

# Imaging Findings on Contrast Enema of Hirschsprung Disease

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**Objective:** To describe imaging findings on contrast enema of Hirschsprung disease at King Chulalongkorn Memorial Hospital.

**Materials and Methods:** The present study was a retrospective study, approved by the present institute ethic Committee. The requirement for informed consent was waived. Between January 2002 and March 15, 2021, the imaging, medical, and histopathologic information of 48 cases with histopathologically proven Hirschsprung disease were evaluated.

**Results:** Forty cases out of 48 (83.33%) that met the inclusion criteria were male. Neonate and infant patients made up 42 cases. According to histopathologic data, there were diseases in the short segment in 60.42% of the study cases, the long segment in 12.50%, and the entire colon in 14.58%. The common symptoms were abdominal distention, delayed passing meconium, constipation, and poor feeding. The transitional zone in 40 out of 48 (83.33%) was the most often observed imaging finding. Out of the 48 patients, additional imaging findings for 17 (35.41%) was reverse rectosigmoid ratio, for nine (18.75%) was serration, for three (6.25%) was microcolon, and for one (2.08%) was short colon. The transitional zone was also most common among cases of short and long segment diseases. In the cases of total colonic disease, transitional zone and non-diagnostic imaging were equal.

**Conclusion:** The transitional zone, reverse rectosigmoid ratio, and serration are common imaging findings for Hirschsprung disease on contrast enema at King Chulalongkorn Memorial Hospital. Non-diagnostic findings were uncommon but frequently observed in total colonic aganglionosis with variety of patterns. Therefore, if a diagnosis is clinically suspected, a biopsy may be required to confirm it.

**Keywords:** Contrast enema; Hirschsprung disease

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Hirschsprung disease is a functional intestinal obstruction caused by the failure of migration of enteric neural crest-derived cells from foregut to hindgut during the fourth to seventh weeks of gestation<sup>(1)</sup>. It results in absence of parasympathetic Auerbach (intermuscular) and Meissner (submucosal) plexuses, causing aperistalsis in the affected bowel. Most patients present clinical symptoms during infancy<sup>(2)</sup>. The common clinical manifestations in neonates are delayed meconium defecation, abdominal distension, poor feeding, and bilious

vomiting<sup>(3)</sup>. Older children have chronic constipation, abdominal distension, and emesis. Enterocolitis associated with Hirschsprung disease causes mortality rate of 1% to 10%<sup>(4)</sup>.

One out of every five thousand live infants is affected by Hirschsprung disease, which causes 15% to 20% of neonatal intestinal blockage. With a ratio of up to 4:1.8, boys are more likely than girls to get the condition<sup>(2)</sup>.

The disease can be classified as short-segment (75%) of the cases and commonly affects rectosigmoid junction, long-segment colonic (15% to 25%), total colonic (3% to 12%), and total intestinal (rare) Hirschsprung disease<sup>(2)</sup>.

A full-thickness rectal biopsy that demonstrates the lack of ganglion cells is the gold standard for the diagnosis of Hirschsprung disease<sup>(2,3,5)</sup>. While suction rectal biopsy can be performed on patients under the age of six months without requiring general anesthesia and with little risk of bleeding or infection<sup>(5)</sup>, a rectal biopsy carries a number of hazards, including the potential for bleeding, stricture, perforation, scarring,

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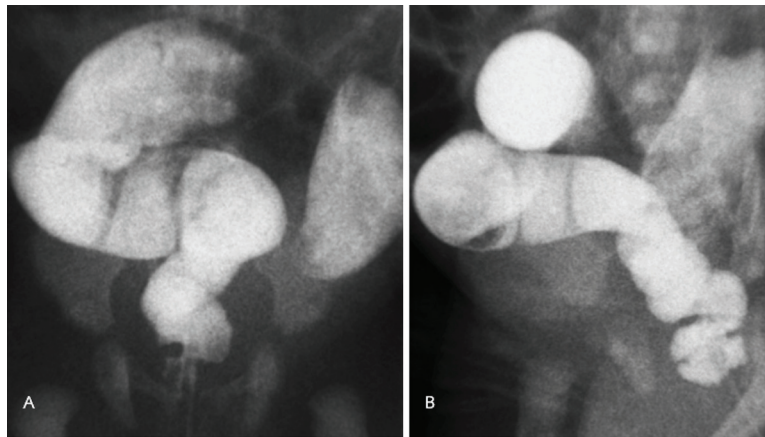
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**Figure 1.** Reverse rectosigmoid ratio. AP (A) and Lateral (B) views from contrast enema show distal sigmoid and rectum to be narrow in caliber. The more proximal sigmoid is wider than the distal sigmoid and rectum with the widest diameter of the rectum divided by the widest diameter of the sigmoid loop less than 1.

and anesthesia-related side effects.

Contrast enema is the imaging of choice for diagnosing Hirschsprung disease<sup>(6-8)</sup>. The type of disease will influence the imaging results. For patients with the short- or long-segment disease, the enema reveals common findings such as an abnormal rectosigmoid ratio of less than 1, or “sawtooth” appearance due to mucosal spasm. In case of short segment disease, a transitional zone is the most sensitive finding<sup>(9,10)</sup>. In cases of total colonic or total intestinal Hirschsprung disease, the enema shows a microcolon and impaired contrast evacuation. Furthermore, rectal manometry may be a useful adjunct study<sup>(2,3)</sup>.

To the best of the author’s knowledge, there has never been any data gathered and published regarding the imaging results of contrast enema in Hirschsprung disease in Thailand. Additionally, the authors discovered that the pre-operative contrast enema results in the present study subjects exhibited atypical findings. Determining the prevalence of non-diagnostic imaging results for cases presenting at King Chulalongkorn Memorial Hospital was the other goal of the present work, which was to report the imaging findings of Hirschsprung disease on contrast enema.

## Materials and Methods

### Study population

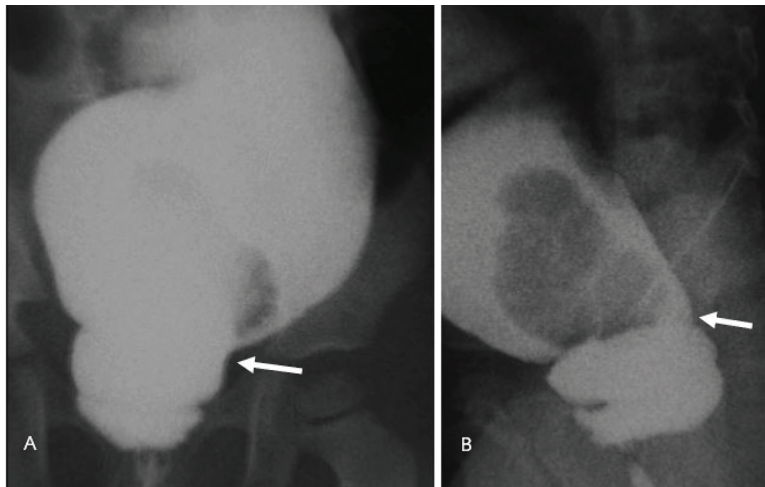
The present study was approved by the Institutional Review Board (COA No. 0154/2022). Between January 2002 and March 2021, the author retrospectively included all patients who had been given a Hirschsprung disease diagnosis using the

ICD-10 code (Q43.1). All of the included patients underwent contrast enema investigation in the present study facility and had their diagnoses verified by pathology. Forty-eight cases met the inclusion criteria.

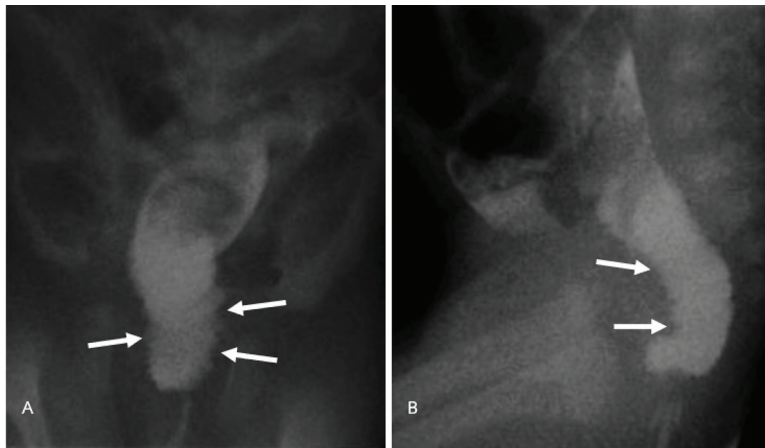
### Data collection

The medical records were used to gather demographic and clinical information, such as gender, age, underlying disorders, and presenting symptoms. The patients were divided into three age groups, the neonates (under a month old), infants (between one month and one year old), and patients older than one year. Water-soluble iodinated contrast or diluted barium enema were used for all contrast enema images. Synapse software was used to examine the pictures on the Picture Archiving and Communication System (PACS) (Version 5, Fujifilm Global, Japan). A pediatric radiologist and radiology resident looked through the imaging results.

Reverse rectosigmoid ratio, transitional zone, serration, microcolon, and short colon were the imaging findings collected. The reverse rectosigmoid was defined as less than one of the widest diameter of the rectum divided by the widest diameter of the sigmoid loop when fully distended by a contrast medium on the lateral view (Figure 1). The normal rectosigmoid ratio is  $\geq 1$ . The transitional zone is defined as the site of obvious caliber change during the retrograde flow of contrast material from non-dilated to dilated bowel (Figure 2). Serration, serrated, or sawtooth appearance is defined as colonic wall irregularity of the aganglionic segment (Figure 3). Microcolon is defined as small-caliber colon, the



**Figure 2.** Transitional zone. AP (A) and Lateral (B) views from contrast enema show the small caliber of the lower rectum and more proximal rectum dilatation with the transitional zone at the lower rectum (arrows).



**Figure 3.** Serration. AP (A) and Lateral (B) views from contrast enema show mucosal irregularity of narrowed rectum with serrated or sawtooth appearance (arrows). Note transitional zone at proximal sigmoid.

largest diameter less than 1 cm or less than the interpedicular space of the L1 vertebra when the colon was fully distended by contrast medium (Figure 4). Short colon is defined as a shorter than the normal colon, usually seen as “question mark sign” (Figure 5).

### Statistical analysis

Statistical analyses were performed in Microsoft Excel (version 16.0). Contrast enema imaging abnormalities were reported as a proportion.

## Results

### Demographic, clinical, and pathologic data

One hundred eight cases had pathologically proved disease. Forty-eight cases out of 108 cases

underwent a contrast enema study and had imaging reports. Thus, forty-eight cases met the inclusion criteria. In the 48 cases, there were 40 males (83.33%) and eight females (16.67%). Twenty-three cases (47.92%) were neonates, 19 cases (39.58%) were infants and six cases (12.50%) were patients, more than one year old. Four cases (8.33%) had Down syndrome.

According to histopathological data, there were 29 short segment disease (60.42%), six long segment disease (12.50%), and seven total colonic segment disease (14.58%). No total intestinal disease was found. In six cases, the pathological reports did not document the involvement of the aganglionic colonic segment.

The most common presentation was abdominal



**Figure 4.** Microcolon. AP view from contrast enema shows the redundant and small caliber of the colon, less than interpedicular space of L1 vertebra.



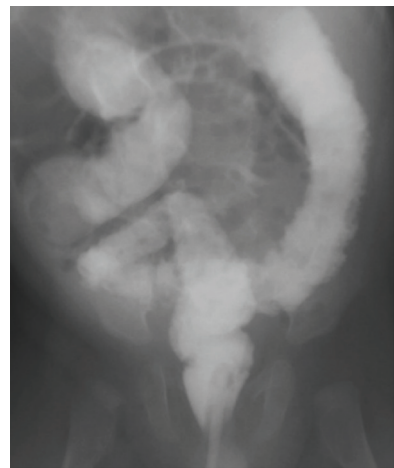
**Figure 5.** Short colon and ileocecal reflux. AP view from contrast enema shows shortening of the colon and also ileocecal reflux. Note the small caliber of the rectum and colon.

**Table 1.** Demographic data and histopathology

Characteristics	Value; n (%)
Patient	48
<b>Age</b>	
Neonate	23 (47.92)
Infant	19 (39.58)
>1 year	6 (12.50)
<b>Male</b>	
Down syndrome	4 (8.33)
<b>Clinical presentation</b>	
Abdominal distention	25 (52.08)
Delayed passing meconium	11 (22.92)
Constipation	9 (18.75)
Poor feeding	7 (14.58)
Bilious vomiting	3 (6.25)
Difficult defecation	2 (4.17)
Others	2 (4.17)
<b>Histopathology</b>	
Short segment	29 (60.42)
Long segment	6 (12.50)
Total colonic aganglionosis	7 (14.58)
No data	6 (12.50)

distention in 25 (52.08%). Other presentations were delayed passing meconium in 11 (22.92%), constipation in nine (18.75%), poor feeding in seven (14.58%), bilious vomiting in three (6.25%), difficult defecation in two (4.17%), and other clinical gut obstruction in two (4.17%).

All demographic data and histopathology are displayed in Table 1.



**Figure 6.** A 3-month-old boy with total colonic aganglionosis presented with abdominal distention and constipation. AP view from contrast enema shows persistent narrowing of the sigmoid without proximal bowel dilatation. Noted mucosal irregularity and bowel wall thickening of the sigmoid and descending colon.

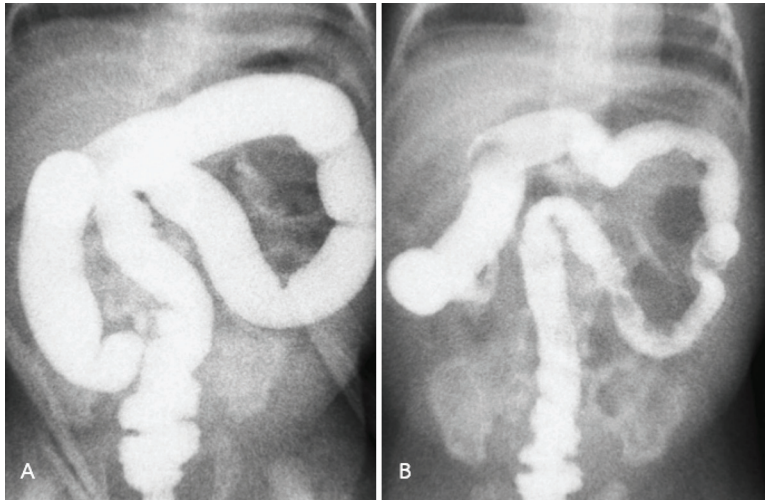
### Imaging findings

The most common imaging finding was the transitional zone in 40 (83.33%). Other imaging findings were the reverse rectosigmoid ratio in 17 (35.41%), serration in nine (18.75%), microcolon in three (6.25%), and short colon in one (2.08%). Three cases had non-diagnostic findings on contrast enema, and all were total colonic aganglionosis. Imaging findings were stratified by type of disease and displayed in Table 2.

The transitional zone was also the most typical

**Table 2.** Imaging findings on contrast enema stratified by type of Hirschsprung disease

	Reverse rectosigmoid ratio	Transitional zone	Serration	Microcolon	Short colon	Atypical imaging findings
Short segment; n	11	27	5	0	1	0
Long segment; n	3	5	0	0	0	0
Total colonic; n	0	3	2	2	0	3
No data; n	3	5	2	1	0	0
Sum; n (%)	17 (35.41)	40 (83.33)	9 (18.75)	3 (6.25)	1 (2.08)	3 (6.25)



**Figure 7.** A 1-month-old boy with total colonic aganglioneuroplasia presented with abdominal distention. (A) AP view from contrast enema shows no abnormal bowel dilatation. (B) The delayed 30-minuted post-evacuation film shows a large amount of residual contrast media in the large bowel.

finding in cases of short and long segment disease. Both the transitional zone finding and the non-diagnostic imaging finding were equally common in cases of total colonic aganglioneuroplasia.

For the three cases with non-diagnostic imaging, they were infants with aged ranging from four days to three months and two cases were male. All cases presented with abdominal distention. Non-diagnostic imaging findings included persistent narrowing of the sigmoid without proximal bowel dilatation (Figure 6), a large amount of residual contrast media in large bowel after 30 minutes delayed evacuation (Figure 7), and persistent small caliber of the rectosigmoid colon with mild proximal bowel dilatation and normal distension of the lower rectum during introducing contrast media (Figure 8).

## Discussion

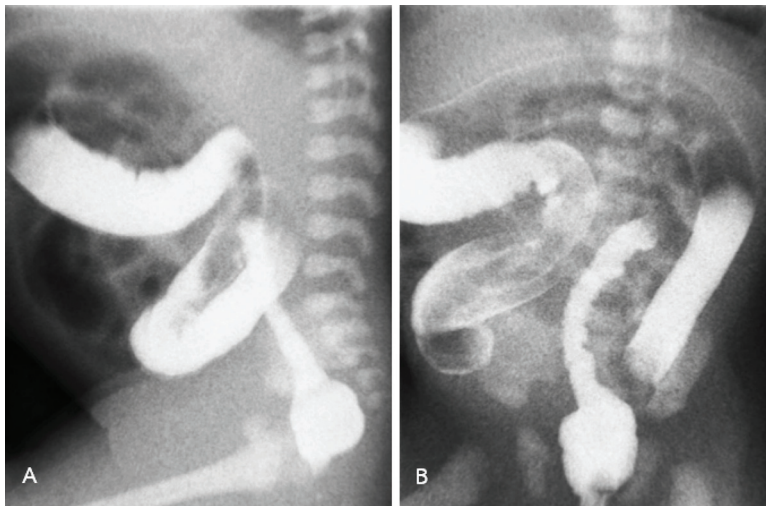
Despite the rarity of Hirschsprung disease, in the past 19 years, there have been 108 cases proven by histopathology in King Chulalongkorn Hospital. Additionally, the present study demonstrates that Hirschsprung disease is more common in males

than in females at a ratio of 5:1, and 87.5% of the cases manifest within the first year of life, which is consistent with the earlier studies<sup>(2,9)</sup>. The majority of the cases presented with abdominal distention in 52.08%.

The short segment is the most common type of disease while total colonic aganglioneuroplasia is uncommon and total intestinal type is very rare. Similar to the present study data, which showed find that short-segment Hirschsprung disease is the most frequent type, total colonic aganglioneuroplasia has few cases, while there was no case of total intestinal disease type.

Like the previous studies, common contrast enema imaging abnormalities in Hirschsprung disease are transitional zone, reverse rectosigmoid ratio, and serration. Among common imaging findings, the transitional zone is the most frequent imaging abnormalities.

The rates of positive results for Hirschsprung disease varied according to the earlier studies. According to a cross-sectional study done on 60 neonates with delayed meconium passage over



**Figure 8.** A 4-day-old-girl total colonic aganglionosis presented with abdominal distention and feeding intolerance. (A) Lateral view and (B) AP view from contrast enema show the persistent segmental small caliber of the rectosigmoid colon with mild proximal bowel dilatation and normal distension of the lower rectum.

the course of a year in Iran<sup>(9)</sup>, the rates of reverse rectosigmoid ratio, transitional zone, and serration were 76%, 90%, and 23%, respectively. To compare with the present study, the results show the rates of reverse rectosigmoid ratio, transitional zone, and serration were 35%, 83%, and 18% respectively. The main difference is the present study much lower rate of reverse rectosigmoid ratio discovery. The exclusion of some cases with a reverse rectosigmoid ratio could be the cause of this outcome. However, there was no results from histopathology or radiology that could cause this outcome.

Although non-diagnostic findings on contrast enema were rarely found, the author demonstrated that non-diagnostic findings were detected in three out of six cases of total colonic aganglionosis. On the other hand, 50% (3 cases), had one of the typical findings of transitional zone, serration, and short colon. Non-diagnostic imaging findings show persistent narrowing of the sigmoid without proximal bowel dilatation, a large amount of residual contrast media in large bowel after 30-minute delayed evacuation and small caliber of the rectosigmoid colon with mild proximal bowel dilatation and normal distension of the lower rectum during introduced contrast media. While the previous single center-retrospective study designed for evaluating barium enema finding in total colonic aganglionosis in 17 neonatal patients found that the reliable radiographic sign is ileocecal reflux, this was found in approximately 50% of the patients<sup>(11)</sup>. The present study has only one case of ileocecal reflux. Another retrospective study aimed to

evaluate the contrast enema findings of total colonic aganglionosis of fourteen cases, and showed that the short and rigid colon, the microcolon, and the transition zone proximal to the cecum were found significantly more often than in the patients without total colonic aganglionosis<sup>(12)</sup>. Furthermore, they documented that poor rectal distensibility, colonic wall irregularity, and poor contrast emptying were also commonly found.

In cases of non-diagnostic findings, the present study also differs from the prior studies. Therefore, if total colonic aganglionosis is clinically suspected, it may be difficult to diagnose, and a biopsy is still required to make a conclusive diagnosis. However, the small total number of cases could be the reason for the discrepancy in these results.

### Limitation

This study has limitations. First, the limitation is similar to other retrospective studies. Many cases lack clinical information due to old data. Secondly, there are few cases.

### Conclusion

Common imaging findings for Hirschsprung's disease on contrast enema at King Chulalongkorn Memorial Hospital are transitional zone, reverse rectosigmoid ratio, and serration, in descending order. Non-diagnostic findings are typically uncommon findings, but total colonic aganglionosis frequently exhibits them in a variety of patterns. Therefore, if a diagnosis was clinically suspected, a biopsy is still

required to confirm it.

### What is already known on this topic?

In prior studies, common contrast enema imaging abnormalities in Hirschsprung disease are transitional zone, reverse rectosigmoid ratio, and serration. In cases of total colonic Hirschsprung disease, the enema shows a microcolon and impaired contrast evacuation.

### What this study adds?

Like previous study, common imaging findings for Hirschsprung disease on contrast enema are transitional zone, reverse rectosigmoid ratio, and serration. However, in cases of total colonic Hirschsprung disease, this study results differed from the prior studies, that it can have a variety of patterns.

### Conflicts of interest

The authors declare no conflict of interest.

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