Hirschsprung Disease

Hirschsprung disease is a congenital disorder of the gut caused by failure of neural crest cells to migrate fully. This causes a segment of the distal intestine, including the rectum, to lack both the Meisnner plexus in the submucosa and Auerbach myenteric plexus, which are the major nerve supplies to the GI tract and play an important role in controlling GI tract motility. Loss of these innervations leads to loss of coordinated peristalsis and the development of a functional obstruction leading to chronic constipation and failure to pass meconium. The obstruction of the distal colon results in dilation proximal to the affected segment and with time, the proximal colon can become severely distended leading to congenital megacolon. 10% of all cases of Hirschsprung disease occur in children with Down syndrome. Hirschsprung Disease is also associated with mutations in the RET gene.



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Failure of Neural Crest Cell Migration

Migrating Flying Neurons with Crests being Interrupted

Hirschsprung disease results when normal migration of neural crest cells is arrested prematurely around the cecum to the rectum. This causes a segment of the distal intestine to lack both the Meissner plexus in the submucosa and Auerbach myenteric plexus.

Involves Rectum

Rectum-rectangle

The rectum is always affected in Hirschsprung disease but the involvement of additional segments varies widely. Most cases are limited to the rectum and can involve the sigmoid colon but severe cases can involve the entire length of the colon.

Lack of Auerbach and Meissner Plexus

Hourglass and Maze

Hirschsprung disease results when normal migration of neural crest cells is arrested prematurely around the cecum to the rectum. This causes a segment of the distal intestine to lack both the Meissner plexus in the submucosa and Auerbach myenteric plexus. These neurons are the major nerve supply to the GI tract and play an important role in controlling GI tract motility.

Chronic Constipation

Corked Con-toilet

Because the Meissner and Auerbach plexus play an important role in controlling GI tract motility, Hirschrpung disease is characterized by uncoordinated peristalsis and functional obstruction of the gut causing chronic constipation.

Failure to Pass Meconium

M-cone

Infants with Hirschsprung disease can present with failure to pass meconium due to uncoordinated peristalsis and functional obstruction of the gut.

Congenital Megacolon

Mega-colon

Functional obstruction of the distal colon results in dilation proximal to the affected segment. With time, the proximal colon may become severely distended leading to megacolon.

Increased Risk with Down Syndrome

Down Syndrome Child

10% of all cases of Hirschsprung disease occur in children with Down syndrome.

RET Mutations

Roulette Mutant

Mutations in RET are most notable for causing multiple endocrine neoplasia type 2 (MEN2). However, mutations in this gene are also notable for their role in Hirschsprung Disease. Epidemiological studies vary, but the co-occurrence of Hirschsprung Disease and RET mutations has been reported to be as high as 70%.

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