infections, among others.

Approximately 70% of babies born with Hirschsprung's disease have only this segment without ganglion cells. Note the narrow aganglionic rectum (called distal segment) creating obstruction and, above this segment, observe the obstructed and dilated large bowel (called proximal segment) which contains ganglion cells or called normoganglionic colon.

Normal colon

Aganglionosis of the rectum

Approximately 20% of babies born with Hirschsprung's disease have this form of aganglionosis. Observe a larger aganglionic distal segment causing obstruction and the proximal normoganglionic dilated colon (named in this case transverse colon).

Aganglionosis of rectum, sigmoid, and left large bowel (also called a descending colon).

This form may affect 10% of babies born with Hirschsprung's disease. The entire colon is narrow causing obstruction and the small bowel (ileum) is the dilated intestine.

Total colonic aganglionosis

How is Hirschsprung's disease treated?

As we previously described, Hirschsprung's has different clinical expressions as well as affected length segments so, we are going to describe the “standard” approach remarking some variables in each stage.

Stage 1: Relieve the obstruction

Rectal tube - More than 70% of newborns with Hirschsprung's disease have only an aganglionic rectum, consequently the obstruction could be resolved with a rectal tube avoiding a colostomy and offering patients the possibility to correct the condition with one surgical procedure commonly called a “One-stage pull-through”. The rectal tube is placed into the rectum allowing the gas and stool to come out, also it is used to instill saline solution into the colon.This procedure could be maintained for several days or even weeks until a comprehensive diagnosis is established.

Colostomy - An intestinal stoma is needed in newborns or infants when a rectal tube does not resolve the obstruction; most commonly, this situation occurs in patients with larger aganglionic segments and usually an abdominal exploration is needed.

Ileostomy - When a total colonic aganglionosis is suspected or confirmed, the last segment of the small bowel, named ileum, is brought out onto the surface of the skin creating a stoma.

Stage 2: Pull-through

All types of Hirschsprung's disease need a surgical procedure achieving 3 surgical principles:

1. to remove the aganglionic segment,
2. to mobilize a normoganglionic intestine through the pelvis to the anal canal and

3. to perform an anastomosis -surgical union- close to the dentate line; these steps commonly are called “the pull-through” and there are different techniques to perform it.

Usually, the colon is pulled-through however, the small bowel is connected to the anal canal in patients with total colonic aganglionosis, in these particular patients, some surgeons use a portion of the aganglionic colon to create a mixed tube with small bowel, this is called “colon patch.” (We typically do not recommend a colon patch because they frequently produce obstructive problems.)

Stage 3: Close the colostomy or ileostomy

The stomas could be closed at the time of the pull-through, if the surgeon decides not to do it; colostomy/ileostomy is closed 4-8 weeks after the pull-through.

The surgical treatment of Hirschsprung's could be made as:

- One-stage pull-through (e.g. primary pull-through)
- Two-stages pull-through (e.g. colostomy and pull-through) or
- Three-stages pull-through (e.g. colostomy, pull-through and colostomy closure)

What can I expect for my son/daughter after the pull-through?

Many children with Hirschsprung's disease have a normal lifestyle after the pull-through procedure however, there is group of children that will have functional problems needing long-term follow-up.

One such problem is enterocolitis. One group of patients, around 30%, will have at any time after the pull-through, acute or chronic symptoms of intestinal obstruction (constipation like), abdominal distention, smelly loose stools (diarrhea like), vomiting and fever. These symptoms could be mild to severe and should be treated immediately, or better yet should be prevented.

Please read below about colonic irrigations.

Other groups of patients may suffer fecal incontinence after the pull-through; most of them are diagnosed at 3 to 4 years of age when mothers are aware that their children cannot use normal underwear due to fecal soiling and cannot achieve bowel control.

Finally, residual constipation is a problem that other group of patients may suffer after the pull-through.

What are colonic irrigations? Why should I know how to make it?

The anal canal is the terminal part of the large bowel located between the rectum and anus. It is a short zone containing the anatomical and physiological elements for fecal continence. The pectinate line is an important anatomical reference of the anal canal, which should be preserved during the anastomosis of the pull-through; the normoganglionic bowel is anastomosed (connected) to the anal canal, a few millimeters above this line.

The consequence of conserve undamaged this structure is that we leave a small zone with “obstructive-functional Hirschsprung's disease”, which is usually overcome for the remaining normoganglionic bowel. For unknown causes, some patients develop acute or chronic obstruction to the outflow causing stasis of stool, bacterial overgrowth and inflammation of the colon named “colitis” or “enterocolitis.” An inflamed bowel does not have normal propulsive movements worsening the obstruction.

In patients with “obstructive colitis,” colonic irrigations are really useful to remove the stool, to clean (wash out) the colon, to prevent stasis of stool helping to take control of bacterial overgrowth.

Contact Us

For more information about Hirschsprungsdisease or to request an appointment with the Colorectal Center for Children at Children's Hospital of Pittsburgh of UPMC, contact us at **412-692-7280** or by email at colorectalcenter@chp.edu.

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