International Journal of Health Sciences and Research
www.ijhsr.org ISSN: 2249-9571

Case Report

Barium Enema Proving to Be a Better Tool for Diagnosing Hirschsprung's Disease: a Case Report

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Received: 29/06/2015 *Revised: 20/07/2015* *Accepted: 22/07/2015*

ABSTRACT

Hirschsprung disease is a neurocristopathy; congenital abnormality occurring due to premature arrest of the craniocaudal migration of vagal neural crest cells in hindgut between 4th to 12th weeks of gestation to form the enteric nervous system. There is aganglionosis, leading to aperistaltic bowel segment effectively preventing the propulsion of fecal stream. The diagnosis and the treatment protocols are decided on the bases of the extension of aganglionosis and the segment involved. The incidence is documented as 1 in 5000 live births with male preponderance, ratio of 4:1. The aganglionosis in HD results from the failure of cells derived from the neural crest to populate the embryonic colon during development. The fundamental defect in the bowel microenvironment prevents the ingrowth of neuroblast.

The patient presents with wide range of clinical symptoms, from wide range of obstipation just after birth in association with incomplete evacuation, leading eventually to distended abdomen, recurrent constipation, and high diaphragm.

Better diagnosis and improvements in surgical techniques have contributed to decrease the mortality of individuals with HD. The greatest morbidity and mortality is observed in children younger than 1 year, owing to the possible set of HAEC, with a mean incidence of 25%, which can be fatal if not diagnosed and treated rapidly. Till current documented data a radiological study alone is not sensitive enough to diagnose HD. Rectal mucosal biopsy is required for an accurate diagnosis. This case report enlightens to rule out the need of rectal biopsy on eliciting the signs on barium enema.

Keywords: Hirschsprung's disease, Aganglionosis, Bowel loops, Barium enema, congenital disorder, Water soluble contrast.

INTRODUCTION

Hirschsprung disease (HD) is a congenital disorder causing colonic obstruction due to absence of ganglionic cells in the distal alimentary canal. [1] There has been evidence of polygenic and varied penetrance genetic condition as a risk factor.

In this clinical entity the children presenting with abdominal distension, explosive diarrhea, vomiting, fever, lethargy, rectal bleeding, and shock may possibly have developed HAEC which can be prevented if diagnosed at early stages. HD presents as an isolated trait in 70%

cases; but can be seen in association with chromosomal abnormality (>90% trisomy 21) in 12% patients and other additional congenital anomalies in 18% cases. [2]

The purpose of the study is to perform the fluoroscopic barium enema for the patient and study the various signs elicited that can prove its utility and emphasis on early diagnosis.

Aims and Objectives

- To formulate an appropriate diagnostic approach and prove its significance by means of eliciting the signs relating Hirschsprung disease (HD) by means of administration of water soluble contrast on fluoroscopic study.
- To prove the eligibility and importance of barium enema as better / non-invasive diagnostic procedure over tissue biopsy in a case of congenital disorder.

This study is ought to determine its significance, by proving the validity of major signs related to HD which have been elicited on BE in a patient who has been clinically diagnosed; suffering from intestinal obstruction and comparing the results of rectal biopsy where post-surgical histopathological correlation was evident.

MATERIALS AND METHODS

Barium Sulphate microbar oral suspension, contrast was instilled per-rectally and Barium Enema was performed under fluoroscopic guidance on Siemens Hellophes D -500ma x-ray with Flurovision.

CASE REPORT

A one year old male child was brought to the radiology department with complaints of tense and distended abdomen with difficulty in passing stools. The complaints were present since birth and were progressive in nature. The other associated complaints were of poor feeding and history of failure to pass meconium in first 24 hours of life.

The patient underwent plain x-ray abdomen standing and BARIUM ENEMA under fluoroscopic guidance. Plain radiograph revealed multiple dilated small bowel loops with air-fluid levels suggesting the distal bowel obstruction, with an empty rectum sign (ERS). On performing barium enema the proximally dilated colonic loop with short segmental narrowing was noted suggesting transition zone (TZ) involving recto-sigmoid junction. Delayed film (48 hours) showed distended colon with residual barium-stool pattern with persistence of barium in colon, suggestive of delayed evacuation. The RSI was measured to be 0.49 as per the measurements stated in fig. 1(b) and 2(b).

Pre-operative measures were taken to stabilize the patient before surgical therapy by fluid and electrolyte imbalance correction. Definitive surgical treatment comprised resection of aganglionic segment followed by a pull-through of ganglionic bowel down to **anus-Duhamel pull-through (recto-rectal).**

Post-operatively the full thickness biopsy specimen was taken above the anal valves.

Rectal biopsy revealed absence of ganglionic cells, hypertrophy/ hyperplasia of nerve fibers and increased acetylcholinesterase positive nerve fibers in lamina propria and muscularis mucosa.

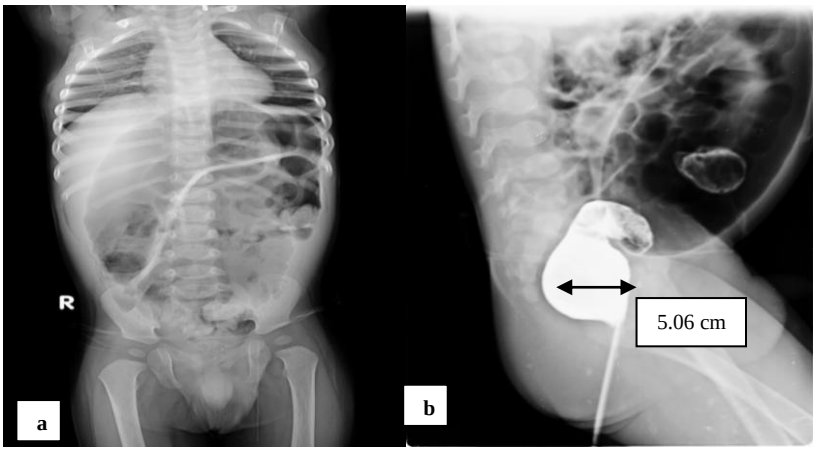

Fig. 1-

(a) Plain X-ray abdomen showing dilated bowel loops, multiple air-fluid levels and empty rectum sign (ERS).
(b) Barium filled rectum with maximum diameter of rectum (5.06cm).

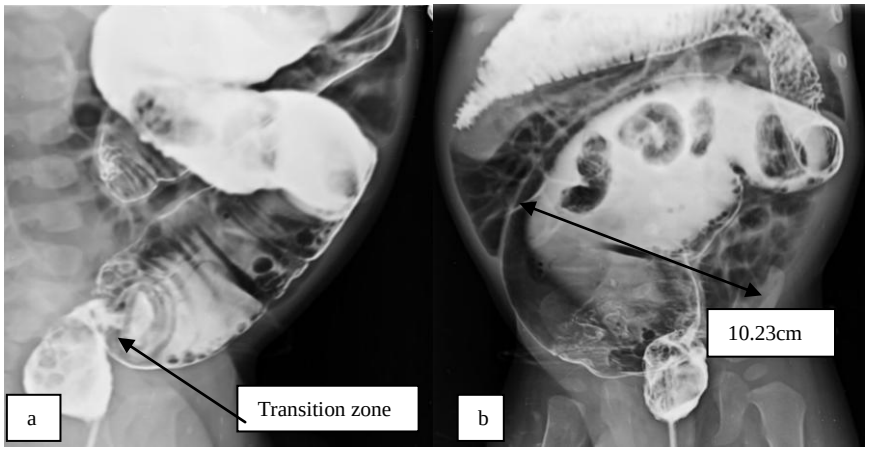

Fig. 2-

(a) Shows occurrence of transition zone at the site of recto-sigmoid junction.
(b) Grossly distended proximal colon with greatest diameter of the sigmoid colon (10.23cm) as demonstrated by the arrow.

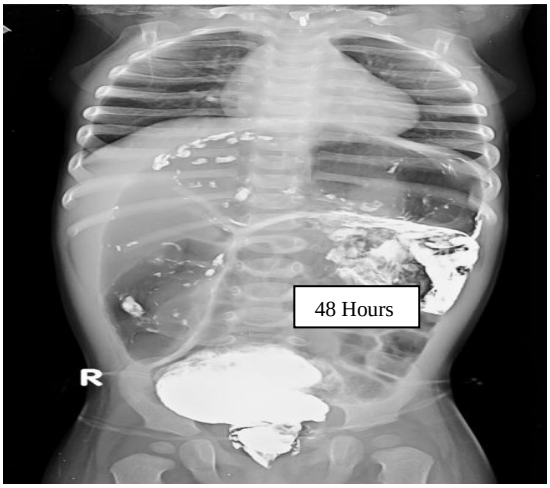

Fig. 3:

- 48 hours delayed film demonstrates the residual barium-stool patterns with dilated bowel loops and persistence of barium in colon s/o delayed emptying.

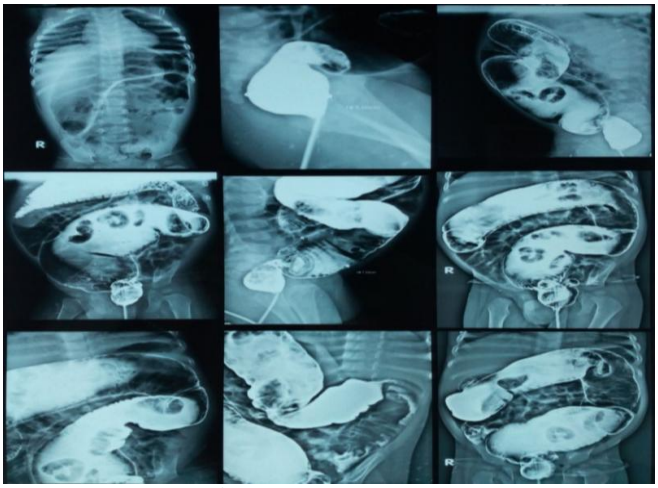

Fig.4 : Complete study of barium enema (BE) and sequential images as per the procedure performed.

TABLE 1

Signs of HD after BARIUM ENEMA administration include the following-

- Transition zone (TZ).
- Abnormal, irregular contraction of aganglionic segment (rare).
- Delayed evacuation of barium.
- Thickening and nodularity of colonic mucosa proximal to transition zone.
- Mixed barium-stool pattern on delayed radiographic film.
- Distended bowel loops on plain radiographs that almost fill after contrast enema.
- Question mark "?" - shaped colon in total colonic aganglionosis.
- Cobblestone mucosal pattern.
- Serrations.
- Spastic colonic appearance.
- Recto-sigmoid index (RSI) <1 for short segment.

TABLE 2

The anatomical division of aganglionic segment according to involved length-

- **Short segment: ~ 75%**
  Rectal and distal sigmoid colon involvement only.
- **Long segment: ~ 15%**
  Typically to splenic flexure or transverse colon.
- **Total colonic aganglionosis: ~ 2-3 %**
  Also known as Zuezler – Wilson Syndrome. Occasional extension of aganglionosis into small bowel.
- **Ultrashort segment disease:**
  Involvement of 3-4 cm of internal anal sphincter only (Controversial entity).

Table 3: Presenting symptoms in case of HD

INFANTS	OLDER CHILDREN
Bilious vomiting	Absence of soiling or overflow incontinence
Enterocolitis associated diarrhea	Failure to thrive
Failure to pass meconium in first 24 hours of life	Fecal impaction
Infrequent, explosive bowel movements; difficult bowel movements	Malnutrition
Jaundice	Progressive abdominal distension
Poor feeding	Chronic progressive constipation, usually with onset in infancy.
Progressive abdominal distension	
Tight anal sphincter with an empty rectum	

TABLE 4: Scoring system based on radiologic signs of HD

SCORE	% PROBABILITY OF HD	RATING
6-8	100	High
4-5	66	Moderate
1-3	40	Low

DISCUSSION

Hirschsprung disease (HD) features failure of ganglionic cells migration cephalocaudally during embryological development, leading to absence of ganglionic cells in whole or part of colon. [3]

The aganglionosis is due to failure of cells derived from the neural crest to populate the embryonic colon during the development. This failure is from fundamental defect in the micro-environment of the bowel wall that prevents ingrowth of neuroblasts.

So far, 11 genetic defects are known to be associated with HD, including mutations to the Endothelin-β receptor gene and tyrosin kinase RET gene, the latter being responsible for a major role in all forms of HD susceptibility. [4] The major gene responsible for HD, identified in the chromosal 10 region, was the RET proto-oncogene.

Associated Syndromes with HD:

- Down's syndrome
- Multiple endocrine neoplasia type 2 (MEN 2)
- Cat Eye Syndrome
- Waardenburg Syndrome
- Bardet-Biedl Syndrome

The incidence of HD has been documented to be approximately 1 case in 5000 to 1 in 10000 live births. There appears a male preponderance with ratio of 4:1 to 5:1 particularly in short segments.

However for short segment disease, the male-to-female ratio is 4.2:4.4 and for long-segment disease the female-to-male ratio is 1.2:1.9

Cases Diagnosed;

- 15% in first month of life.
- 45-50% in first 3 months.
- 60% in end of first year of age.
- 85%by 4 years of age.

HD is often termed as congenital Aganglionic Megacolon, accounting for 15-20% of all intestinal obstructions in the neonate. [5] The segment is usually seen as begin at the anus extending proximally to the colon. [6] Most patients present in infancy, and early diagnosis is important to avoid further complications.

HD can also be noted in association with cardiovascular, urologic, neurologic and gastrointestinal abnormalities.

Here the ganglionic cells are absent in both, the sub-mucosal (Meissner's plexus) and intermuscular (Aurbach's plexus) layer. [7]

Differential Diagnosis;

- Intestinal neuronal dysplasia
- Meconium ileus
- Pediatric constipation
- Functional Megarectum
- Necrotizing Enterocolitis
- Microcolon

The first reporting of a patient with HD was made in 1691 by Fredrick Ruysch, but it was Danish pediatrician Harald Hirschsprung who in 1888 published the classic description of congenital megacolon. [8]

DIAGNOSTIC APPROACH:

Diagnostic techniques involve anorectal manometry, Barium enema (BE) and rectal biopsy.

Definitive diagnosis of HD is made by suction biopsy of the distally narrowed segment. [9] A histologic examination of tissue would show lack of ganglionic nerve cells.

Suction rectal biopsy is currently the gold standard for diagnosing HD.

HD is more definitely diagnosed by means of contrast enema examination, which can show the presence of transition zone, irregular contractions, mucosal irregularity and delayed evacuation of contrast material. Colonic perforation is not uncommon in HD, being reported in 3-4% of affected patients. [10, 11] The risk of perforation in the neonatal population justifies the use of low-osmolality contrast agents used. The margin of safety is more when low-osmolality contrast agents are used.

Sensitivity & specificity of a contrast enema in diagnosis of HD are reported as being 76% and 97%, respectively.

Plain abdominal radiography

Contrast enema examination of colon.
Occasionally assisted by USG findings to suggest the diagnosis

Rectal suction biopsy or full thickness rectal biopsy.

Radiographic imaging features:

Radiographic images show multiple loops of dilated small bowel with air-fluid levels that can be determined as distal bowel obstruction.

- **Empty Rectum Sign (ERS)**: dilatation of small bowel with no gas in rectum. Classic image is that of a dilated proximal colon with the aganglionic cone narrowing towards distal gut.
- **Transition Zone (TZ)**: is the term applied to the region in which a marked change in the caliber occurs with the dilated, normal colon above and the narrowed, aganglionic part below; although this is a highly reliable sign of HD. Hallmark of diagnosis is demonstration of transition zone from dilated bowel to the reduced caliber bowel.

- **Recto-Sigmoid Index (RSI):** is the ratio of the widest diameter of the rectum to the widest diameter of sigmoid colon.

$$RSI = \frac{\text{Widest diameter of rectum}}{\text{Widest diameter of the sigmoid colon}}$$

RSI can help diagnosing the HD in cases of inability to visualize transition zone.

RSI <1 is diagnostic for HD.

The scoring system as stated below in table is simple. When a sign is present, the study radiologist records a score of 1; when the sign is absent, the score is 0. The maximum score of a patient can be 8. The scoring system provides a means of attaching a level of probability to the reporting that should benefit the radiologist and the physician. [12]

Treatment:

- Pre-operative therapy by fluid and electrolyte imbalance correction, antibiotics in case of enterocolitis followed by rectal decompression with use of rectal irrigations and rectal tubes until time of surgery.
- Definitive surgical therapy comprises resection of aganglionic segment followed by a pull-through of ganglionic bowel down to anus.

Various surgical procedures used in treating the case are-

- Swenson pull-through (recto-sigmoidectomy)
- Duhamel pull-through (recto-rectal)
- Soave pull-through
- Trans-anal pull through
- Endo-rectal pull through

Post surgical complications:

- Obstructive symptoms
- Fecal incontinence
- Enterocolitis.

CONCLUSION

Even in recent times of advances in diagnostics, Barium enema a water soluble contrast based procedure is still a useful and definitive diagnostic tool for cases of Hirschsprung Disease (HD).

Rectal biopsy a gold standard & invasive method of diagnosis with 100% specificity can be ruled out if fluoroscopic Barium Enema (BE) is done in the right manner with demonstration of various suggestive radiological findings.

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ABBREVIATIONS-

HD	=	Hirschsprung Disease
BE	=	Barium Enema
AM	=	Aganglionic Megacolon
TZ	=	Transition Zone
ERS	=	Empty Rectum Sign
RSI	=	Recto-Sigmoid Index
WSCE	=	Water Soluble Contrast Enema
HAEC	=	Hirschsprung-Associated Enterocolitis

How to cite this article: Jagdale A, Malhotra R. Barium enema proving to be a better tool for diagnosing Hirschsprung's disease: a case report. Int J Health Sci Res. 2015; 5(8):708-714.

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