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Accuracy of contrast enema for the diagnosis of hirschsprung disease in tertiary medical centre

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Abstract
To compare radiological findings with the histological diagnosis of Hirschsprung disease (HD) to establish the usefulness of contrast enema as an initial screening and diagnostic tool and role of scoring system.
Material and Methods: Retrospective study of patients aged 0–6 years, with clinically suspected HD, for whom both contrast enemas and rectal biopsies were performed between 01 January 2018 and 31 August 2020 in a tertiary-level hospital. A total of 10 such patients were identified. Diagnostic accuracy levels were calculated by comparing radiological results with histology results, which is the gold standard.
Results: Diagnostic accuracy of contrast enema was 90%.
Conclusion: Contrast enema remains useful as an initial screening and diagnostic test for HD. Results of our hospital were consistent with the best international results for sensitivity of the contrast enema (approximately 90% in excluding the disease).

Keywords: diagnosis, hirschsprung disease, tertiary medical centre

Introduction
Hirschsprung disease (HD) is a functional obstruction of bowel caused by the lack of distal enteric ganglion cells and also known as colonic aganglionosis, is a rare cause of constipation with a reported incidence of 1:5000–7200 in newborns [1]. There has been considerable debate about the most appropriate initial test to diagnose HD because all the diagnostic modalities contrast enema, ano-rectal manometry (ARM) and rectal suction biopsy will have false-negative and false-positive results [2]. Full-thickness biopsy confirming the absence of ganglion cells remains the gold standard for diagnosis [3, 4]. HD may affect variable length and segments of large bowel and will have significant morbidity and mortality if not diagnosed and treated early. The contrast enema is often readily available and is the first imaging procedure, due to the risks associated with rectal biopsy, including bleeding, sepsis, perforation and anaesthesia-related risks [5], led to the development of the rectal suction biopsy as an alternative diagnostic modality.

Methods
A retrospective study was undertaken at Sapthagiri institute of medical sciences and research centre, of all contrast enemas and rectal biopsies performed on patients aged 0–6 years between 01 January 2018 and 31 August 2020 inclusive. Only children investigated with both contrast enema and full-thickness rectal biopsy for the clinical suspicion of HD were included in the study.
10 patients who underwent plain X-ray abdomen, USG, iodinated water soluble contrast enema and full thickness rectal biopsy were studied, positive enema results were based mainly on the presence of a transition zone (TZ) or caliber change, cobble stone appearance, reversed recto-sigmoid ratio (RRSR) and other ancillary findings such as delayed post-contrast evacuation of bowel, saw-tooth mucosal pattern or mucosal irregularity.
Full-thickness rectal biopsies were performed under general anesthesia by pediatric surgeon at 15 mm above the dentate line, posteriorly, and sent for haematoxylin and eosin (H&E) staining Specimens were examined by a specialist histopathologist of our hospital, who looked for the presence or absence of ganglion cells in Meissner's submucosal plexus and Auerbach's intermyenteric plexus, at times with the additional finding of neuronal hyperplasia and acetyl choline esterase.
Clinical history of constipation, abdominal distension, vomiting and delayed passage of meconium were recorded for each patient.

Results

Table 1: Demographic and clinical characteristics.

N=10	
Age (range)	**8 days to 6 years**
Gender:	
Male	4
Female	6
Clinical symptoms:	
Abdominal distension	6
Bilious emesis	1
Non-bilious emesis	Nil
Delayed passage of meconium	3
Diarrhea	Nil
Constipation	6
Constitutional symptoms	Nil
Details of risk factors (n)	ASD with left to right shunt
Family history of HD	Nil

Table 2: Contrast Enema Findings in patients with Hirschsprung disease.

CE Findings	Number	%
Transitional zone	9	90
Reversed Recto- sigmoid index	9	90
Irregular contraction	Nil	
Mucosal irregularity with granularity	1	10
Cobblestone appearance	Nil	

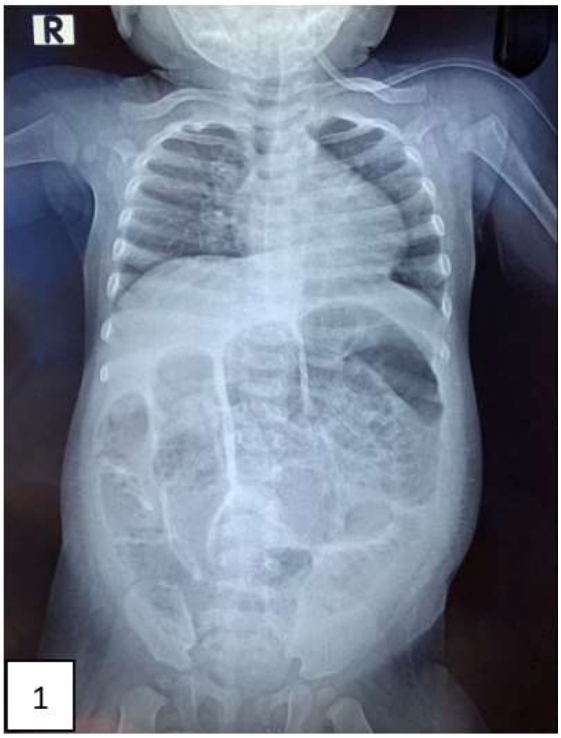

Fig 1: Plain X-ray of abdomen erect showing dilated bowel loops with paucity of gas in the pelvic region.

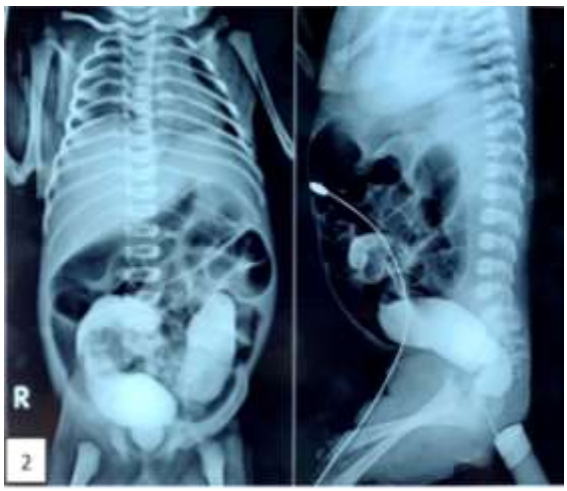

Fig 2: Water soluble contrast enema AP and lateral showing transition zone.

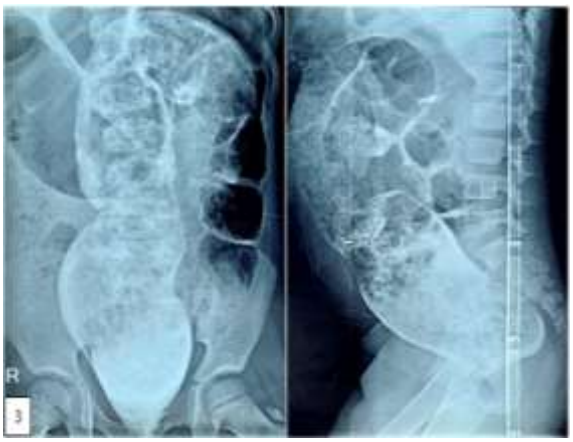

Fig 3: Water soluble contrast enema AP and lateral showing transition zone and RRSR.

Table 3: Details of patients with HD

Sl no	Male	Female	Age	Wt	TZ	RRSR	Rectal biopsy	Procedure	Family history	Associated anomaly
1		F	6 m	5.9kg	Rectum	-	+	Myectomy	-VE	-VE
2	M		1m	3.5kg	Recto-sigmoid	+	+	HSD stage 1	-VE	ASD
3	M		1yr	6.9 kg	Recto-sigmoid	+	-VE	HSD stage II	-VE	-VE
4	M		6yr	15kg	Granulated R-S junction	-VE	-VE	HSD stage 1	-VE	-VE
5	M		3days	4.1kg	Recto-sigmoid	+VE	+VE	HSD stage 1	-VE	-VE
6		F	5 m	4.9 kg	Recto-sigmoid	+VE	+VE	HSD stage ii	-VE	-VE
7		F	7 m	6.2kg	Recto-sigmoid	+VE	-VE	DAMA	-VE	-VE
8		F	18 day	2.7kg	Recto-sigmoid	+VE	+VE	HSD stage ii	-VE	-VE
9		F	8 day	3.7kg	Rectum	+VE		Myectomy + colostomy	-VE	-VE
10	M		10day	3.5kg	Recto-sigmoid	+v	+VE	HSD stage 1	-VE	-VE

Discussion

Intestinal obstruction in the newborn contributes a significant proportion of neonatal admissions in a pediatric surgical unit. The obstruction may be due to a variety of conditions including atresia and stenosis, HD, malrotation, meconium ileus, neoplasia, trauma, and other rare causes. HD causes functional obstruction. Detailed history, thorough physical examination, and well-directed investigation will ensure early diagnosis and prompt treatment of HD.

HD is a developmental disorder of intrinsic enteric nervous system characterized by the absence of ganglion cells in submucosal and myenteric plexus of distal intestine. Ganglion cells are derived from the neural crest. By 13 weeks of gestation, cranial to caudal migration of neural crest cells occurs followed by maturation into ganglion cells. The disruption of either of these, i.e., migration or maturation leads to HD [14].

HD is due to abnormal development of neural crest-derived cells, it may be associated with other neurocristopathies, for example, central hypoventilation syndrome, sensorineural hearing deafness, Waardenburg's syndrome, and MEN Type II B syndrome and Down's syndrome.

The incidence was 1.54 per 5000 live births, female preponderance noted.

In developed countries, most of the patients get diagnosed in neonatal age group, while in developing countries; most of children are diagnosed later [13]. Classical HD was found restricted to recto-sigmoid junction in 75% cases, (similar to our study) long segment disease in 15% cases, ultra short segment disease in 5% cases and variable length was found in 5% of cases [15].

The most common presenting symptoms for HD are delayed passage of meconium (n=3, 30%) abdominal distension (n=6, 60%) and bilious vomiting (n=1, 10%) In older children, the most frequently reported symptom is constipation [1, 11].

Delayed passage of meconium (beyond 24 h) was documented in n=3, child, and while constipation and abdominal distension were reported in patients with the disease[6] (n=6, 60%).

Our study is in concordance with at least three of these findings. Ninety per cent of patients are diagnosed when they are newborns, and the disease is reported to be more common in than females than males [1].

An upright abdominal X-ray was showed features of distal intestinal obstruction (n=10). All patients showed the absence of gas in the pelvis on initial X-ray at the time of presentation [10]. In a neonate with suspected HD, the absence of gas from the pelvis especially on an erect lateral X-ray is helpful in diagnosis [11].

A study by O'Donovan *et al*. assessed the validity of using low osmolality WSCE instead of barium enemas in neonates and infants for the investigation of HD [3]. It found good agreement in the sensitivities and specificities for both groups, which justifies the use of WSCE in neonates and infants. In our study showed TZ at recto-sigmoid junction (n=7, 70%), rectum (n=2, 20%) and one case showed granular appearance at recto-sigmoid junction.

The RRSR and the TZ were the only radiological features that were significantly associated with the HD. The RRSR (reversed recto-sigmoid ratio) was even more significantly associated with the disease than the TZ. An RRSR was present in (n=9, 90%) of children in our study. The TZ was present in (n=9, 90%) of children with the disease [12]. This is consistent with most literature reports which state that the presence of a radiographic TZ on barium enema remains the most accurate diagnostic sign for HD [7].

Although the rectal biopsy is the diagnostic gold standard, the first diagnostic method is CE [8]. The challenging issues of rectal biopsy are higher cost, more invasiveness, longer hospital stay and need to follow-up [9]. The HPE features are increased acetylcholine esterase and absent ganglion cells and Calretinin, a calcium-binding protein, is an important new marker in HD.

Previous studies have shown the TZ and RSI as the most common signs in HD. The sensitivity and specificity of CE in the diagnosis of HD has shown a wide range, between 90% [10].

The overall accuracy of PARTZ and CETZ concordant to the pathological level of aganglionosis was 92% and 72% respectively [8]. Our results though correlate with the study, correlation of contrast enema transition zone and its pathologic location, correlates with our study.

Regarding the diagnostic accuracy of radiologic scoring system, we decided to consider two points for positive results in RSI and TZ and one point for positive results related to lack of meconium defecation, cobblestone appearance, irregular contraction, and level of filling defect, since our statistics is small so our results are not significant.

The anorectal manometry is a useful screening test in the constipated young child.

The surgery for Hirschprung's disease is a staged procedure. In 1st stage, the obstruction is relieved by establishing a loop enterostomy just above the transition zone to allow dilatation and inflammation to resolve and no attempt at resection is made. In 2nd stage, the definitive surgery is performed when child is 10 months old and 10 lbs. in weight. The basic principle of definitive surgery is the removal of poorly functioning aganglionic bowel and an anastomosis of a normally innervated portion of the intestine to the distal rectum. The various definitive procedures are Swenson's-proctocolectomy, the Duhamel procedure –

posterior pull-through with side-to side anastomosis to aganglionic rectum and Soave operation - pull of ganglionated bowel through the sleeve of rectum.
One case went on DAMA and one case died of post-operative sepsis.

Limitations
Limitation of our study is small number of cases.

Conclusion
HD is an important cause of neonatal intestinal obstruction. HD is usually not associated with other congenital anomalies. It has excellent survival in comparison to other causes of neonatal intestinal obstruction. The contrast enema remains useful as a screening test for HD. Results of our tertiary medical care hospital were consistent with the best international results for sensitivity of the contrast enema (approximately 90%) in excluding the disease. However, full-thickness rectal biopsy remains the gold standard for diagnosis of HD.

Abbreviations
1. HD: Hirschsprung's disease
2. CE: Contrast Enema
3. TZ: Transition Zone
4. FTRB: Full Thickness Rectal Biopsy
5. RRSR: Reversed Recto-Sigmoid Ratio
6. HPE: Histo-pathological correlation

Conflict of interest: Nil.
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Authors' Contributions
Dr. B. Mallikarjunappa performed literature review, prepared research proposal protocol for ethics board approval, gathered data for analysis, and prepared the primary write-up of the research manuscript. Dr. Vishwaprem Raj D R and Dr. Naveen D, were the co-investigators of the research project, assisted with data collection for analysis and helped with manuscript editing. Dr. Shivaprasad V R supervised the research project and helped with the manuscript editing.

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