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# Hirschsprung Disease at Children: Report of One Case

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**Abstract:** ***Hirschsprung disease is a rare motor disorder of the gut that is characterized by absence of ganglia in distal colon resulting in a functional obstruction. Most cases of Hirschsprung disease are the diagnosted in the newborn period and is characterized by the total absence of intramural ganglion cells of the submucosal (Meissner) and myenteric (Aurbach) neural plexus in the affected segment of the bowel. Hirschsprung disease should be considered in any newborn that fails to pass meconium within 24-48 hours of birth 94% of cases are diagnosted before the patient reaches 5 years of age,mild cases of Hirschsprung disease may go underdiagnosted until he or she reached adulthood. In this study, we describe one case of infant with Hirschsprung disease ,5months old, presenting to the University Hospital Center of Tirana. He had a history of longlasting recurrent constipation, realived by laxatives, progressive abdominal distendion, colicky pain, emesis. Barium Enema reveal a grossly distended descendent colon, sigma and rectum. HD should be considered in the differential diagnosis of cases where patients present with constipation, acute megacolon, hypothyroidism, intestinal motility disorder, irritable bowel sindromy, toxic megacolon. The treatment of HD is surgery.***

**Keywords:** Hirschsprung Disease, constipation, functional obstruction, treatment

## 1. Introduction

Hirschsprung Disease was described by Ruysch in the 1691 and popularized by Hirschsprung in 1886, the pathophysiology was not clearly determined until the middle of the 20-th century, when Whitehouse and Kernohon reported aganglional of the distal colon as the cause of obstruction in a case series.[1]

In 1949, Swenson described the first consistant definitive procedure for Hirschsprung Disease, rectosigmodectomy with coloanal anastomosis. In patient with Hirschsprung Disease, both myenteric and submucosal plexures are absent. The cause of Hirschsprung Disease is most commonly of neuroblasts originaty from the neural crest in functional intestinal obstruction.[2.3]

Enteric ganglion cells are the primary pathogenic entity in HD, some studies suggested that other cell types may also be implicated.[4.5.6.7.8]

In about 75% of all cases affected by this abnormality, the aganglionosis is confined to the rectum and sigma. About 17% of all patients show an extended disease and in about 5-8% the absence of ganglia is present in the total large bowel and terminal ileum. Only a small number of patients exist with extended aganglionosis including a large portion of the small bowel and upper gastro-intestinal tract. The classical Hirschsprung Disease patient, syndromic Hirschsprung disease and rare neurocristopathy exist. Ondine syndrome or Waardenberg syndrome are part of this entity.

20% of patient with Hirschsprung Disease have associated abnormalities. Down Syndrome 8%, cardiac defects 8%, gastrointestinal abnormalities 4%.The disease occurs more often in male than in females, with a male to female ratio of approximately 4:1.

Hirschsprung disease affects all races; however, it is roughly 3 times more common among Asian-Americans.[9]

The typical clinical signs of Hirschsprung Disease are delayed meconium passage, abdominal distension, vomiting and enterocolitis. More than 80% of all Hirschsprung cases present symptoms in the neonatal period. Only a few of these are having a prenatal diagnosis (mostly performed by intrauterine MRI and ultrasound).

Enterocolitis is present in one third of babies and toddlers with HD and associated with diarrhea. Enterocolitis is still the most common cause of death in HD and especially in Down's Syndrome a severe hazard for the patient's life.[1] The contrast study was one of the most important exams. Barium Enema showed the typical proximal dilatation above the distal narrow segment is usually always present in elder children. In the newborn the dilatation can be absent. The histological exam to confirm Hirschsprung Disease, is showing the absence of ganglia structures in a rectal biopsy.

Acetylcholinesterase staining reveals hypertrophied nerve trunks throughout the lamina propria and muscularis propria layers of the bowel wall. Recent studies suggest that immunohistochemical (IHC) staining for calretinin might be more accurate than acetylcholinesterase staining in diagnosing congenital aganglionosis in suction biopsy specimens.[15]

Many pathologists applied prefer to repeat the exam to wait for a "maturation" of nerve structures avoiding the risk a false positive diagnosis.

Anorectal manometery showed high pressure peristalsis in proximal ganglionic bowel and lack of progressive peristalsis in a normal pressure zone lacking ganglion cells and failure of relaxation of rectal sphincter in response to rectal distention. Although rarely used as primary diagnostic modality.

This examination is valuable for evaluating functional results after reconstuctive procedures.

There are many different surgical procedures Swenson, Duhamel, Soave, Rehbein, Boley that are described and performed. In the last 20 years different authors introduced laparoscopic assisted techniques, made popular by Keith Georgeson and others.

The most recent modification was the introduction of the transanal approach, known as the de la Torre technique. For long and total Hirschsprung Disease many modifications of the above mentioned techniques were described and applied. All techniques follow the above-mentioned principles to bring ganglionotic, normal bowel as close as possible to the sphincter complex. The resection at the level of the rectum is in all cases different due to technical details. The Swenson procedure is clearly the most radical approach, doing a total resection right above the sphincter complex with an end-to-end anastomosis. Soave's techniques ends up with a similar anastomosis, but leaves the cuff of aganglionotic muscle inside, which is divided and opened. The Duhamel leaves the rectum with aganglionotic muscle in place, doing a lateral posterior anastomosis. Rehbein did in his approach an extremely deep resection of the rectal part, leaving a short distance to the sphincter complex. The modifications of De la Torre and the Georgeson approach repeat a Soave-like technique, but starting the mucosectomy from the anus, preparating the rectum from below. Each technique has its own technical difficulties and risk, as well as surgical complications which might lead to the known consequences and problems of surgery for Hirschsprung`s disease. Therefore the aim of any surgery should be a patient who is having regular stool frequency and bowel function, no more enterocolitis and stool retention and faecal continence. Surgery should be performed ideally as early as possible in life, using a minimal-invasive technique for short post-op course and excellent cosmetically results. The risk for surgical early and late surgical complications should be minimized. All this factors finally contribute to a good quality of life. In criticall ill babies with a septic situation or a long aganglionosis an enterostomy can be a lifesaving procedure.

## 2. Case Report

A 5- months old male admitted to General Pediatric the University Hospital Center of Tirana with diagnosis of admission Acute Diarrhea. The child A.B has 2 days with bilious emesis, abdominal distention, loss of appetite, dimness, subfebrile temperature.

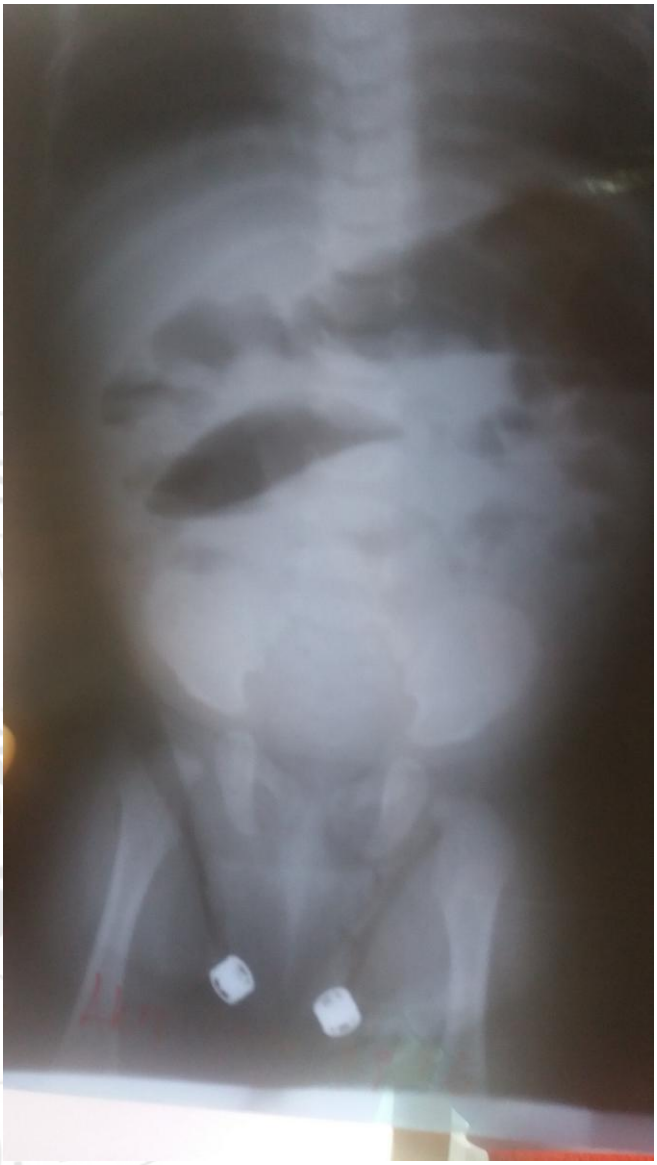
In physical examination he appeared ill. The history of life the mother refered that child has passed meconium 48 hours after births. The child was born premature with cesarean secion, birth weight 3200gr.

On physical examination showed a markedly distended abdomen, his facies was pale and suffered.

Laboratory investigation on admission revealed a blood cell count WBC 5500 (45 % lymphocytes, 7% monocytes, 48% granulocytes ), RBC 4 540000, hemoglobin level 9.6 g⁄dl, hemotocrit value 36.5 %, plated count 460000, total protein 5.1g⁄dl, kalium value 3.1mmol⁄l, chloride value 112mmol⁄l.

Abdominal ultrasonography showed dilatation of descendent colon, sigma and rectum.

Plain abdominal radiography showed a dilatation descedent and ascedent colon.(Fig 1)



Barium enema showed the aganglionic distal segment was narrow with distended normal ganglionic bowel above it. We looked a transition zone, delayed evacuation ›24 h.

In the examination of rectum that make gastro-hepotologic pediatric showed rectal ampulla was empty. We consulted with pediatric surgeon and decided to make rectal biopsy that result no ganglion cells.

The patient was started antibiotic treatment with cephalosporin for prevent complication of HD. He treated with rehydration therapy.

## 3. Discussion

Hirschsprung Disease is a disease that should be diagnosted in the newborn period. Hirschsprung Disease should be

considered in any newborn that fails to pass meconium within 24-48 hours of birth.

Untreated aganglionic megacolon in infancy may result in a mortality rate as high as 80%.

Possible complication of surgery include anastomic leak 5%, anastomic stricture 5-10%, intestinal obstruction 5%, pelve abscess 5% and wound infection 10%.

Long segment disease could have long term complication include chronic obstructive symptoms, incontenience chronic constipation, enterocolitis and late mortality.

Long term follow up studies have shown that greater than 90% of children experience significant improvement.[10] Patients with a syndromic association and those with long-segment disease have poorer outcomes.[11.12.13]

Symptoms of Hirschsprung Disease in children include bowel obstruction with bilious vomiting, abdominal distension, poor feeding and failure to thrive.
About 10% of children present with diarrhea caused by enterocolitis, which is hypothesized to to stem from stasis and bacterial overgrowth. This may progress to colonic perforation, causing life-threatening sepsis.[14]

Diagnosis of Hirschsprung Disease made from from physical examation, abdominal X-ray, contrast Enema, Rectal biopsy, Anorectal manometry.

In all cases of Hirschsprung Disease, surgery is the definitive treatment.

## 4. Conclusion

Our case was presented in General Pediatric with Acute Diarrhea and undiagnosed until the child arrived 5 months old. Neonates should be careful in evaluation in any infant who does not pass meconium with the first 24 hours of life after birth. Pediatrician should be suspected in newborn intestinal obstruction, constipation, chronic abdominal distention in the first year of life.

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