Hirschsprung’s disease in adults.

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INTRODUCTION

Hirschsprung’s disease is the malformation of the hindgut characterised by the absence of intramural ganglion cells in the submucosal and myenteric plexuses and manifested by megacolon. Congenital character of this disease stipulates its belonging in general to the scope of activity of the pediatric clinics. Thanks to the progress of the pediatricians problems of diagnosis and treatment of Hirschsprung’s disease have been successfully resolved.

Key words: Hirschsprung’s disease, adults, treatment

However, this disease in many cases can be presented without pronounced clinical manifestation when constipation has periodical and unstable character and is easily subdued by enemas. All this allows such patients to reach rather an advanced age. Usually their first visit to a doctor can happen in the case of functional decompensation of the colon which requires urgent surgical intervention.


However, in modern literature there is no information about peculiarities of the clinical development of the Hirschsprung’s disease in adults. There is not enough data on criteria of diagnostic and surgical strategy used for treatment of this abnormality.

This work is based on the results of the investigation and treatment of 84 adult patients with aganglionosis of the bowel in the State Scientific Centre of coloproctology from 1977 to 2005. The age of the patients varied from 14 to 47. The average age was 24.3 ± 8.3 years. There were 59 (70.2%) male patients and 25 (29.8%) female patients. The distribution of the Hirschsprung’s disease patients according to their gender and age is presented in table 1.

The table clearly shows that majority of the patients - 62 (73.0%) - were at the age of 14 – 29. Most of the patients were male – 59 (70.2%).

The main criteria of inclusion was their confirmed aganglionosis of the colon and absence of anamnesis of any surgical interventions because of the Hirschsprung’s disease and its complications until 14 years of age.

Upon admission to the clinic 61 (72.6%) out of 84 patients made complaints of absence of defecation. The inflation of abdomen was registered in 41 (48.8%) of them. The abdominal pain was presented in 32 (38.1%) cases. The combination of complaints was found in 54 (64.3%) patients.

Majority of the patients had constipation already during their first year of life, although their low intensity and regular conservative treatment allowed them to reach rather an advanced age. The periods of life during which the clinical manifestation of the Hirschsprung’s disease appeared, are stated in table 2.

The periods of life in which the first clinical signs of the disease appeared (n = 84).

The clinical manifestation of the Hirschsprung’s disease in adults in 64 (76.2%) cases developed in neonatal period or early childhood, in 18 (21.4%) cases – in childhood,
and only 2 (2.4%) cases corresponded to the so called latent type of disease, when its first appearance occurred in a more advanced age (Vorobyov G.I., 1982).

However, due to the unclear clinical picture of disease in childhood the correct diagnosis was made only in 16 (19.0%) out of 84 patients and surgical treatment was not carried out.

Failure of conservative treatment resulted in the necessity of urgent operation in the advanced age in 26 (30.9%) patients prior to observation in our clinic. The surgical procedures had palliative character and did not include the resection of the aganglionic segment which led to the referral of those patients to specialized hospital for further treatment. The types of surgical procedures are stated in Table 3.

All patients underwent a complex investigation including clinical and instrumental methods. Barium enema examination was the main method of the Hirschsprung’s disease diagnosis. The presence of the narrow zone in the distal part of the colon with a proximal cone-shaped segment is a symptom of the abnormality. According to our observations the typical X-ray picture was marked in 69 (83.1%) out of 83 patients who underwent this examination.

Most of the patients – 76 (90.5%) out of 84 - in addition to the barium enema anorectal manometry to define rectoanal inhibitory reflex was carried out. In those cases, when patients had suffered from constipations since early childhood and narrow zone with dilated colon during X-ray examination was found, and rectoanal reflex was absent we could definitely determine that the Hirschsprung’s disease was present. In our study, according to the manometry the negative reaction of the internal sphincter was registered in 47 (61.8%) out of 76 patients.

In the rest 29 (38.2%) patients that reaction was significantly weaker. This data was not adequate to confirm the final diagnosis of Hirschsprung’s disease. In such cases we performed the additional diagnostic test – the acetylcholinesterase histochemistry. Given the usual characteristics of the Hirschsprung’s disease in the anamnesis and typical X-ray pictures, though the presence of the weak rectoanal reflex, the increased acetylcholinesterase activity allowed us to determine the ultimate diagnosis of the disease in 51 (67.9%) out of 58 patients who underwent those diagnostic procedures.

Thus, we determined the diagnostic criteria of the Hirschsprung’s disease in adults:

a) constipation since early childhood in the anamnesis 
b) presence of the narrow zone in the distal part of the colon with proximal cone-shape dilation in the X-ray pictures 
c) absence of the rectoanal inhibitory reflex 
d) increased acetylcholinesterase activity.

In those cases when on the basis of barium enema, anorectal manometry, histochemical exams the diagnosis of the Hirschsprung’s disease was not made, we carried out the rectal biopsy (Swenson O.).

TABELA 1
THE DISTRIBUTION OF THE PATIENTS ACCORDING TO THEIR GENDER AND AGE

<table>
<thead>
<tr>
<th>sex</th>
<th>Age (years)</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>14-19</td>
<td>20-29</td>
</tr>
<tr>
<td>Male</td>
<td>20</td>
<td>22</td>
</tr>
<tr>
<td>Female</td>
<td>10</td>
<td>10</td>
</tr>
<tr>
<td>Total</td>
<td>30</td>
<td>32</td>
</tr>
</tbody>
</table>

TABLE 2
THE PERIODS OF LIFE IN WHICH THE FIRST CLINICAL SIGNS OF THE DISEASE APPEARED (N=84)

<table>
<thead>
<tr>
<th>Period of life</th>
<th>Age</th>
<th>Patients</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neonatal</td>
<td>0-28 days</td>
<td>18</td>
<td>21.4%</td>
</tr>
<tr>
<td>Early childhood</td>
<td>29 days-3 years</td>
<td>46</td>
<td>54.8%</td>
</tr>
<tr>
<td>Childhood</td>
<td>4-14 years</td>
<td>18</td>
<td>21.4%</td>
</tr>
<tr>
<td>Adult</td>
<td>20-29 years</td>
<td>1</td>
<td>1.2%</td>
</tr>
<tr>
<td></td>
<td>30-39 years</td>
<td>1</td>
<td>1.2%</td>
</tr>
</tbody>
</table>

The full-thickness rectal biopsy were performed in 26 (30.9%) out of 84 patients. In all cases were registered the absence of intrinsic ganglion cells that allowed to diagnose Hirschsprung’s disease.

Taking into account the fact that up to now there is no common view about the length of the physiological hypoganglionic zone and the level of the biopsy taking (Duhamel B., 1956, Hofmann R.F., Venupogal S., 1981), we performed our own research of the density and spread intrinsic ganglia in the distal part of the rectum in adults. Research was based on 35 cadaver specimens of distal part of rectum and anal canal. Exclusion criteria were catamnestic data on diabetes, rectal and anal canal tumors, chronic constipation, megacolon. The age of deceased varied from 30 to 71 (57.2 ± 8.4). There were 21 (60.0%) male and 14 (40.0%) female.

As the result of the study it was found that the length of the physiological hypoganglionic zone in the distal part of the rectum in adults was 24.4 ± 10.9 mm. Its parameters varied from 7.5 to 50 mm. In 4 out of 35 cases the length of the physiological hypoganglionic zone reached 40.0 mm., and in one case – 50.0 mm. The biopsy taken according to the Swenson method would have inevitably led to diagnostic mistakes in those patients.

For more precise diagnosis of the Hirschsprung’s disease and other abnormalities of the intrinsic nervous system of the colon, we worked out the modification of the rectum biopsy. Method includes excision of the full-thick-
ness stripe 60 mm. long proximally to the dentate line. Histological study of the biopsy specimen which includes physiological hypoganglion zone and some part with potentially normal nervous system above it, permitted us to make more complete and objective evaluation of the nervous structure of the rectum.

Thus, as a result of the complex study the supraanal form of the Hirschsprung’s disease was detected in 5 (5,9%) patients, rectal form – in 45 (53,6%), rectosigmoidal form – in 33 (39,3%), and in 1 (1,2%) female patient the total aganglionosis of the colon was found out.

All 84 patients with Hirschsprung’s disease underwent surgical treatment - resection of the aganglionic zone and dilated decompensated segments of the colon.

One-stage radical surgical treatment were performed with stable general condition of the patients, megacolon limited by one or two parts of the colon and when there were no complications. This tactic was applied in 56 (66,7%) out of 84 adult patients with Hirschsprung’s disease. Intensive preoperative therapy was necessarily carried out when there was no effect of enemas, progressed decompensation of the colon with signs of intestinal obstruction, general condition was getting worse due to chronic intoxication and metabolic disorders. The surgical treatment of such patients consisted of several stages. The first stage was elimination of the intestinal obstruction. After restoration of the colon function and improvement in the patient’s general condition, the radical surgical treatment with reconstruction of the colon continuity was carried out.

The multistage treatment was applied in 28 (33,3%) patients with Hirschsprung’s disease. As the result 13 (46,4%) of the patients were cured of the intestinal obstruction caused by coprolite in sigmoid colon and radical surgery was performed.

In 8 (28,6%) patients with expanded megacolon multistage treatment helped reach compensation of proximal parts of the dilated colon and save them for further functioning.

Another 6 (21,4%) patients with expanded megacolon, despite the absence of the colon compensation, helped to eliminate intoxication and improve general condition significantly. At last 1 (3,6%) patient with megileum and total aganglionosis of the colon had successful surgical treatment – subtotal resection of the colon with subsequent restoration of the bowel continuity.

For treatment of the adults with Hirschsprung’s disease a modification of the Duhamel procedure was worked out in the Scientific Centre of Coloproctology (Vorobyov G.I., 1982), which passed the test of time and has been applied in our clinic since 1977. This method of treatment was used in 76 (90,4%) out of 84 patients with aganglionosis. While working out the surgical treatment of the Hirschsprung’s disease in 3 (3,6%) patients the Swenson procedure was performed and in 2 (2,4%) patients – Duhamel procedure with application of the clamps on the colorectal septum was performed. In another 3 (3,6%) cases subtotal resection of the colon with "end to end" anastomosis was carried out.

Early postoperative complications were registered in 20 (23,8%) out of 84 adult patients. One of the complications led to fatal outcome. The conservative treatment of complications was performed in 7 (35,0%) out of 20 patients. The rest 13 (65,0%) people underwent surgical treatment.

Late complications were detected in 12 patients after modified Duhamel operation and in one patient after the Duhamel procedure with using clamps for formation anastomosis. All complications were represented by the colorectal anastomosis stricture. In 7 (53,8%) out of 13 patients stricture resulted from early inflammatory process in the pelvis.

The stable positive effect from the conservative treatment – bougienage of the stricture, was achieved only in 1 (7,7%) out of 13 patients. In the other 12 (92,3%) cases surgical intervention was required to correct this complication.

The functional outcomes of the surgical treatment of the Hirschsprung’s disease within the period of 1 to 25 (5,7 ±5,9) years after the operations were observed on 62 (74,7%) out of 83 patients (one patient died from the progressing peritonitis in the early postoperative period).

<table>
<thead>
<tr>
<th>TABLE 3</th>
<th>THE SURGICAL TREATMENT WHICH PATIENTS UNDERWENT BEFORE THEIR ADMISSION TO THE CENTRE OF COLOPROCTOLOGY</th>
</tr>
</thead>
<tbody>
<tr>
<td>The types of the surgical treatment</td>
<td>Patients</td>
</tr>
<tr>
<td>Resection of the colon with primary anastomosis</td>
<td>12</td>
</tr>
<tr>
<td>Hartman procedure</td>
<td>4</td>
</tr>
<tr>
<td>Colotomy, coprolite removing</td>
<td>3</td>
</tr>
<tr>
<td>Detorsion of the sigmoid colon</td>
<td>2</td>
</tr>
<tr>
<td>Colostomy</td>
<td>5</td>
</tr>
<tr>
<td>Total</td>
<td>26</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>TABLE 4</th>
<th>THE DEPENDENCY BETWEEN THE LONG-TERM OUTCOMES OF MODIFIED DUHAMEL PROCEDURE AND MEGACOLON EXTENTION (N=55)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Result</td>
<td>Megacolon extention 1 part of the colon More than 1 part of the colon</td>
</tr>
<tr>
<td>Good</td>
<td>30 (88.2%) 14 (66.7%)</td>
</tr>
<tr>
<td>Satisfactory</td>
<td>3 (8.8%) 6 (28.5%)</td>
</tr>
<tr>
<td>Unsatisfactory</td>
<td>1 (3.0%) 1 (4.8%)</td>
</tr>
<tr>
<td>Total</td>
<td>34 (100%) 21 (100%)</td>
</tr>
</tbody>
</table>

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Good results were seen in 50 (80.7%) patients, satisfactory — in 10 (16.1%) and unsatisfactory — in 2 (3.2%) cases.

Most of the adult patients with Hirschsprung’s disease — 76 (90.4%) out of 84 underwent the modified Duhamel procedure. To reveal statistical significance and to make conclusions we analyzed factors influencing long-term results in this particular group because it was the most representative one.

The long-term outcomes of treatment were observed in 55 (72.4%) out of 76 patients who underwent the modified Duhamel procedure. Good results were achieved in 44 (80.0%) patients, satisfactory — in 9 (16.4%), unsatisfactory results were seen in 2 (3.6%) patients.

The study of correlation between the long-term outcomes after surgical treatment of the Hirschsprung’s disease and the length of the aganglionic segment and megacolon extension, has been carried out. Analysis of all the data compared gave us statistically proven dependency between the long-term outcomes and megacolon extension (p <0.05) (table 4).

The dependency between the long-term outcomes of modified Duhamel procedure and megacolon extension (n = 55).

Thus, megacolon extension limited by the sigmoid colon gave good result in 88.2% cases, while in case of the megacolon extension to the proximal segments the value of good results went down to 66.7%. Other results were statistically insignificant.

CONCLUSION

1. The first clinical signs of the Hirschsprung’s disease in adult patients appear in neonatal period or in early childhood in 97.6% of cases. However, due to the latent type of the disease the diagnosis at that age is made only in 19.0% of cases. Due to the progressing of complications of the Hirschsprung’s disease the necessity for urgent surgical treatment arises in 30.9% of cases.

2. Diagnostic criteria of the Hirschsprung’s disease in adults are:
   a) constipation since early childhood in the anamnesis
   b) presence of the narrow zone in the distal part of the colon with proximal cone-shape dilation in the X-ray pictures
   c) absence of the rectoanal inhibitory reflex
   d) increased acetylcholinesterase activity
   e) absence of intrinsic ganglia according to morphological study.

3. The length of the physiological hypoganglionic zone in adults varies between 7.5 and 50.0 mm., making up on average 24.4 ± 10.9 mm. which required the modified rectal biopsy.

4. One-stage surgical treatment of the Hirschspring’s disease in adults is justified when the general condition of the patient is stable, megacolon is limited, and there are no complications of the disease. If megacolon is extended or disease is complicated, the surgical treatment should be divided into several stages. Such tactic allows us to achieve favorable results in 96.4% of cases.

5. The long-term outcomes of the surgery of the Hirschsprung’s disease depend on extension of the megacolon. In case only the sigmoid colon is dilated, good results could be achieved in 88.2% of cases. If dilation extends to the proximal parts of the colon this value reduces to 66.7% (p< 0.05, statistically significant).

SUMMARY

Hiršprungova bolest je malformacija debelog creva koja je uzrokovana nedostatkom intramuralnih gangliona u submukoznom i mijenteričnom pleksusu i manifestuje se megakolonom. Kongenitalne karakteristike je svrstavaju u dečje bolesti i zahvaljujući progresu pedijatrijske dijagnostike sada se uspešno leć.

Ali ova bolest može biti prezentovana sa prominentnim kliničkim manifestacijama kada opstipacija ima periodični i nestabilni karakter i lako se rešava klizmama. Sve ovo dozvoljava ovim bolesnicima da dožive starost. Obično je njihova prva poseta lekaru u slučaju funkcionalne dekompencije kolona koja zahteva hitnu hiruršku intervenciju.

Key words: Hiršpringova bolest, odrasli, tretman