Hirschsprung’s Disease in Africa in the 21st Century

1. Definition and background
Hirschsprung’s disease (HD) is a form of functional intestinal obstruction caused by absence of ganglion cells in the myenteric and submucosal plexuses of the intestine, which results in absent peristalsis in the affected bowel. It is also referred to as congenital megacolon or congenital colonic aganglionosis, and occurs in 1 in 5,000-7,200 newborns. (1-3) In Africa only 20-40% present as neonates, compared to more than 90% in developed countries. (4-8) The male: female ratio is 4:1, but this becomes equal in long segment disease and familial cases.
Congenital megacolon was first described by Ruysch in 1691, and then more widely reported by Danish Paediatrician Harald Hirschsprung in 1886. The pathophysiology of aganglionosis was not determined until the middle of the 20th century following which Swenson recommended rectosigmoidectomy as the optimal treatment in 1948. (9-10) Initially this operation was performed without colostomy, but the debilitated and malnourished state in which many children presented caused most surgeons to adopt a multi-staged approach. Recent advances and refinements in surgical technique have resulted in a shift towards one-stage and minimal access procedures for the treatment of this disease. (1-2) In Africa, ignorance and poverty on the part of the parents, late presentation with attendant complications, limited access to trained paediatric surgeons and limitation of facilities for prompt diagnosis characterize management of this disease. Hence, multiple stages of management still predominate in sub Saharan Africa, (7)(8) (11)

2. Embryology and aetiology

Neural crest cells originate in the vagal neural crest and then migrate craniocaudally into the embryonic intestine reaching the rectum at the 12th week. Auerbach’s myenteric plexus layer is formed first and Meissner’s submucosal plexus develops later, with cells maturing after arriving at their destination. (2)

Abnormalities in the microenvironment result in the neural crest cells failing to reach the distal bowel. There are differences in extracellular matrix proteins (fibronectin, laminin), abnormal cell-cell interactions (absent neural cell adhesion molecule) and absence of neurotrophic factors in aganglionic bowel when compared with normal bowel. (12-13)

Other investigators suggest that neural crest cells originate in both vagal and sacral sites and migrate toward the middle of the intestine, raising the possibility that the neural crest cells get to their destination, but then fail to survive, proliferate, or differentiate. (14) Additionally, the observation that the smooth muscle cells of aganglionic colon are electrically inactive points to a myogenic component in the development of HD. (15) Abnormalities in the pacemaker Interstitial Cells of Cajal, have also been postulated as an important contributing factor. (16)

3. Genetic Abnormalities

Sporadic occurrence accounts for 80% to 90% of cases of HD. Variable expressivity and incomplete sex-dependent penetrance are observed, suggestive of a more complex pattern of inheritance and the involvement of several genes.

A positive family history occurs in approximately 10% of children, especially those with longer segment disease. Children with Down syndrome and other genetic abnormalities also have a higher incidence of HD. HD has been associated with the RET proto-oncogene the endothelin family of genes, SOX-10 gene and SIP1. It is unclear exactly how these genetic abnormalities result in the phenotype of HD. Development of the disease is a multi-genic phenomenon that can occur at any number of stages during the normal process of neural crest cell migration, differentiation, and survival.

Animal models demonstrate that some mutations may produce early maturation or differentiation of neural crest cells, and mutations in the RET proto-oncogene likely act by depriving the migrating neural crest cells of an adequately supportive microenvironment. (17) The genetic mutations associated with HD can best be understood by examining how they relate to the family of neurocristopathies, many of which have similar genetic patterns (Table 1).
TABLE 1  
Syndromes and Genetic Abnormalities Associated with Hirschsprung's Disease.

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>Identified Genetic Basis</th>
</tr>
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<tbody>
<tr>
<td>Down syndrome</td>
<td>Trisomy 21</td>
</tr>
<tr>
<td>Neurocristopathy syndromes</td>
<td>Endothelin and SOX-10</td>
</tr>
<tr>
<td>Waardenberg-Shah syndrome</td>
<td></td>
</tr>
<tr>
<td>Yemenite deaf-blind-hypopigmentation</td>
<td></td>
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<tr>
<td>Piebaldism</td>
<td></td>
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<tr>
<td>Other hypopigmentation syndromes</td>
<td></td>
</tr>
<tr>
<td>Goldberg-Shprintzen syndrome</td>
<td>Possibly SIP1</td>
</tr>
<tr>
<td>Multiple Endocrine Neoplasia 2</td>
<td>RET</td>
</tr>
<tr>
<td>Central hypoventilation syndrome (Ondine's curse)</td>
<td>Unknown</td>
</tr>
</tbody>
</table>

4. Pathology

Normal intestinal motility is primarily under the control of intrinsic neurons that control both contraction and relaxation of smooth muscle, with relaxation predominating. Extrinsic control is mainly through the cholinergic and adrenergic fibers (2) In HD, ganglion cells are absent, leading to a marked increase in smooth muscle tone and an imbalance of smooth muscle contractility, uncoordinated peristalsis, and a functional obstruction.

The gross appearance of a bowel segment affected by HD is an aganglionic distal spastic and narrow segment with a proximal hypertrophic and dilated bowel separated by a 5-10cm transition zone.

Histologically, the absence of ganglion cells in the distal intestine is the hallmark of the disease. Ganglion cells are absent in both the submucosal (Meissner's) plexus and the myenteric (Auerbach's) plexus. There is usually a marked hypertrophy of nerve fibers that extend into the submucosa that may be seen on routine hematoxylin-eosin stained slides but are more easily seen using an acetylcholinesterase stain (Figure 1 and 2) Cases with long segment or total colon HD may not have nerve hypertrophy on rectal biopsy.

![Figure 1](image.png)

**Figure 1:** Normal ganglion cells in myenteric plexus
Figure 2: Staining with acetylcholinesterase shows abnormal pattern of hypertrophic nerves in HD (source BHC)

Aganglionosis is always present in the rectum and progresses proximally and continuously for a varying distance. Exceptions have been documented (skip lesions (18)), though these cases are so rare that the finding of ganglion cells proximal to an aganglionic segment indicates that the biopsy was taken within the transition zone. The transition zone has a 5-10cm progressive decrease of ganglion cells until the aganglionosis level is reached. The transition zone may not be symmetric circumferentially, which has implications in deciding how much bowel to remove.

5. Classification

Hirschsprung’s disease is classified based on the length of involved bowel and location of transition zone.

<table>
<thead>
<tr>
<th>Length of bowel involved</th>
<th>Proportion of HD cases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Rectosigmoid</td>
<td>70-80%</td>
</tr>
<tr>
<td>Long segment (above sigmoid)</td>
<td>10- 25%</td>
</tr>
<tr>
<td>Total colonic</td>
<td>3- 15%</td>
</tr>
<tr>
<td>Total intestinal</td>
<td>0.4- 4%</td>
</tr>
<tr>
<td>Ultrashort segment</td>
<td>&lt;1%</td>
</tr>
</tbody>
</table>

6. Clinical presentations

There are three ways that HD characteristically presents: neonatal bowel obstruction, chronic constipation, and enterocolitis. Most patients in the African series presented with intestinal obstruction while about 30% presented with constipation, 11% with enterocolitis, and 2% with intestinal perforation. (5-7) (11)

In developed countries, the age at which HD is diagnosed has progressively decreased over the past century so that 90% of patients with HD are now diagnosed in the newborn period. In Africa less than 50% of HD cases present as neonates, and those usually present with complete intestinal obstruction or cecal perforation and suffer high morbidity and mortality. (7) (8) (11) (19) (20) Presentation in adulthood has also been reported. (21), (22)

6.1. Neonatal bowel obstruction
There is a history of delayed passage of meconium in about 80% of newborns with HD. A study of normal African newborns found that 75% passed meconium within 24 hours of birth, 92% within 48 hours and 98% within 72 hours. (23) A delay of more than 48 hours in passage of meconium should raise concern about HD.

6.2. Chronic constipation
Many children in the African settings with delayed passage of meconium or infrequent passage of stool are managed with traditional enemas. (Figure 3 (24))

Children with HD may present later with chronic constipation, failure to thrive, gross abdominal distention and dependence on enemas without significant encopresis. (Figures 4 and 5) The impacted sigmoid megacolon may also undergo volvulus (Fig 6).
Figures 4 and 5: Gross abdominal distension and failure to thrive in children with chronic constipation and intestinal obstruction due to Hirschsprung’s disease (Courtesy Prof. E. A. Ameh, ABUTH, Zaria).

Figure 6a. Dilated rectosigmoid segment loaded with faeces above the transition zone, leading to a sigmoid volvulus (arrow shows point of volvulus).

Figure 6b: Hugely dilated sigmoid above transition zone has become gangrenous. Note the viable pink bowel adjacent to gangrenous sigmoid. (source, author LOA)
6.3. Enterocolitis

Hirschsprung’s Enterocolitis (HEC) is characterized by fever, abdominal distention, and explosive or foul-smelling diarrhea, and may lead to life threatening septic shock. Approximately 10% of children with HD have diarrhea as part of the presentation, and the diagnosis may therefore be missed. (8) Suspicion of HD should be raised if a history of failure to pass meconium and intermittent obstructive episodes is elicited.

6.4. Associated Anomalies and Syndromes

The incidence of associated congenital anomalies is approximately 20% involving the neurological, cardiovascular, urological, or gastrointestinal system. (25) In addition, HD may be part of a large number of recognized syndromes, some of which have an identifiable chromosomal or genetic basis (Table 1). HD should therefore be suspected in any child with constipation or neonatal intestinal obstruction who is known to have one of these syndromes. In addition, a diagnosis of HD should alert the clinician to the increased possibility of these associated anomalies.

7. Diagnosis

The differential diagnosis of HD in infancy includes other causes of neonatal intestinal obstruction. (Table 2) HD may be suspected in older children presenting with functional constipation who do not respond to usual treatments.

Table: 2 Differential diagnoses for Hirschsprung’s disease according to age of presentation

<table>
<thead>
<tr>
<th>Neonates</th>
<th>Older children</th>
</tr>
</thead>
<tbody>
<tr>
<td>Distal intestinal obstruction:</td>
<td>Functional constipation</td>
</tr>
<tr>
<td>Ileal atresia</td>
<td>Fecal impaction</td>
</tr>
<tr>
<td>Meconium ileus</td>
<td>Abdominal tumor</td>
</tr>
<tr>
<td>Meconium plug syndrome</td>
<td>Abdominal Tuberculosis and lymphoma</td>
</tr>
<tr>
<td>Small left colon syndrome</td>
<td>Metabolic abnormalities</td>
</tr>
<tr>
<td>Prematurity</td>
<td>Pseudo-obstruction</td>
</tr>
<tr>
<td>Sepsis and electrolyte imbalance</td>
<td></td>
</tr>
<tr>
<td>Cretinism and myxedema</td>
<td></td>
</tr>
<tr>
<td>Intestinal neuronal dysplasia</td>
<td></td>
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</tbody>
</table>

The appropriate diagnostic approach varies, depending on the age of the patient and the presenting clinical picture. After a careful history eliciting delayed passage of meconium, chronic constipation and or repeated diarrhea and adequate physical examination including a digital rectal examination which gives a tight rectum with absent fecal content, the diagnostic steps should include radiographic studies, and rectal biopsy.

7.1. Plain radiographs usually show dilated bowel loops with characteristic flank fullness (Figure 7a). The colon may show stippled shadows which are evidence of large amounts of fecal stasis proximal to the obstruction. Pneumoperitoneum may be evident in those who have developed intestinal perforation. In newborns, a prone lateral x-ray (Figure 7b) can demonstrate that the rectum is narrower than the proximal bowel in HD.
Figures 7a and 7b: Grossly dilated large bowel loop on anteroposterior view. The prone lateral x-ray shows a more dilated sigmoid compared with the rectum (arrow): a rectosigmoid index of less than 1 is abnormal and suggests HD. (source BHC)

7.2. Contrast enema
In older children with HD, a water soluble contrast enema will demonstrate a funnel shaped transition zone between the normal and aganglionic bowel. These features are better elicited on the lateral or oblique films since superimposed loops of sigmoid colon make the interpretation difficult on the anteroposterior film. (Figures 8a and 8b) The transition zone may be absent in 25% of neonates and in older children with a very short aganglionic segment. (26) A rectosigmoid index (the ratio of rectal diameter/sigmoid diameter) less than 1.0 and retention of barium on a 24-hour post evacuation film are other findings supporting the diagnosis of HD.
The findings in total colonic HD may include a normal barium enema, a short colon of normal caliber, a microcolon, or a transition zone in the ileum. There may also be an easy, extensive reflux far back into small bowel, a pseudo-transition zone in the colon, and intraluminal small bowel calcification.

Figure 8a and 8b: Contrast enema demonstrating the funnel shaped transition zone (arrow) (source LOA)

Figure 9: CT scan of abdomen in a child presenting with an abdominal ‘tumor’ (source BHC)
Although a computerised tomographic scan is not generally indicated in the diagnosis of HD, this 20 month old infant with an abdominal ‘tumor’ has faecal loading in the grossly dilated rectosigmoid segment and was found to have HD. (Fig 9)

7.3. Anorectal Manometry may show absence of the rectoanal inhibitory reflex. It is not widely available in Africa and not used frequently in Canada because it is unreliable and not diagnostic.

7.4. Rectal Biopsy
The gold standard for definitive diagnosis of HD is rectal biopsy, looking for the absence of ganglion cells and the finding of hypertrophied nerve trunks. The biopsy is taken 2-3 cm above the dentate line on the posterior wall of the rectum. Going too distally may result in a false-positive diagnosis of HD because ganglion cells are normally absent in the anal canal.

The most common technique used in Africa is full thickness rectal wall biopsy, which requires close collaboration with a good pathologist. Disadvantages of full thickness biopsy include the potential for bleeding and scarring and the need for general anesthesia. Evaluation of biopsy specimens may be enhanced by staining of the tissue for acetylcholinesterase and immunohistochemistry with calretinin. (27) Histochemistry is also useful in differentiating between HD and intestinal neuronal dysplasia.

A suction rectal biopsy can be used to obtain tissue for histologic examination. Rectal mucosa and submucosa are sucked into the suction device, and a self-contained cylindrical knife cuts off the tissue (Figure 10). The distinct advantage of the suction biopsy is that it can be easily performed at the bedside without general anaesthesia and can be repeated several times. In experienced centres, the accuracy is 99.7%. Interpretation of the specimen is technically more demanding for pathologists, and some will require a full thickness biopsy to confirm the diagnosis of HD. This device is expensive and not readily available in Africa.

![Figure 10: rbiz™ rectal mucosa suction device (Aus Systems PTY Ltd.)](image)

8. Treatment

Once the diagnosis of HD has been established, the goals of initial management are 2-fold:

   i) Rehydration and correction of electrolyte derangements.
   ii) Nasogastric decompression in patients with gross abdominal distension and vomiting.
   iii) Intravenous broad spectrum antibiotics to prevent progression of septicaemia.
   iv) Nutritional rehabilitation through nasogastric feeding or parenteral nutrition if available.
8.2. Colonic decompression

There are 2 options for colonic decompression.

i) **Rectal irrigations** using normal saline (20mls/kg per instillation) given through a large rectal tube. Care should be taken to ensure that the tip of the tube reaches above the level of aganglionosis and perforation is avoided by lubrication and gentle push on the catheter. The irrigation is repeated until most of the accumulated faeces is evacuated, ensuring that no more than 20 ml/kg of fluid is retained at once (retention of enema fluid may lead to perforation). Fecal disimpaction may be necessary before embarking on colonic irrigation, and it must be ensured that there is no evidence of intestinal perforation. Subsequently, the irrigation is done 1-4 times daily to keep the colon decompressed until the time of definitive surgical correction.

ii) **Colostomy** is necessary if colonic irrigation is contraindicated or fails to achieve adequate decompression especially in very sick babies, grossly dilated, redundant and flabby colon, hard stools feculoma and uncooperative parents.

The desirable colostomy is one which sites the stoma just above the level of aganglionosis in normally ganglionated colon (leveling colostomy). This can generally be identified by contrast enema and gross appearance. (Figure 11) Biopsies should be taken from the colostomy site. If the colostomy is placed mistakenly on or just above the Transition Zone, obstruction and Hirschsprung’s enterocolitis will persist. (Figure 12) It may be safer to resect most of the distal colon at the time of colostomy otherwise, the large dilated colon may take months to shrink enough for safe pull through and may be responsible for persistent postoperative constipation.

![Transition zone](image)

**Figure 11.** Rectosigmoid transition zone
Some paediatric surgeons prefer using a transverse colostomy (particularly in the absence of facilities for frozen section as it is in most centres in Africa) as it places the stoma as far as possible from the usual rectosigmoid level of aganglionosis. However, a transverse colostomy may make it difficult to clean out the intestine distal to the stoma before definitive correction. A high transition zone may make it necessary to take down the transverse loop colostomy and either resect additional bowel, have a second anastomosis, or refashion a more proximal colostomy. Loop colostomies also invariably prolapse (Figure 13a and b).

An ileostomy may be required when cecal perforation has occurred or in cases of total colonic aganglionosis.
Figures 13a and 13b: Prolapsed loop colostomies with skin escoriation in an infant (a) and older child (b) (source LOA and Prof. E. A. Ameh, ABUTH, Zaria, Nigeria).

8.3. Bowel preparation

Formal bowel preparation is important in the African child to reduce bulk although the practice is becoming less favoured in the developed countries. The significance of bowel preparation prior to definitive pull-through surgery to avoid faecal contamination of the anastomosis and resultant leakage to reduce morbidity and mortality, as emphasized by Nmadu (28) in a review of 2 decades of Hirschsprung’s disease managed in Zaria, Nigeria.

The options for mechanical bowel preparation include:

i) A 3- day preparation comprising mechanical bowel irrigation with warm saline and use of cathartic and antibiotics- neomycin, dulcolax and or thalazole are used in our practice in Ilorin, Nigeria.

ii) One or 2 day bowel preparation with the use of lactulose, Polyethylene glycol (PEG) or Picosalax. These are not readily available in Africa.

8.4. Definitive surgical options

The goals of surgical management for Hirschsprung's disease are to remove the aganglionic bowel and reconstruct the intestinal tract by bringing the normally innervated bowel down to the anus, while preserving normal sphincter function.

We will discuss the available operations, the operative approaches, and whether or not to perform a staging colostomy.

The most commonly performed operations are the Swenson, Soave, and Duhamel procedures (Figure 14). Most of the evidence available reports systematic reviews of the outcome these procedures without a comparison between them. There are no prospective trials comparing surgical treatments of Hirschsprung's disease and surgeons will get the best results doing the operation they have been trained to do, and do with some frequency.
The Swenson procedure is a low anterior resection of the rectum with an end-to-end anastomosis performed by prolapsing the rectum and pulled-through bowel outside the anus.

The Duhamel procedure entails leaving the native rectum in situ and bringing the normally innervated colon behind the rectum with an end-to-side anastomosis 2 cm above the dentate line and joining the two lumens side to side. Originally, this was accomplished by leaving a clamp across the common wall until it fell off, but in more recent years most surgeons use a linear stapler. Adeniran et al (29) have devised a Hand sewn technique for African surgeons who do not have access to the staplers. (Figure 15) This has also assisted in preventing the complications caused by premature dislodgement of clamps. (28)

The Soave endorectal pull-through consists of stripping the rectal mucosa with preservation of the rectal muscular cuff. Ganglionated colon is pulled through the muscular cuff and anastomosed just above the dentate line. The operation was designed to avoid injury to pelvic vessels and nerves, which are theoretically at risk with the Swenson procedure. In the original description, the pulled-through bowel was left hanging out for
several weeks, and was then amputated and the anastomosis was completed. Boley's modification involved a primary anastomosis with the mucosa being excised at same sitting and is the technique employed today.

8.5. Multi-stage vs. Single-stage (primary) Pull-through
Surgery can be done in 2 or 3 stages with a colostomy to protect the anastomosis. Over the years, as surgical and magnification techniques improved, many surgeons began to do the definitive operation at an earlier age and in one-stage. (30), (31), (32) This avoids the known morbidity of stomas in infants and is more cost effective, but a one-stage approach is only advisable with pathologic frozen section support to confirm the transition zone. (33), (34) Many centers in Africa use the barium enema and intra-operative findings to determine this, however, a recent study suggests that 8-10% of children with a rectosigmoid transition zone on contrast study actually have a more proximal pathological transition zone. (35) This may give rise to the ‘acquired Hirschsprung disease’ when an aganglionic remnant is left behind. A single stage pull-through in Africa would be indicated if patients present early without complications and the expertise is available to reduce morbidity of repeated surgery, cost to the parents and long waiting list time.

8.6. Laparoscopy assisted and transanal pull through
Pull-through procedures originally entailed a combination of abdominal and perineal approach but recently laparoscopic assisted (Figures 16 and 17) or exclusively transanal dissection has been applied for all procedures.

Figure 16: Infant positioned with legs suspended for laparoscopy assisted transanal pull-through (BHC)
Figure 17: Laparoscopic view of rectum and pelvis. The colon can be biopsied and the rectosigmoid mobilized laparoscopically before proceeding to the transanal pull through. (source, BHC)

The transanal Soave procedure starts with a submucosal dissection from below with or without laparoscopic biopsy and mobilization of the intra-abdominal rectum. (32)(33) The rectal mucosa is incised circumferentially 1-2cm above the dentate line, and the dissection is continued along the rectal wall. The entire rectum and part of the sigmoid colon can be delivered through the anus. The transition zone is identified, and the anastomosis is performed from below. This operation has been shown to be safe and associated with a short hospital stay, early feeding, and minimal analgesia requirements compared with the open Soave operation. (36) Options for accessing the proximal bowel in order to do the biopsy include laparoscopy or a small umbilical incision, which can also be used to mobilize the splenic flexure in children with higher transition zones.

The transanal approach can be used in a patient with a preexisting colostomy, either taking down the colostomy to use for the pull-through or closing it at a third procedure.(Figures 18-21)

Figure 18: Infant positioned for a trans-anal pull through procedure (LOA).
Figure 19: Anal retraction with sutures in the absence of an expensive ‘lone-star’ retractor (seen in Fig.20)

Figure 20: Transanal pullthrough of sigmoid colon prior to anastomosis. (BHC)
8.7. Total Colonic Hirschsprung’s disease
Laparotomy is done to take multiple biopsies including the appendix to identify the pathological transition zone, which may differ from what the surgeon sees grossly. An ileostomy is left in the patient and a definitive reconstructive procedure is planned for an older age (6-18 months).
The options for reconstruction are: **colonic patch, straight ileoanal pull-through** and **J-pouch construction**. A segment of colon has been retained for water absorption (2), but the aganglionic colon gradually dilates and some of the patients develop severe enterocolitis that requires removal of the patch. (37) Hence, the short Duhamel small bowel-rectal patch is recommended.
Straight pull-throughs are performed using any one of the standard pullthrough techniques. The J-pouch procedure is the same as that performed commonly for children and adults with ulcerative colitis and polyposis. (38) Little has been written about the results of this approach in the management of HD.

8.8. Near-total intestinal aganglionosis
Children with HD involving the colon and entire small bowel usually do not survive because they do not have adequate nutrient absorptive capacity. The surgical options available are:
a. Bowel tapering, imbrication, or bowel-lengthening procedures, such as the Bianchi or STEP (Serial Tapering EnteroPlasty) operation for children who develop significant proximal dilatation of the normally innervated bowel. (39)
b. Zeigler technique of myectomy-myotomy. (40)
c. Small bowel transplantation. (41)

8.9. Postoperative care
Following the formation of a stoma, bowel function usually returns within 24-48 hours and oral feeding can be commenced usually with breast milk in the infants and maize or sorghum-based paste in older children. The mothers are taught about how to take care of the stoma on the wards before discharge which usually takes place after the 7th day of operation if there are no other concerns. Because of the cost and non availability of stoma bags, Zinc oxide in petrolatum paste (Vaseline) is used to protect the skin and local clothing materials or napkins are used to wrap the stoma to receive the faeces (Figure 22). Mothers are advised to change the receptacle as soon as faeces are collected on it so that contact with the skin is limited.

![Figure 22: A home-made ostomy appliance from Guyana. (BHC)](image)

After a pull-through procedure most infants begin bowel movements within 24 hours and can start liquid feeds right away. In trans-anal pull-through with or without laparotomy oral intake in form of liquid diet is usually commenced after 48 hours to delay fecal contact with the peri-anal wound. Adequate parenteral and enteral analgesia is provided and a caudal block, when available, provides additional pre-emptive analgesia. Anal dilatation is commenced after 2 weeks and mothers are taught to use their fingers, Hegar’s dilator or trimmed candle stick 1-2 times daily for up to 3-6 months to prevent anal stricture.

### 9. Complications

#### 9.1. Intra-operative and Early Postoperative Complications

The complications of surgery for HD include the general group of complications of any abdominal surgery, including bleeding, infection, injury to adjacent organs, and the risks of anesthesia. Those children who undergo a staged procedure with a preliminary stoma may experience stoma-specific complications such as prolapse, skin breakdown, stricture, and retraction. (42)

Anastomotic leak and strictures occur infrequently, and can be avoided by close attention to adequate blood supply of the pulled-through bowel. Minimizing tension on the anastomosis will help prevent ischemia and retraction of the pull-through. The incidence of anastomotic leak in laparoscopic and transanal pull-through appears to be lower than that reported in the older literature of open pull-throughs. The complications reported with primary pull through are less than with staged pull through. (43) Postoperative perineal excoriation is common and can be treated with a zinc based cream. In children with a colostomy, the perineal skin can be toughened by applying stool to the perineum for several weeks preoperatively to diminish postoperative excoriation.
9.2. Late Complications

Two categories of patients have been identified that are not doing well after the pull through for HD (44) (45). They are:
i) those that are distended with recurrent episodes of enterocolitis which may cause failure to thrive and
ii) those who are soiling or have fecal incontinence.

The incidence of postoperative enterocolitis is reported from 5-35% and partly depends on how strictly one defines enterocolitis. (46) Enterocolitis may occur postoperatively, and it is extremely important that the surgeon educate the family about the risk of this complication which may be life-threatening. Parents should be urged early return to the hospital if the child should develop any concerning symptoms such as distension associated with foul-smelling diarrhea and signs of systemic infection.

Enterocolitis and obstructive symptoms may be associated with internal anal sphincter dysfunction. This can be treated by injection of botulinum toxin into the sphincter, anal myectomy, or topical nifedipine cream.

In some severe cases of obstructive symptoms, the child may be best served by use of a cecostomy and administration of antegrade enemas as described by Malone, or even by the creation of a proximal stoma. (47) Malone operation may not be acceptable in the African setting and the patient may do better with dietary management. (Table 3)

**TABLE 3**: Potential causes of Late Complications following a Pull-through

<table>
<thead>
<tr>
<th>Incidence</th>
<th>8-30%</th>
<th>5-35%</th>
</tr>
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<tbody>
<tr>
<td>Causes</td>
<td>Mechanical obstruction</td>
<td>Abnormal sphincter function</td>
</tr>
<tr>
<td></td>
<td>Persistent or acquired aganglionosis</td>
<td>Abnormal sensation</td>
</tr>
<tr>
<td></td>
<td>Colonic motility disorder</td>
<td>Overflow incontinence due to constipation</td>
</tr>
<tr>
<td></td>
<td>Internal sphincter achalasia</td>
<td>Abnormal mucin</td>
</tr>
<tr>
<td></td>
<td>Stool-holding behavior</td>
<td>Obstruction</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Genetic or immunologic predisposition</td>
</tr>
</tbody>
</table>

Postoperative bowel dysfunction is evaluated by taking a detailed history noting the patient’s bowel habits, use of anti-motility drugs or laxatives, need for dilation or irrigation, and the type of previous surgery. Patients with an associated syndrome and those with long-segment disease have been found to have poorer outcomes. (37), (48)

A contrast enema will provide information about the degree of megacolon, fecal impaction and presence of a stricture or transition zone.

The digital rectal examination, preferably under general anaesthesia, should evaluate the integrity of the anal canal, location of anastomosis in relation to the dentate line, presence of stricture, sphincter tone, and presence of large rectal pouch or a Soave cuff. A full thickness rectal biopsy should be done to rule out persistent...
aganglionosis. Persistent aganglionosis may be due to pathologist’s error on the original specimen, or a transition zone pull-through, and in some cases there may be ganglion cell loss after a pull-through.

Although some strictures can be managed using repeated dilations, some require revision of the pull-through. Duhamel spurs can be resected from above or managed by extending the staple line from below, with or without laparoscopic visualization.

It is well recognized that children with HD may have associated motility disorders, with histologic abnormalities such as intestinal neuronal dysplasia. (49) Investigations for motility disorder may include a radiologic shape study, colonic manometry, and laparoscopic biopsies looking for intestinal neuronal dysplasia. (50), (51) If a focal abnormality is found, consideration should be given to resection and repeat pull-through using normal bowel. If the abnormality is diffuse, the appropriate treatment is bowel management and the use of prokinetic agents.

10. Outcomes

Most children with Hirschsprung’s disease overcome the postoperative problems and do well. (52) Studies have suggested that obstructive symptoms and incontinence seem to resolve with time, and that the risk of enterocolitis, in the absence of an ongoing obstructive cause, is almost eliminated after the first 5 years of life. Sexual function, social satisfaction, and quality of life all appear to be relatively normal in the vast majority of patients. (53-54) A recent large multicenter review showed no significant differences in continence or stooling patterns after transanal versus transabdominal pull through procedure. (55)

11. African experience

Hirschsprung’s Disease management is a major problem in the practice of surgery in Africa; many patients may default on follow up appointment and would never get to complete their scheduled procedures for various reasons. (6-7) Reasons include: lack of funds for treatment (non-availability of health insurance), parents or babies having an illness at the time of appointment or parents having another engagement. (56) Perhaps some patients may have died from complications of initial intervention or the disease. There are instances when patients are referred to colleagues for follow up and possible completion of intervention either by choice of the parents because of reason of proximity/closeness. Feedbacks are never gotten back from these referrals which make completion of reports difficult.

12. Recommendations

1. Public enlightenment and awareness about Hirschsprung’s Disease should be stepped up.
2. Parents and other care givers (including general practitioners) should be made aware of the classical features of presentation and that the supposed ‘fountain sign’ (projectile flush of watery offensive stool on digital rectal examination) in children with constipation is a feature of enterocolitis which suggest danger that needs urgent and adequate intervention.
3. Parents should also be educated as to the signs and symptoms of enterocolitis so that they can seek medical attention early.
4. For patients that are not doing well post pull-through, efforts should be made for proper evaluation and expert treatment so that the quality of life of the patients is optimized.
5. There should be feedback or a form of collaboration among practitioners and surgeons so that many questions, yet to be answered about epidemiology and pathology of Hirschsprung’s disease in Africa, are addressed and support from developed countries is secured.
6. The algorithm at the end of this article summarizes our approach to diagnosis and treatment of HD (Figure 23)

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Figure 23: Algorithm for management of patients with suspected Hirschsprung’s disease

Newborn with delayed passage of meconium
- Plain abdominal x-rays
- Contrast enema

Enterocolitis, suspected HD
- Fluid resuscitation
- IV Antibiotics
- Rectal irrigations
- +/- Colostomy

Chronic constipation
- Contrast enema

Full thickness or suction rectal biopsy confirms HD

Rectal irrigations followed by one-stage pullthrough

Trans-anal, laparoscopic or open pullthrough procedure

Post-op anal dilation

Levelling colostomy above transition zone
OR transverse loop colostomy

Postoperative incontinence, encopresis or enterocolitis
- EUA, rectal biopsy, contrast study, motility studies

Anal dilation, anal sphincter botox or myectomy, redo pullthrough or bowel management program

 +/- 3rd stage
Close colostomy
13. References


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24. **Wellcome.** [Online] [Cited: November monday, 2010.]
[http://images.wellcome.ac.uk/indexplus/obf_images/f2/06/11b8086bf856558f2595791db273.jpg](http://images.wellcome.ac.uk/indexplus/obf_images/f2/06/11b8086bf856558f2595791db273.jpg)


45. Levitt MA, Dickie B, Pena A. Evaluation and treatment of the patient with Hirschsprung disease who is not doing well after a pull-through procedure. 2010, Vol. 19, pp. 146-153. [http://simplelink.library.utoronto.ca.myaccess.library.utoronto.ca/url.cfm/122660](http://simplelink.library.utoronto.ca.myaccess.library.utoronto.ca/url.cfm/122660)


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