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HIRSCHSPRUNG'S DISEASE IN CHILDREN.

Abstract: *This article provides information about Hirschsprung's disease in children and its origin, symptoms, treatment methods, diagnosis, etiology*

Key words: *Hirschsprung's disease*, agangliosis, *treatment, Sprinzen-Goldberg* syndrome, *Meissner and Auerbach, Genetic testing.*

Hirschsprung's disease is a congenital anomaly of innervation of the lower intestine (usually limited to the colon), which leads to partial or complete functional obstruction. Signs include constipation and bloating. The diagnosis is made on the basis of irrigation and rectal biopsy. Thanks to anal manometry, it is possible to determine the severity and assess the insufficiency of relaxation of the internal anal sphincter. The treatment is surgical.

Hirschsprung's disease is caused by the congenital absence of vegetative plexuses of Meissner and Auerbach (agangliosis) in the intestinal wall. The estimated frequency is 1 in 5,000 live births. The disease is usually limited to the distal part of the colon (in 75% of cases), but it can also involve the entire colon (in 5% of cases) or even the entire large and small intestine; areas in which there is no innervation are always adjacent. If the entire colon is not involved, men are more likely to get sick (male ratio:women = 4:1); otherwise, there are no gender differences.

The cause of agangliosis is considered to be insufficient migration of neuroblasts from the neural crest. There is a significant genetic component to this disease and at least 12 different genetic mutations associated with Hirschsprung's

disease. The probability of this disease among family members increases with an increase in the length of the involved part of the intestine: from 3 to 8% in cases where the distal colon is involved, and up to 20% when the entire large intestine is affected. Approximately 20-25% of patients with Hirschsprung's disease have another congenital malformation, and approximately 15% have a genetic anomaly (Down syndrome is the most common). About 20% of patients with congenital central hypoventilation syndrome also have Hirschsprung's disease; this combination is called Haddad syndrome. About 20% of patients with intestinal neuronal dysplasia (IND) have Hirschsprung's disease. Other diseases associated with Hirschsprung's disease include Waardenburg syndrome, Bardet-Bidl syndrome, Sprinzen-Goldberg syndrome, and cartilage and hair hypoplasia. Peristalsis in the affected segment is absent or pathological, which leads to continuous spasm of smooth muscles, partial or complete obstruction with accumulation of intestinal contents, massive dilatation of the normally innervated parts of the intestine above. It is almost impossible to miss pathological changes.

Symptoms and signs of Hirschsprung's disease:

Symptoms often appear in early childhood, although some do not develop the disease until school or even adulthood. Normally, 98% of newborns have meconium excretion from the body during the first 24 hours of life. In approximately 50-90% of newborns with Hirschsprung's disease, meconium is not excreted in the first 48 hours of life. Infants have constipation, bloating and, finally, vomiting – as well as with other forms of peripheral intestinal obstruction. Sometimes infants with agangliosis of a very short segment develop only mild or intermittent constipation, often interspersed with diarrhea, which leads to a delay in diagnosis. In older infants and children, symptoms and signs may include anorexia, constipation, lack of physiological urge to defecate and, with finger rectal examination, an empty rectum with a stool palpable higher in the colon, and rapid passage of the stool after removal of the examining finger (a sign of an explosion).

Infants may also be stunted. Less often, infants may have Hirschsprung's enterocolitis.

Diagnosis of Hirschsprung's disease:

Initially, as a rule, a contrast enema with barium and / or rectal aspiration biopsy is performed. Irrigation with barium can show a transition in diameter between an enlarged, normally innervated colon located proximally and a narrowed distal segment (not having normal innervation). Irrigation with barium should be done without prior preparation, which can lead to the expansion of the abnormal segment, which makes the study undiagnostic. Since the characteristic signs in the neonatal period may not be observed, it is worth conducting an X-ray examination within 24 hours after bowel emptying: if the colon is still filled with barium, Hirschsprung's disease can be suspected. Rectal aspiration biopsy may indicate the absence of ganglion cells. Acetylcholinesterase staining can be performed to isolate enlarged nerve trunks. In some centers, it is possible to perform rectal manometry, which can reveal motor disorders characteristic of abnormal innervation. To make a final diagnosis, it is necessary to conduct a deep biopsy of the rectum or colon to assess the degree of the disease and the subsequent volume of surgical treatment.

Genetic testing is not routinely used, but can be carried out in the presence of manifestations of a genetic syndrome.

Treatment of Hirschsprung's disease:

Reconstructive surgery

The treatment of Hirschsprung's disease is a restorative operation to bring the normally innervated intestine to the anus with the preservation of the anal sphincter. In newborns, this was usually a two-stage procedure starting with a colostomy proximal to the aganglionic segment for decompression of the colon. Then the newborn will grow up to the 2nd stage of the procedure, in which the entire aganglionic part of the colon is resected and the reduction operation is

performed. However, many clinical centers are currently conducting a one-stage procedure in the neonatal period for the lesion of a short segment. The results using laparoscopic techniques are similar to those obtained with open access, and are associated with shorter hospitalization, earlier start of feeding, and less pronounced pain sensations.

After the final recovery, the prognosis is good, despite the fact that some children have chronic motor disorders with constipation, obstructive problems, or both conditions.

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