

Hirschsprung Disease

Congenital Aganglionic Megacolon
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From birth: Absence of cluster of nerves Enlarged colon

- Autosomal dominant disorder due to mutation in RET oncogene usually associated with MEN 2 and 3.
- RET is important for the neural crest cells to migrate and differentiate - so lack in that causes absence of ^{neural} migration to the distal colon and rectum.

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Absence of submucosa (Meissner plexus)
& Myenteric plexus (Auerbach plexus)

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No dilatation on contraction

C/F

- No Peristalsis (Constipation, distension, vomiting).
- No Meconium - 2 days within
- ~~No~~ Dilatation of the sigmoid colon
↓ Rupture & Peritonitis
- RET oncogene associated with Down Syndrome

Investigation

Rectal Suction Biopsy (Full Thickness Biopsy)

↳ Absence of plexus

- X-ray Abdomen with Barium Enema
Colonic dilatation.