



## Hirschprung's Disease in Neonates

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### Abstract

**Background:** *Hirschprung's Disease in Neonates (HD) is an important cause of intestinal obstruction. To evaluate incidence / clinical investigational profile / treatment modality in HD in neonates, the following study was done.*

**Method:** *The neonates who came to pediatric gastroenterology OPD of PGI MER, Chandigarh with clinical feature of HD (e.g. delayed passage of meconium at birth, abdominal distension, vomiting, enterocolitis with fever diarrhea etc.) were admitted from July 1993 to June 2003. History, clinical feature, investigation (USG whole abdomen, barium enema, rectal suction biopsy, CBC, electrolyte etc.) were noted. Surgery (i.e. colostomy followed by definitive surgery of abdominal pull through etc.), and nutritional care like parenteral nutrition etc was done.*

**Results:** *Out of 50 neonates of HD, male was 30, female was 20. Age of presentation was 5 – 15 days. Birth weight was 2.5 to 3.5 kg. (average 2.8 kg.).*

*Delayed passage of meconium was in 45 (90%) cases, abdominal distension in 35 (75%), bilious vomiting in 40 (80%), fever in 5 (10%), diarrhea in 3 (6%), perforation in 5 (10%) cases.*

*Site of aganglionic segment seen in rectum is 10 (20%), sigmoid colon in 30 (60%), descending colon in 5 (10%), splenic flexure in 3 (6%), 1 (2%) in ileum, total aganglionic colon in 1 (2%) cases. Forty (80%) cases had initial colostomy followed by definitive surgery (Boley's endorectal pull through). In 10 (20%) cases, definitive surgery was done in single phase.*

**Conclusion:** *HD is an important cause of neonatal intestinal obstruction. Early detection / management is very important.*

### Introduction

Hirschprung's Disease (HD) is a serious birth defect where nerve cells are completely missing from end of bowel. The area of bowel where enteric nerves are missing will stay contracted and bowel obstruction occurs.<sup>(1)</sup>

HD is an important differential diagnosis of neonatal intestinal obstruction.<sup>(2)</sup>

The presenting feature of HD depends on length of aganglionic segment and severity. HD rarely

presents in preterm babies. Clinical presentation may be delayed passage of meconium, neonatal bowel obstruction, neonatal bowel perforation, neonatal enterocolitis, constipation. The length of aganglionic segment are short (75%), long segment (15%), total aganglionosis (2 – 13%) (it is also known as zuebler – Wilson syndrome), with short segment (3 – 4 cm of internal sphincter only).

5% children with HD have bowel perforation. It may be colonic or ileal and accounts for 10% of neonatal bowel perforation.<sup>(3,4)</sup>

70 – 75% of HD can be diagnosed BY Barium / contrast enema. The diagnostic feature are transitional zone, absence of peristalsis in aganglionic segment, saw tooth appearances in aganglionic segment, retention of barium in colon after 24 hours. Clinical radiological feature. Is absent in 25% neonate.<sup>(5)</sup>

Seventy percent of HD has isolated lesion. Well documented associated anomalies are down syndrome (5 – 10% of HD), congenital deafness (9%), congenital Heart defects (8%), Mental retardation and seizure (6%), Neurocristopathy syndrome (e.g. MEN II a etc.), non-neurocristopathy syndrome (e.g. Smith-Lemli-Opitz syndrome etc).<sup>(6-8)</sup>

HD can cause severe complications and occasionally lead to serious bowel infection and treated early but HD is picked up soon after birth and treated with surgery as soon as possible.<sup>(9)</sup>

Signs of HD in neonates are swollen belly and tummyache, not feeding well, not gaining weight.<sup>(10)</sup>

High temperature of 38<sup>0</sup> C or more / watery foul smelling diarrhea could be a sign of bowel infection (enterocolitis) which may be serious and lead to sepsis. Digital rectal examination and x-ray can be done to show blockage and bulge in bowel.<sup>(11,12)</sup>

Diagnosis is confirmed by rectal biopsy which is done by inserting a small instrument into rectum to remove a tiny sample of affected bowel.<sup>(13)</sup>

## Method

The neonates who came to pediatric gastroenterology OPD of PGI MER, Chandigarh with clinical feature of HD (e.g. delayed passage of meconium at birth, abdominal distension, vomiting, enterocolitis with fever diarrhea etc.) were admitted from July 1993 to June 2003. History, clinical feature, investigation (USG whole abdomen, barium enema, rectal suction biopsy, CBC, electrolyte etc.) were noted. Surgery

(i.e. colostomy followed by definitive surgery of abdominal pull through etc.), and nutritional care like parenteral nutrition etc was done.

## Results

Out of 50 neonates of HD, male was 30, female was 20. Age of presentation was 5 – 15 days. Birth weight was 2.5 to 3.5 kg. (average 2.8 kg.). Delayed passage of meconium was in 45 (90%) cases, abdominal distension in 35 (75%), bilious vomiting in 40 (80%), fever in 5 (10%), diarrhea in 3 (6%), perforation in 5 (10%) cases.

Site of aganglionic segment seen in rectum is 10 (20%), sigmoid colon in 30 (60%), descending colon in 5 (10%), splenic flexure in 3 (6%), 1 (2%) in ileum, total aganglionic colon in 1 (2%) cases. Forty (80%) cases had initial colostomy followed by definitive surgery (Boley's endorectal pull through). In 10 (20%) cases, definitive surgery was done in single phase.

## Discussion

Various genes are involved in HD. Some genes are associated with down syndrome but in most cases it is not.<sup>(14)</sup>

HD enterocolitis may occur either pre-operatively (upto 50% incidence) or post-operatively (22% incidence). Fever, vomiting, abdominal distension occurs. Fecal stasis from aganglionic segment leads to bacterial over growth of gut followed by inflammation of mucosa with high blood count and fever.

Bowel perforation, septicemia, death can occur. Rectal irrigation, naso gastric decompression, IV fluid, antibiotic coverage for gram negative anaerobic organisms are done. Pediatric surgeon should be consulted.<sup>(15)</sup>

Enterocolitis may be recurrent, difficult to treat in post operative scenario if pre operative enterocolitis is present. It may require reoperation. Even diverting colostomy may be required.<sup>(16)</sup>

Most children with HD will have "Pull-Through" operation. Here affected segment of bowel is removed and remaining healthy section of bowel are connected together.

If child is not well enough to withstand this surgery, then this may be done in two stages. A few days after birth, surgeon will divert bowel through temporary opening (stoma) made in tummy. This procedure is called colostomy. Colostomy can be done by open surgery or by laparoscopy.<sup>(17)</sup>

Risks of surgery in HD are small. These include bleeding during or after operation, bowel becoming infected (enterocolitis), bowel contents going outside causing peritonitis which is a serious infection, bowel being narrowed or bowel requires repeat surgery.<sup>(18)</sup>

Most children will have normal functioning bowel after surgery, some may experience constipation and require high fiber diet and laxatives.<sup>(19)</sup>

A small number of children may experience bowel incontinence. This may last up to teen age and very much distressing.<sup>(20)</sup>

The parents may be given advice about soiling in children.<sup>(21)</sup>

Colostomy in neonates under local anesthesia is feasible, safe, effective. Lukong CS et al performed colostomy in neonates in a series of 38 neonates with anorectal malformations (97%) and colonic atresia (3%). Sigmoid colostomy, descending colon colostomy, transverse colon colostomy was done in 76%, 18%, 6% respectively. Early complication was skin excoriation, superficial site infection, bowel evisceration.

Late complications were stomal stenosis, colostomy diarrhea, hernia. (22%)

Colostomy stoma care is very much important in HD surgery. Healthy stoma is red and moist. The skin around stoma should be free from rash and broken down area. The stoma should not be painful as there is no nerve endings in stoma.

Pouch should be free from emptied into toilet when it is half filled with gas or stool. If it gets too full it will leak and not last long.

It should be emptied before nap time, bed time.

The pouch should be changed if it is loose or start leaking (leakage will damage skin).

The skin around stoma should be cleaned with water on soft paper towels, wipes, oil, powder, ointment or lotion should not be used.<sup>(23)</sup>

After HD surgery, stenosis can be persistent in anastomotic site. Anal dilatation can be recommended by surgeon. It may be continued at home.

Catheter dilator, K-Y Lubricant, clean diaper, wipes may be needed.

Small amount of bleeding may occur. It may be normal gradual increment of dilator size is done as directed by surgeon. Increasing size of dilator may be stopped when maximum size is achieved.<sup>(24)</sup>

In our series of 50 patients of HD, 40 (80%) needed initial colostomy followed by definitive surgery later on. 10 (20%) cases received definitive surgery on first occasion.

### Conclusion

HD is an important cause of neonatal intestinal obstruction. Early detection / management is very important.

### Conflict of interest – Nil

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