DIAGNOSIS AND TREATMENT OF HIRSCHSPRUNG’S DISEASE IN CHILDREN

Bălănescu R.1, Topor Laura1, Nedelea S.2

1 Surgery Clinic of “Grigore Alexandrescu” Emergency Hospital for Children
2 Carol Davila” University of Medicine and Pharmacy, Bucharest, Romania

Abstract. Background. Hirschsprung disease is a relatively rare condition, initially described as a cause of constipation in early infancy, and it is managed by pediatric surgeons. Significant advances have been made in understanding its etiologies in the last decade, especially with the explosion of molecular genetic techniques and early diagnosis. The surgical management has progressed from a two- or three-stage procedure to a primary operation.

Methods. This retrospective study involves 74 patients diagnosed with Hirschsprung’s Disease at the Surgery Clinic of “Grigore Alexandrescu” Emergency Hospital for Children from 1.1.2000 to 1.1.2011. For better analysis of diagnosis and treatment evolution, the group was split according to admission date into two smaller groups, each one containing 37 patients.

Results. Age at diagnosis has decreased in 2006-2011 interval, 32.4% of patients being newborns and 24.3% infants. In 2000-2005 only 24.3% were newborns and 21.6% infants. This results are correlated with a shorter hospital stay and period from diagnosis to surgery. Our results are all compared with the literature dates at the moment.

Conclusion. Our clinic has improved its diagnosis and treatment methods for HD during the last 11 years. The age at diagnosis has decreased, along with the hospital stay and the time from diagnosis to surgery. Different procedures are used according to the case. As a result, patients are getting better and more suitable treatment with lower costs and lesser complications. Follow-up studies have shown very good results in long-term quality of life.

Keywords: congenital, megacolon, aganglionosis, pull-through, soave

Introduction

Congenital megacolon is eponymously named after Harold Hirschsprung, who accurately described the clinical features in 1886.[1] Hirschsprung disease is a relatively rare condition managed by pediatric surgeons. Significant advances have been made in understanding its etiologies in the last decade, especially with the explosion of molecular genetic techniques and early diagnosis [2].

International studies have reported rates ranging from approximately 1 case per 1500 newborns to 1 case per 7000 newborns. This condition is a developmental disorder of the enteric nervous system and is characterized by the absence of ganglion cells in the myenteric and submucosal plexuses of the distal intestine. This results in absent peristalsis in the affected bowel and the development of a functional intestinal obstruction. [3]

Congenital megacolon is classified depending on the aganglionic segment into the following types:

- ultrashort segment HD – only anal canal and terminal rectum is aganglionic.
- short-segment HD – anal canal and rectum is completely involved (80%).
- long-segment HD – anal canal, rectum and pert of the colon is involved (10%).
- total colonic HD – anal canal, rectum and full length of colon is involved.[4]

The appropriate diagnostic approach may vary, depending on the age of the patient and the presenting clinical picture [3]. Contrast enema (CE), anorectal manometry (ARM), and rectal suction biopsy (RSB) are the most important tests used for the diagnosis. RSB is the most accurate test for diagnosing HD, and it has the lowest rate of inconclusive test results. [5]
The surgical management has progressed from a two- or three-stage procedure to a primary operation. [1]

The difficulties encountered in the early procedures were in large part due to technical errors resulting from a poor understanding of the pathophysiology of this disease. As our understanding of Hirschsprung’s disease improved, operations evolved into those that are presently performed. [6]

One-stage correction of Hirschsprung’s disease is a safe procedure in all age groups. It offers economical and social advantages to families in developing countries. Benefits of 1-stage treatment include avoidance of multiple operations, elimination of complications associated with a colostomy, shorter duration of hospital stay, and completion of treatment at an earlier age. [7] More recently, definitive surgery for Hirschsprung disease through minimally invasive techniques has gained popularity. In neonates, the advancement of treatment strategies for Hirschsprung disease continues with reduced patient morbidity and improved outcomes. [1]

**Objectives**

Our objective was to review the actual diagnostic and treatment principles and methods, to point out the importance of a correct and rapid diagnosis and finally to analyze the diagnosis and treatment evolution in our clinic.

**Material and methods**

This retrospective study involves 74 patients admitted to the Surgery Clinic of “Grigore Alexandrescu” Emergency Hospital for Children from 1.1.2000 to 1.1.2011. We have selected all the patients with a confirmed diagnosis of Hirschsprung’s Disease (HD). We have recorded the following information for every patient: identification data, sex, age at diagnosis and surgery, hospital stay both in Intensive Care Unit and Surgery Clinic, clinical manifestations, comorbidities, histopathological diagnosis, disease type, type of treatment, surgical procedure and evolution.

For better analysis of diagnosis and treatment evolution we have split the group of patients into two smaller groups, each one containing 37 patients. The first group consists of all patients admitted from January 2000 to November 2005, and the second one all the patients admitted from December 2005 to January 2010.

**Results**

**Sex Ratio**

HD occurs more often in males than in females, with a male-to-female ratio of approximately 4:1. However, with long-segment disease, the incidence increases in females. 76% of the patients in our group were males and only 24% females (Male: Female ratio of 3:1).

**Disease types**

Short segment HD is the most common type of disease (44,6% of the cases) followed by ultra-short segment HD (25,7% of the cases), long segment HD (20,3% of the cases) and total colonic HD (9,5% of the cases). These findings resulted from both paraclinical examinations (irrigography and histopathological exams) and intraoperative assessment. The high prevalence of ultra-short segment HD is correlated with studies that report it in up to 25% of the cases.

![Figure 1. Disease types](image)

**Age at HD diagnosis**

HD is uncommon in premature infants. Currently, approximately 90% of the patients with HD are diagnosed in the newborn period. HD should be considered in any newborn who fails to pass meconium within 24-48 hours after birth, or in any child with a history of chronic constipation since birth. Although contrast enema is useful in establish the diagnosis, full-thickness rectal biopsy remains the key for diagnosing HD. It is important that the infant should not have rectal washouts or even digital examinations prior to barium enema, as such interference may distort the transitional zone appearance and give a false-negative diagnosis [15].

In our study, age at diagnosis has decreased over time, probably due to a higher HD suspicion. The number of patients diagnosed before the age of 1 has increased in the second group, with a decrease in the number of patients diagnosed after the age of 1. Diagnosis at 0-30 days of age has increased from 24.3% to 32.4%, at 1-12 month from 21.6% to 24.3%.

**Clinical manifestations**

The most important symptom in neonates with HD is delayed passage of meconium. About 90% of the neonates having HD fail to pass meconium
in the first 24 hours of life [15].

Almost one third of the babies who were diagnosed with HD before age of 3 months have diarrhea as a cardinal symptom [14]. In this situation, diarrhea always means enterocolitis, which remains the commonest cause of death [15].

As we could easily see, clinical manifestations are often digestive complications of the HD. Chronic constipation is the main symptom that takes the patient to the doctor in both groups, followed by bowel obstruction and enterocolitis. Their prevalence is higher in the later interval: constipation (from 60% to 65%), bowel obstruction (from 33% to 47%) and enterocolitis (from 3% to 26%). Late meconium passage as a clinical manifestation was only present in the second interval probably due to a better post-natal surveillance and a higher HD suspicion. Only a few patients presented to our clinic for perforation.

Comorbidities and complications

About one fifth of the patients have a neurological, cardiovascular, urological or gastrointestinal problem [15]. Many patients have general complications due to chronic malnutrition: carential anemia (24.3%) and failure to thrive (25.7%). A small percentage presented with pneumonias (4.1%) and urinary tract infections (6.8%). HD has been described in association with numerous genetic abnormalities and congenital malformations, such as Down Syndrome, neurocristopathy syndromes, multiple endocrine neoplasia type II. 5.4% of our patients had genitourinary malformations, represented by hypospadias and undescended testicle, 2.7% had associated Down Syndrome and 2.7% had Atrial Septal Defect.

Hospital stay

Hospital stay has decreased due to multiple factor such as: a more rapid diagnosis, a decrease in complicated forms of the disease and a more effective postoperative care. The medium hospital stay was 27.47 days in 2000-2005 interval and only 20.86 days in 2006-2011 interval. The number of days in the Intensive Care Unit has decreased subsequently.

The results are better reflected in a lower cost hospital care, a decrease in nosocomial infection risk and a faster psychosocial reintegration.

Age at first intervention

Although age at diagnosis has dropped in the second interval, there are more important differences in age at first intervention. The percentage of newborns has risen from 8.1% to 21.6%, and that of infants from 21.6% to 29.7%. Subsequently, we recorded a drop of surgical interventions at patients older than 1 year.

First intervention

In the last ten years, most of the cases are diagnosed in the neonatal period, and this is probably the most important reason for which there have been created one-stage pull-through operations, with minimal morbidity rates and encouraging results. [15]

In this study, according to patient’s status, age, anatomo-pathological type and clinic’s experience the first intervention may be temporary or definitive. The percent of definitive procedures has increased from 54.1% in 2000-2005 to 62.2% in 2005-2011

Time from diagnosis to surgery

There are some advantages in operating on the newborn with HD, and these are resulting from the
fact that we can easily control the colonic dilation by washouts. Another very important aspect is that the caliber of the pull-through bowel is almost normal, and this will permit an appropriate anastomosis, with minimum leakage and cuff infection [15].

Here, the time from HD to diagnosis has decreased in the second group. In the 2000-2005 interval 59.5% of the patients had surgery in the first month from diagnosis. The percentage has increased to 78.4% in the second period. The number of surgeries in the 1-12 months interval is approximately the same but no surgeries were made at more than 12 months from diagnosis in the last 5 years.

Pull-through procedure

The surgical options vary according to the patient's age, mental status, ability to perform activities of daily living, length of the aganglionic segment, degree of colonic dilation, and presence of enterocolitis. Surgical options include colostomy at the level of normal bowel, rectal irrigations followed by rectal resections with a pull-through procedure once bowel caliber is restored to normal, and a staged procedure with placement of a diverting colostomy followed by a pull-through procedure.

There are a number of definitive procedures that have been used over time. 4 most commonly performed repairs are the rectosigmoidectomy described by Swenson and Bill, the retrorectal approach developed by Duhamel, the endorectal procedure developed by Soave and deep anterior colorectal anastomosis described by Rehbein. All these procedures have a common idea, which is that of bringing the aganglionic bowel down to the anus [15]. Recently, the surgery is being performed in the newborn period, using minimally invasive surgical techniques, like laparoscopy.

The Swenson procedure was the first one used to treat HD. The aganglionic segment is resected down to the sigmoid colon and the remaining rectum, and after that the normal colon and the low rectum are stuck by an oblique anastomosis.

In the Duhamel procedure, the aganglionic bowel is resected down to the rectum, and the rectum is oversewn. The proximal bowel is then brought through the rectorectal space and an end-to-side anastomosis is performed to the rectum.

The Soave procedure consists of removing the mucosa and submucosa of the rectum and pulling the aganglionic bowel through the aganglionic muscular cuff of the rectum. This procedure was modified by Boley to include a primary anastomosis at the anus.

TERPT – The resection and endo-rectal transanal pull-through – described for the first time by De la Torre-Mondragon and Ortega-Salgado – this technique realizes both the resection and the pull-through by transanal way, and there's no need of any abdominal approach. The whole procedure is done from below in a similar way to perineal rectosigmoidectomy. The advantages of this technique are those of avoiding both the laparotomy and the laparoscopy, the scars of the abdomen wall, the contaminations of the peritoneum.

Laparoscopic pull-through technique – was first described in 1999 by Georgeson; first, the transition zone is identified using laparoscopy, and then the rectum is mobilized below the peritoneal reflection. After that, a transanal mucosal dissection is done, and finally the rectum is prolapsing through the anus and linked to the segment from above.

In our clinic, except for the ultra-short segment HD, the treatment is represented by an abdomino-perineal pull-throgh. Duhamel procedure was used in 83% of the cases. In the last five years we have successfully started using a modified Soave procedure in which rectal mucosal dissection is done during the perineal time of the procedure. Swenson and Lester Martin procedures were other used procedures. Due to our limited experience, the minimally invasive surgical techniques are not used yet for treatment of HD, but in the next future we will manage this problem, too. However, it’s a real challenge to compare the results of the three techniques used until now, because the incidence

![Figure 5. First surgical intervention](image)

![Figure 6. Rectal mucosal dissection during perineal time](image)
of complications is related to the skill of the individual surgeon and even to the year of the study. The long term outcomes of these procedures appear to be similar.

![Figure 7. Abdomino-perineal pull-through procedure](image)

### Evolution

Untreated, aganglionic megacolon in infancy may result in a mortality rate of as much as 80%. Although the diagnosis and treatment were substantially different in the two intervals, the evolution was mainly correlated with the type of disease.

There were 4 cases of exitus, 2 of them at patients with long-segment HD and 2 of them at patients with total colonic HD. 2 newborns with total colonic HD requested transfer to HD specialized clinics abroad. We have defined satisfactory evolution as presence of temporary ileostoma or colostoma. All the patients with ultra-short segment disease and more than 90% of those with short segment disease had a favorable evolution.

### Follow-up

The outcome in infants and children with HD is generally quite good. Most children obtain fecal continence and control. However, children with Down syndrome may be expected to have lower rates of continence.

Although many patients will encounter one or more of some problems postoperatively, long-term follow-up studies have shown that more than 90% of most children experience significant improvement and will do relatively well. Patients with long-segment disease have been found to have poorer outcomes.

Advances in surgical technique, including minimally invasive procedures, and earlier diagnosis have resulted in decreased morbidity and mortality for patient with HD. There are some advantages we have to consider when thinking in laparoscopic techniques: minimal postoperative pain, the bowel is gaining faster its function, the hospital stay is shorter and also the cosmetic aspect is significantly improved.

### Discussion and conclusion

Age at diagnosis has decreased in 2006-2011 interval, 32.4% of patients being newborns and 24.3% infants. In 2000-2005 only 24.3% were newborns and 21.6% infants. This results are correlated with a shorter hospital stay and period from diagnosis to surgery leading to lower costs and a shorter healing process.

Sex ratio (Male: Female=3:1) and disease type (short segment being the most frequent) are in concordance with international studies. Age at HD diagnosis is in a continual decrease. An increasingly number of patients are diagnosed as newborns and infants. We have recorded an important decrease in total hospital days as well as in the ICU.

We aim for more one-stage interventions, thus their prevalence has increased from 54.1% to 62.2% in the second interval. In 2006-2011 all the interventions have taken place in the first year from diagnosis, most of them (78.4%) in the first 30 days. Duhamel procedure is the most common but our modified Soave procedure has gained field recently due to better recovery, lesser complications and shorter hospital stay.

### References


9. Hackam DJ, Reblock K, Barksdale EM, et al. The infl-


