

Hirschsprung's Disease

What is it?

Hirschsprung's disease (pronounced HIRSH-sproongz) is a condition of the **large intestine (colon)**. It occurs when nerve cells are missing from the large intestine (colon). Normally, the intestines are lined with nerve cells that help move waste by wave-like muscle contractions. Babies with Hirschsprung's disease have a portion of the large intestine where the nerve cells are missing. Because the nerve cells are missing, waste cannot be moved through the intestines and a blockage occurs. This makes it difficult to have a bowel movement.

Signs and symptoms seen shortly after birth may include:

- Swollen belly
- Vomiting, including vomiting that is green or brown
- Constipation or gas
- Diarrhea

How common is it?

About 1 out of every 5,000 babies is born with Hirschsprung's disease each year in the United States.

What causes it?

Currently, the exact reason children with Hirschsprung's disease are missing nerve cells is not known. Hirschsprung's disease can be **familial**, meaning that multiple family members may be born with it. Hirschsprung's disease also can be associated with other birth defects, including genetic conditions

How is it diagnosed?

There may be many steps to getting a correct diagnosis of Hirschsprung's disease. Signs and symptoms appear shortly after birth, but sometimes they might not appear until later in life. Your child's doctor may do multiple tests, including x-rays and biopsy, to confirm the diagnosis. When a biopsy is done, a sample of colon tissue is tested to check for nerve cells.

How is it treated?

Hirschsprung's disease can be corrected by surgically removing the portion of the colon that is missing nerve cells. Your child's doctor(s) will discuss appropriate treatment options with you.



For more information:

Mayo Clinic

<https://www.mayoclinic.org/diseases-conditions/hirschsprung-s-disease/symptoms-causes/syc-20351556>

National Institute of Diabetes and Digestive and Kidney Diseases

<https://www.niddk.nih.gov/health-information/digestive->