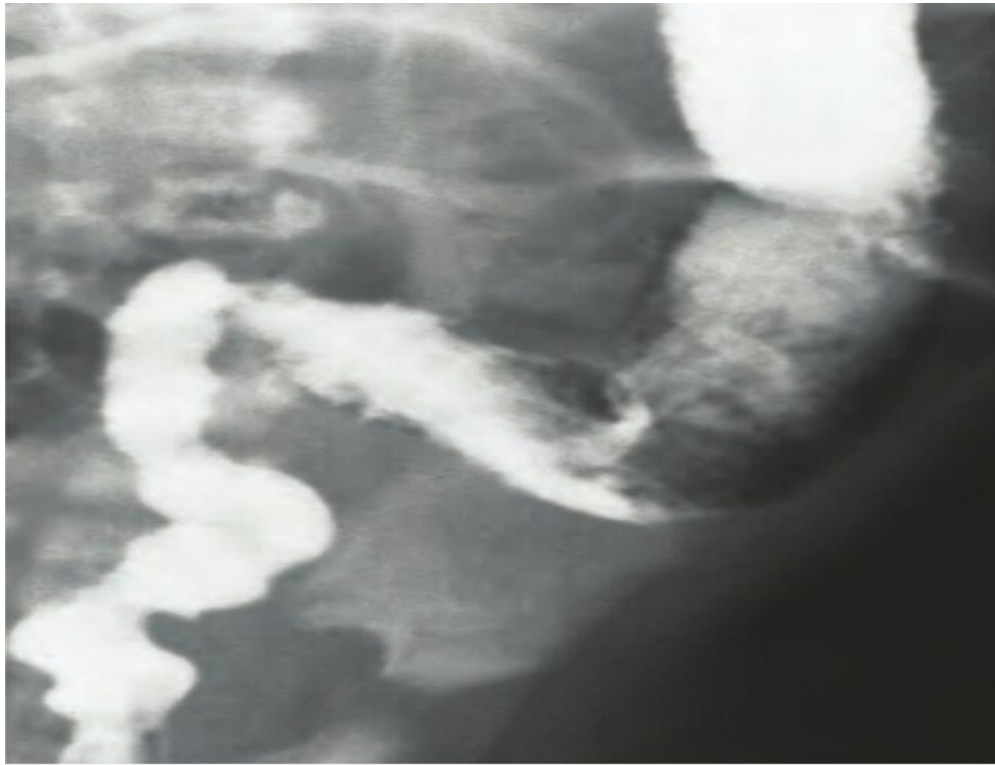


# Hirschsprung's disease



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)



# Incidence

- It occurs as Congenital and Familial.
- Also known as Congenital mega colon.
- Gene mutation also have identified on chromosome 10 ( involving the RET proto oncogene) and on chromosome 13 in some patients.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Incidence

- Some of these conditions occur in an autosomal dominant manner.
- The cause of certain remaining cases are unclear.
- If otherwise normal parents have one child with the condition, the next child has 4% of being affected.



# Pathophysiology

- Occurs in newborn due to absence of ganglion cells in the Auerbach's and Meissner's plexus.
- And there is hypertrophic nerves in the in the distal large bowel.
- The absence of ganglion is due to a failure of migration of vagal neural crest cells into the developing gut.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Pathophysiology

- The cranicaudal migration of neuroblast originating from the neural crest that occurs during first 12 weeks of gestation.
- Defects in the differentiation of the neuroblasts into ganglion cell destruction within the intestine may also contribute to this.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Pathophysiology

- 3 zones in a affected colon.
  1. Distal immobile spastic segment.  
(Aganglionic zone)
  2. Cone
  3. Normal ganglionic dilated and hypertrophied zone.



# Pathophysiology

- A transition zone exist between the dilated , proximal, normally innervated bowel and the narrow , distal aganglionic segment.



# Types

1. Ultra short segment HD – Terminal part of the rectum is affected.
2. Short segment HD – Rectum is affected. (80%)
3. Long segment HD- Rectum and part of the colon are affected.
4. Total colonic HD- 5%.





# Clinical features

- 80% occur in males
- Common in infants and children.
- 10% associated with Down's syndrome.
- 90% are symptomatic within 3 days of age.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Clinical features

- Common presentation of this condition,
  1. Failure to pass meconium.
  2. Toothpaste like stools when DRE.
- Enterocolitis is a potentially fatal condition.



# Clinical features

- This is typically in neonates with delayed passing meconium, abdominal distension and villous vomiting.
- But it may not be diagnosed until later in the childhood or even adult life, when it manifests as chronic constipation.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Investigations

1. Plain X ray – intestinal obstruction.
2. Biopsy from all zones.
3. Ba enema.
4. Anorectal Manometry.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Investigations

- Diagnosis requires an adequate rectal biopsy and an experienced pathologist.
- In contrast enema shows the narrow aganglionic segment, cone and dilated proximal bowel.
- Surgery aims to remove this aganglionic segment.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Diagnosis

- Definitive diagnosis require a suction biopsy of the distally narrowed segment.
- A histologic examination of the tissue would show lack of ganglionic nerve cells.
- Suction rectal biopsy is considered currently the gold standard .



# Diagnosis

- Radiological findings may also assist with diagnosis.
- Cineanography assists in determining the level of the affected intestines.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Treatment

1. Rectal washouts
2. Colostomy
3. Nutrition
4. Definitive surgery – pull through surgery when child reaches 10kg of body weight.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)



# Treatment

- Surgery aims to remove the aganglionic segment and pull through ganglionic bowel to the anus.
- Swenson, Duhamel, Soave and transanal procedures are there.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Treatment

- Surgery can be done in single stage or in stages after first establishing a proximal stoma in normally innervated bowel.
- Outcome is good in most cases.



# Complications

1. Colitis
2. Intestinal obstruction
3. Growth retardation
4. Constipation
5. Perforation
6. Peritonitis



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)

# Prognosis

- Following the surgery most patients achieve good bowel control but significant minority experience residual constipation and faecal incontinence
- someone also experienced enterocolitis.



**RISH**ACADEMY

educate yourself to empower yourself

[www.rishacademy.com](http://www.rishacademy.com)