Hirschsprung's Disease

Hirschsprung's disease is a rare congenital disorder affecting about 1 in 5000 newborns. These babies don't have normal nerve cells in the lower part of their large intestine. As a result, they don't have normal bowel movements. Surgery is usually successful in restoring normal function of the large intestine.

What is Hirschsprung's disease?

Hirschsprung's disease is a birth defect that occurs when nerve cells don't develop normally in part of the large intestine (colon). This results in obstruction (blockage) of the intestines. In most infants, the defect is limited to the lowest one third of the colon. In about 10%, the entire colon lacks nerve cells. Hirschsprung's disease is sometimes called "congenital aganglionic megacolon."

Babies with Hirschsprung's disease usually don't have normal bowel movements. The bowel material gets backed up and cannot be passed, causing the colon to enlarge. To avoid complications, it's important to recognize this condition as early as possible. Babies with Hirschsprung's disease need surgery to create a normally functioning large intestine. Although there is a risk of complications, most children do well after surgery.

What does it look like?

- Usually, Hirschsprung's disease is first suspected when there is a delay in passing the normal first meconium bowel movement after birth.
- Some infants pass the meconium stool but then have continued problems with constipation. Your baby may have a lot of problems passing bowel movements—they start to happen less and less often. In some infants, the bowel movements are small and pellet-like in appearance.
- Your baby may have feeding problems and poor growth.
- As material builds up in the large intestine, you may notice a bulge or swelling in your baby's abdomen. This can lead to infections and damage to your baby's intestines. Early diagnosis and treatment are essential to prevent these complications.

What causes Hirschsprung's disease?

- Hirschsprung's disease is caused by the absence of normal nerve cells in parts of the large intestine. As a result, the large intestine can't contract normally to move stool out of the body.
- Several abnormal genes have been linked to Hirschsprung's disease. If anyone in your family has had the disease, your child may be at a higher risk.

What are some possible complications of Hirschsprung's disease?

- The main complication is enlargement of the large intestine. This may lead to infections (enterocolitis) and endanger the blood supply to the large intestine.
- Even after surgery to create a normally functioning large intestine, your child may be at risk of infections and other complications. These risks are higher if a very long segment of the large intestine was involved (lacked nerve cells) and had to be removed at surgery.

What puts your child at risk for Hirschsprung's disease?

- It is four times more common in boys than in girls.
- It is more common in babies with certain genetic diseases, especially Down's syndrome.
- It may run in some families.

Can Hirschsprung's disease be prevented?

There is no way to prevent Hirschsprung's disease.

How is Hirschsprung's disease treated?

- First, tests may be performed to determine whether Hirschsprung's disease is present:
  - Barium enema: x-rays of the intestines, performed using a material called barium that shows abnormalities.
  - Manometry: performed to measure pressure changes in the rectum (the very bottom part of the large intestine).
  - Biopsy: A sample of the intestine is examined for the absence of nerve cells. This is the most certain way to make the diagnosis.
- Babies with Hirschsprung's disease may be quite ill. A doctor specializing in diseases of the stomach and intestines (gastroenterologist) will probably direct your baby's care.
- *Surgery* is needed for all infants with Hirschsprung's disease. The operation is planned to make the most use of your baby's normal intestinal tissue. If more of your

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baby's intestine lacks normal nerve cells, the surgery will be more extensive.

- The surgeon may decide to delay part of the surgery and wait to perform it until your child is a little older. In this situation, a relatively minor operation called a *colostomy* is performed. An opening is made in the wall of the abdomen, allowing bowel material to drain out. You'll receive the training you need to take care of your baby's colostomy.
- The good news is that surgery is usually highly successful in creating a normally functioning large intestine. With modern surgical techniques, most infants recover quickly.

- Your baby will need regular medical follow-up visits to make sure the intestines are working properly. If the surgery was very extensive, various complications may develop in the future. Most of these complications are manageable.

When should I call your office?

Call our office or go to the emergency department if any of the following occurs:

- Bulging (distended) abdomen; belly looks swollen, is sticking out.
- Fever.
- Abdominal pain.
- Frequent diarrhea.

Please type your custom instructions and/or office contact information here.

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