



PERSPECTIVE

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Pathophysiology of Hirschsprung's Disease and its Diagnosis

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Description

Hirschsprung's Disease (HD or HSCR) is a birth defect in which nerves in parts of the intestine are missing. The most prominent symptom is constipation. Other symptoms may include vomiting, abdominal pain, diarrhea and slow growth. Symptoms usually appear in the first two months of life. Complications may include enterocolitis, megacolon, bowel obstruction, and bowel perforation. The disorder can occur on its own or in association with other genetic disorders such as Down syndrome or Waardenburg syndrome. About half of isolated cases are associated with a specific genetic mutation, and about 20% run in families. Some of them occur in an autosomal dominant manner. The cause of the remaining cases is unclear. If otherwise normal parents have one child with the condition, the next child has a 4% risk of being affected. This condition is divided into two main types, short segment and long segment, depending on how much of the intestine is affected. Rarely, the small intestine can also be affected. Diagnosis is based on symptoms and confirmed by biopsy. Treatment is generally surgery to remove the affected part of the intestine. The surgical procedure most commonly performed is known as a "stretch". Occasionally, a bowel transplant may be recommended. Hirschsprung's disease occurs in about one in 5,000 newborns. Men are more often affected than women. The condition is believed to have been first described in 1691 by the Dutch anatomist Frederik Ruysch and is named after the Danish physician Harald Hirschsprung after his description in 1888.

Pathophysiology

During normal prenatal development, cells from the neural crest migrate into the large intestine (colon) and form networks of nerves called the myenteric plexus (Auerbach's plexus) (between the smooth mus-

cle layers of the wall of the gastrointestinal tract) and the submucosal plexus (Meissner's plexus). In the submucosa of the wall of the gastrointestinal tract. In Hirschsprung's disease, migration is incomplete and part of the colon is missing these nerve cells that regulate colon function. The affected segment of the colon cannot relax and pass stool through the colon, creating an obstruction. The most accepted theory for the cause of Hirschsprung's is a defect in the cranio-caudal migration of neural crest-derived neuroblasts that occurs during the first 12 weeks of pregnancy. Defects in the differentiation of neuroblasts into ganglion cells and accelerated destruction of ganglion cells in the gut may also contribute to the disorder. This lack of ganglion cells in the myenteric and submucosal plexus is well documented in Hirschsprung's disease. In Hirschsprung's disease, the segment lacking neurons (aganglionic) narrows, causing the normal proximal part of the intestine to distend with feces. This narrowing of the distal colon and failure to relax in the aganglionic segment is thought to be due to a lack of neurons containing nitric oxide synthase. The most cited feature is the absence of ganglion cells especially in men, 75% have none at the end of the colon (rectosigmoid) and 8% lack ganglion cells throughout the colon. The enlarged part of the intestine is located proximally, while the narrowed, aganglionic part is located distally, closer to the end of the intestine. The absence of ganglion cells results in persistent overstimulation of the nerves in the affected area, leading to contraction.

Diagnosis

Definitive diagnosis is established by aspiration biopsy of the distal narrowed segment. Histological examination of the tissue would show a lack of ganglion nerve cells. Diagnostic techniques include anorectal manometry, barium enema, and rectal bi-

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opsy. Aspiration rectal biopsy is considered the current international gold standard in the diagnosis of Hirschsprung's disease. Radiological findings can also help with the diagnosis. Cineanography (fluoroscopy of a contrast agent passing through the anorectal region) helps in determining the level of the affected intestines.