**HIRSCHSPRUNG DISEASE**

- **Congenital defect**
- **Incidence**: 1:5000 live births
- **M:F = 4:1**
- **85% of the time limited to the rectosigmoid segment of the colon**

**PATHOPHYSIOLOGY**
Migration failure of neural crest cells to the distal colon
- Absence of ganglion cells in the submucosal and myenteric plexuses
- Absence of peristalsis in the aganglionic colon
- Functional obstruction

**CLINICAL PRESENTATION**

<table>
<thead>
<tr>
<th>NEWBORN</th>
<th>OLDER CHILD</th>
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<tbody>
<tr>
<td>• Failure to pass meconium in first 24 hours after birth</td>
<td>• Severe chronic constipation</td>
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<tr>
<td>• Bowel obstruction with bilious vomiting</td>
<td>• Failure to thrive, malnutrition and feeding problems</td>
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**TREATMENT**

- **Surgical “pull-through” procedure**: removal of the aganglionic segment and anastomosis between the normally innervated part of the intestine and the anus, preserving the anal sphincter

**Hirschsprung-associated enterocolitis**

- **Life-threatening complication**
- **Inflammation of the colon which causes bowel obstruction & sepsis**
- **Can occur either before or after the pullthrough surgery**

**Signs and symptoms**
- Abdominal distension
- Explosive diarrhea
- Vomiting
- Other: fever, rectal bleeding, shock
- X-ray: megacolon (severely dilated colon)

**Treatment**
- Broad spectrum antibiotics
- IV fluids
- Labs: CBC, CRP, blood culture
- Nasogastric decompression
- Rectal irrigations
- General surgery consultation

**INVESTIGATIONS**
- **TSH** (rule out hypothyroidism), electrolytes (rule out hypercalcemia, hypokalemia)
- Abdominal X-Ray → will show dilated loops of intestine
- Contrast enema → will show the “transition zone” between the narrow rectum and distal colon and the dilated proximal colon
  - Contraindicated if enterocolitis is suspected → risk of perforation
  - For infants > 12 months old → anal manometry can show absence of rectoanal inhibitory reflex

**DIAGNOSIS**
- **Rectal biopsy** → will show aganglionosis, hypertrophy of nerve trunks, and abnormal calretinin staining

**PHYSICAL EXAM**
- Abdominal distension
- Digital rectal exam:
  - Empty rectum
  - Explosive ejection of stool as the finger is removed ("squirt sign")

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