I. RADIOLOGY

HIRSCHSPRUNG DISEASE – THE CONTRIBUTION OF THE RADIODIAGNOSTIC CASE REPORT OF ONE RARE VARIANT WITH COMPLICATIONS

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Abstract

The following describes the case of a female suckling who,at 6 weeks of life, has been hospitalized at the

Infant Surgery Clinics within "Louis Turcanu" Emergency Hospital from Timisoara.

Both her anamnesis and clinical aspect determined the clinician (surgeon) to demand a series of investigations such as abdominal radiological exams which showed modifications (resulted from the presence of some hydroaeric levels).

The radiological exam with contrast substance confirmed our suppositions regarding the diagnostic of Hirschsprung Disease (HD). Moreover, a thorough analysis of the patient's radiological aspect determined the radiologist to diagnose her with a rare form of Hischsprung Disease, more precisely the ultra – short segment HD.

Infectious complications as enterocolitis and septicemia, forced the clinician (surgeon) to resort a two times surgical intervention.

Key words: Hirschsprung Disease (HD), congenital aganglionosis, water-soluble contrast enema (barium enema), ultra - short segment HD.

Introduction

Harold Hirschsprung published the classic description of the congenital megacolon in 1886. Hirschsprung Disease (HD) is caracterised by the absence of the myenteric and submucosal ganglion cells in the distal digestive tract. The disease result in decreased motility in the affected bowel segment (5). This causes a blockage. Intestinal contents build up behind the blockage, causing the bowel and abdomen to become swollen (8). Hirschsprung Disease causes about 25% of all newborn intestinal obstruction (8).

HD is regarded as a neurocristopathy because it involves a premature arrest of the craniocaudal migration of vagal neural crest cells in the hindgut at weeks 5-12 of gestation to form the enteric nervous system (5). The aganglionic, aperistaltic bowel segment effectively prevent the propulsion of the fecal stream, resulting in dilatation and hypertrophy of the normal proximal colon (5).

Hirschsprung Disease (congenital aganglionosis) is caused by a single gene mutation of the RET proto-oncogene on band 10q 11.2 (3). As a congenital disorder, HD is manifested mostly in the first several weeks of life (5).

HD is estimated to occur at a rate of 1 case per 5000 live births (5). Males are affected more often than females, with a ratio of 4:1 (5).

HD is sometimes associated with other inherited or congenital conditions such as Down syndrome (2,5).
Tests used to help diagnose HD may include: abdominal x-ray, anal manometry, barium enema and rectal biopsy.

Anamnesis and clinical findings

Female suckling with habitual constipation is hospitalized at 6 (six) weeks at Arad County Hospital between May 21, 2008 and May 23, 2008. The patient is hospitalized with the following diagnoses: hemolytic anemia (infectious?), post-transfusion reaction (hemoglobinuria, hematuria, icterus) overlapped with the initial anemia, invasive acute enterocolitis, dynamic ileus.

The patient is transferred at the Universitary and Emergency Hospital for Children from Timisoara where initially is hospitalized in the Clinic no.3 Pediatrics. On May 23, 2008 the clinician observes: general state profoundly altered, no fever, intens pale teguments with sclerotic and tegument icterus. Between May 24, 2008 and May 28, 2008 the clinician observes and notes the state of the abdomen: distended, flat and relaxed, discret distended, distended, distended. She is then transferred at the Surgery Clinic where repeated abdominal radiographic

examinations revealing modifications, the barium enema is recommended. On June 2, 2008 this examination was performed and the radiologist has offered the diagnostic – congenital megacolon; on June 11, 2008 the patient has undergone a operation when the aganglionic segment was resected and the sigmoid colostomy was performed. The patient was discharged with the pathology (congenital megacolon, acute enterocolitis, severe sepsis with enteroccocus, severe secondary anemia) removed.

Paraclinic findings

A. Laboratory tests:

a. Blood cells count:

Date:	Arad,	Timisoara	29.05.2008,	08.06.2008
Hemoglobin g%	4,6		6,2	
Erytrocytes/mm3	1.690.000		2.080.000	4.310.000
Leucocytes/mm3			24.400	7.230

b. Serum electrolytes level:

Date:	Timisoara	29.05.2008,	03.06.2008
Na mmol/l		129	128
K mmol/l		4,3	4,5
Ca mmol/l		1,23	1,24
Cl mmol/l		93	92

c. Cultures: blood cultures

Date:	Timisoara	02.06.2008
		enterococcus

B. Radiological aspects:

a. Plain abdominal radiography – Dates: 24.05.2008; 27.05.2008; 28.05.2008

Marked dilatation of the bowel (small and large bowel).

Air-fluid level. No gas in the pelvis (rectum) (fig. 1).

b. Barium enema - Date: 02.06.2008

Reduced caliber of the terminal rectum (with a tubular aspect: 43mm/length and 12 – 16 mm/width) followed by a transition zone to an enlarged caliber of the proximal rectum and sigmoid. Dolicosigma (fig. 2).

C. Histopathological exam – Date: 16.06.2008

Congenital megacolon.

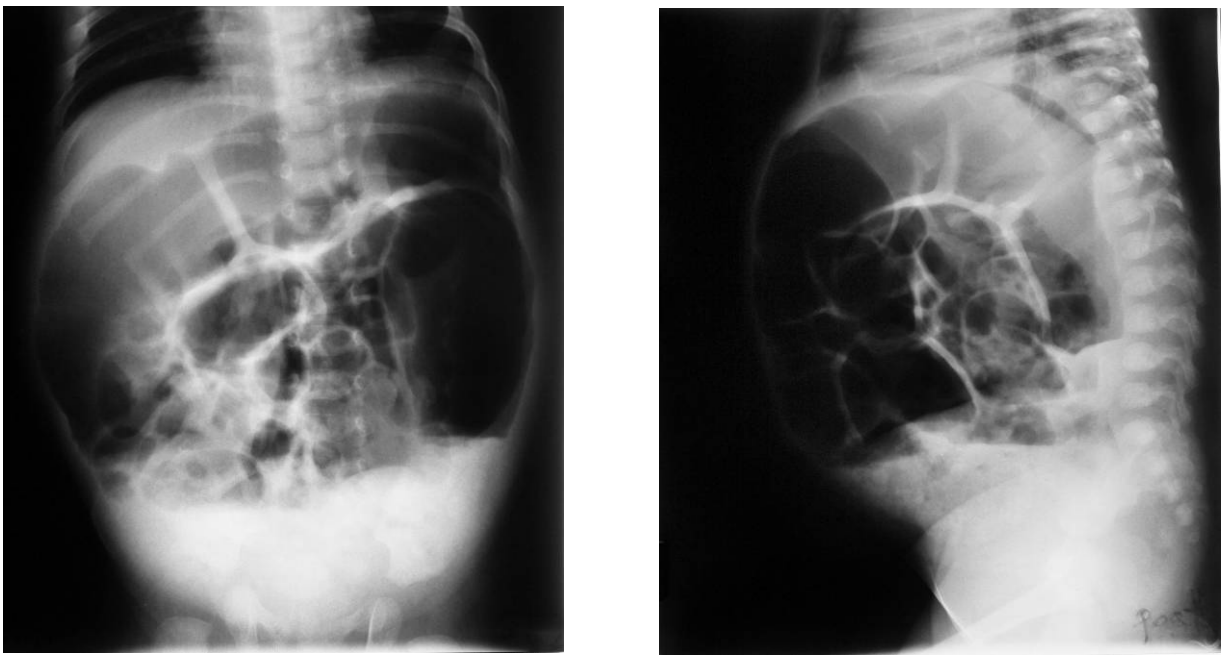

Fig. 1. Marked dilatation of the bowel (small and large bowel).
Air-fluid level. No gas in the pelvis (rectum).

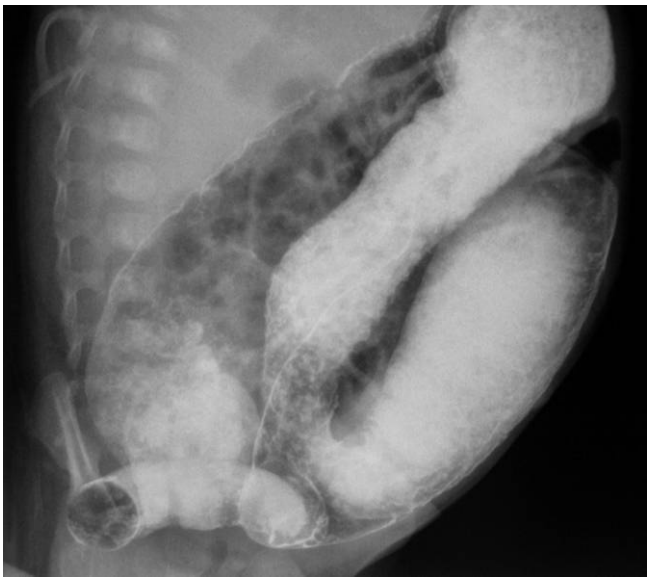

Fig. 2. Reduced caliber of the terminal rectum (with a tubular aspect: 43mm/length and 12 – 16 mm/width) followed by a transition zone to an enlarged caliber of the proximal rectum and sigmoid. Dolicosigma.

Discussions

Normally, as a baby grows in the womb, bundles of nerve cells (ganglia) begin to form between the muscle layers along the length of the colon. This process begins at the top of the colon and ends at the bottom (rectum). In children with Hirschsprung Disease, this process does not finish and the ganglia do not form along the entire length of the colon. Other times a longer portion may be affected (7).

Aganglionosis begins with the anus, wich is always involved, and continues proximally for a variable distance (2). The precise mechanism underlying the development of Hirschsprung disease is unknown (2).

Hirschsprung Disease (HD) can be classified by the extension of the aganglionosis as follow (5):

1.Classical HD (75% of cases): The aganglionic segment does not extend beyond the upper sigmoid.

2. Long segment HD (20% of cases).

3. Total colonic aganglionosis (3-12% of cases).

Some rare variants include the following:

4. Total intestinal aganglionosis.

5. Ultra – short segment HD.

Ultra – short segment HD is caracterised by a few centimeters of aganglionic bowel in the rectum, adjacent to the anus (2).

Water-soluble contrast enema is the key study for diagnosis (6). Lateral views of early rectal/sigmoid colon filling are the most important images. The normal rectum should always be equal or of larger caliber than the sigmoid colon: the rectum/sigmoid ratio. In HD rectum/sigmoid ratio is reversed (6).

About 10% children may present with diarrea caused by enterocolitis, wich is thought to be related to stasis and bacterial overgrowth. This may progress to colonic perforation, causing life threatening sepsis(2).

The case in discussion can be classifid as a rare form of HD, more specifically, the ultra – short HD, with a 4-5 cm length stenotic segment (emphasized by water soluble contrast enema), affecting not only the anal channel (1) but also the distal part of the rectum. The rectum/sigmoid ratio has been reversed.

Both the enterocolitis and septicemia, proved through positive hemoculture analyses, confirmed the fact that this case can be considered as belonging to the rare group of 10% cases - of HD – diagnosed with infectious complications (2,4).

Under these circumstances and taking into account the type of disease (HD associated with septicemia infection), the two times therapeutic surgical intervention was selected.

Conclussions

1. In Hirschsprung Disease (HD), water soluble contrast enema is the key for the diagnostic. The rectum/sigmoid ratio is reversed.

2. The case into discussion can be classified as a rare form of HD, ultra – short HD.

3. The infectious complications have determined the surgeon to choose a two times surgical intervention.

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