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Suspected Hirschsprung's Disease in Infants: The Diagnostic Accuracy of Contrast Enema

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Abstract ***Objective:*** To determine the accuracy of contrast enema (CE) in diagnosis of Hirschsprung's disease (HD) in infants. ***Methods:*** A total of 23 CE films of infants with suspected HD were collected in a period of two years. Of those, 11 cases had histological confirmed diagnosis of HD, and 12 cases had HD excluded. The radiographs were reviewed independently by three pediatric radiologists without knowing the histological diagnosis. Radiological signs of HD, including transition zone, spastic colon and reversed recto-sigmoid index were commented by the radiologists in all radiographs. Also, they need to commit the overall impression for or against the radiological diagnosis of HD in each case at the end of examination. ***Results:*** For transition zone sign, the mean false positive and false negative rates were 23.1% and 40.6% respectively. For spastic colon sign, the false positive and false negative rates were 15.3% and 59.1% respectively. Reversed recto-sigmoid index gave false positive and false negatives rates of 18.1% and 57.6% respectively. For overall impression of radiological diagnosis of HD, the false positive and false negative rates were 18.8% and 44.3% respectively. Among all three radiologists, concordant and correct radiological diagnosis occurred in 11 out of 23 cases (47.8%). ***Conclusions:*** Radiological signs shown in CE have high false positive and high negative rates in diagnosis of HD. There is significant discordance among different radiologists in reading CE radiographs. Diagnosis of HD should not be relied on CE alone.

Key words Contrast enema; Diagnosis; Hirschsprung's disease

Introduction

Hirschsprung's disease (HD) is a congenital intestinal disorder characterised by aganglionosis of the distal bowel.[1] The clinical symptoms of HD may include delayed passage of meconium beyond 24 hours of life, signs and symptoms of large bowel obstruction such as bilious vomiting, a distended abdomen, feeding intolerance, failure to thrive, severe defaecation problems and enterocolitis.[2] Several diagnostic tests have been described for patients whom HD is suspected such as demonstration of the absence of recto-anal inhibitory reflex (RAIR) in anorectal manometry and an elevated cholinesterase activity and aganglionosis in rectal suction biopsy. However, these tests can sometimes be expensive and invasive, and specialised equipments may also be required if the test is to be performed on a small size infant or even neonate. As a relatively non-invasive procedure, the use of contrast enema (CE) is sometimes a favourable diagnostic option for suspected HD in infants. A systemic review by De Lorijin et al,[3] had quoted a sensitivity rate of 70% and a specificity rate of 83% in using CE as the initial diagnostic test for the workup of HD. However, the evaluation of the CE can often be subjective and the sensitivity of diagnosis of HD from a CE may sometimes be observer dependent.

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The objective of this study was to review the diagnostic accuracy of contrast enema in infants with suspected HD and to investigate the potential concordant rate among different radiologists in the interpretation of the CE radiographs.

Methods

We retrospectively reviewed the contrast enema radiographs of infants with suspected HD collected in a period of two years in our centre. All enema were done using water soluble contrast via a non-ballooned rectal catheter. All children did not undergo bowel washout 24 hours prior to the contrast enema procedure. And digital manipulations including digital per rectal examination were also avoided prior to the contrast enema procedure.

These contrast enema radiographs were reviewed independently by three post fellow level paediatric radiologists with more than ten-year experience in interpreting paediatric radiographs. They were all blinded to the histological diagnosis and clinical conditions of each infant. Commonly used radiological signs of HD, including transition zone, spastic colonic segment and reversed recto-sigmoid index were commented by the radiologists in all radiographs. The radiologists were asked to make decisions on whether there was presence or absence of the radiological sign, or if it was an equivocal finding. They were also asked to commit the overall impression for or against the radiological diagnosis of HD in each case at the end of the review.

The data were then analysed and tabulated.

Results

A total of 23 CE radiographs of infants aged six months or below with suspected HD were reviewed. Of these, 11 patients had histological confirmed diagnosis of HD and 12 patients had HD excluded by rectal biopsy.

For the 11 patients with histological confirmed HD, the sensitivity of detecting transition zone were 63.6%, 54.5% and 60.0% by the three independent paediatric radiologists respectively, giving a mean sensitivity of 59.4%. For the 12 patients with confirmed non-HD, the specificity of detecting the absence of transition zone were 75.0%, 66.7% and 88.9% by the three independently paediatric radiologists respectively, giving a mean specificity of 76.9%. The concordant rate for the correct radiological diagnosis was 12/23 (52.2%).

For the 11 patients with histological confirmed HD, the sensitivity of detecting spastic colon were 27.2%, 45.5% and 50.0% by the three independently paediatric radiologists respectively, giving a mean sensitivity of 40.9%. For the 12 patients with confirmed non-HD, the specificity of detecting the absence of spastic colon were 81.8%, 83.3% and 88.9% by the three independently paediatric radiologists respectively, giving a mean specificity of 84.7%. The concordant rate for the correct radiological diagnosis was 9/23 (39.1%).

For the 11 patients with histological confirmed HD, the sensitivity of detecting reversed recto-sigmoid index were 50.0%, 27.3% and 50.0% by the three independent paediatric radiologists respectively, giving a mean sensitivity of 42.4%. For the 12 patients with confirmed non-HD, the specificity of detecting the absence of reversed recto-sigmoid index were 83.3%, 75.0% and 87.5% by the three independent paediatric radiologists, giving a mean specificity of 81.9%. The concordant rate for the correct radiological diagnosis was 7/23 (30.4%).

For the 11 patients with histological confirmed HD, the sensitivity of diagnosing the correct diagnosis of HD from an overall impression were 57.1%, 50.0% and 60.0% by the three independent paediatric radiologists respectively, giving a mean sensitivity of 55.7%. For the 12 patients with histological confirmed non-HD, the specificity of diagnosing the correct diagnosis of non-HD from an overall impression were 83.3%, 72.7% and 87.5% by the three independent paediatric radiologists respectively, giving a mean specificity of 81.2%. The concordant rate for the correct radiological diagnosis was 10/23 (43.5%).

The detailed breakdown of the summary of the results is shown in Tables 1-4.

None of the histologically confirmed HD had aganglionic bowel segment extending proximal to the distal two third transverse colon. i.e. no long segment HD and no total colonic aganglionosis were included in this study.

In summary, the mean sensitivity of detecting the radiological signs of transition zone, spastic colon, reversed recto-sigmoid index and the overall impression in histological confirmed HD patients are 59.4%, 40.9%, 42.4% and 55.7% respectively. This would in turn give an overall mean sensitivity rate of 49.6% in positively identifying HD patients with the above mentioned radiological signs from the contrast enema.

The mean specificity of detecting the absence of the radiological signs of transition zone, spastic colon, reversed recto-sigmoid index and the overall impression in

Table 1a Histological confirmed HD patients: transition zone sign

	1	2	3	4	5	6	7	8	9	10	11	Sensitivity
A	✓	✓	×	×	✓	×	✓	✓	✓	×	✓	63.6%
B	×	×	×	×	✓	✓	✓	✓	✓	×	✓	54.5%
C	O	×	✓	×	✓	×	✓	✓	✓	×	✓	60.0%

✓ = transition zone present (correct diagnosis); × = transition zone absent (incorrect diagnosis); O = equivocal

Table 1b Histological confirmed non-HD patients: transition zone sign

	1	2	3	4	5	6	7	8	9	10	11	12	Specificity
A	✓	✓	✓	✓	✓	✓	✓	✓	×	×	×	✓	75.0%
B	✓	✓	✓	✓	✓	✓	✓	✓	×	×	×	×	66.7%
C	✓	✓	✓	✓	✓	✓	O	✓	O	✓	×	O	88.9%

✓ = transition zone absent (correct diagnosis); × = transition zone present (incorrect diagnosis); O = equivocal

Table 2a Histological confirmed HD patients: spastic colon sign

	1	2	3	4	5	6	7	8	9	10	11	Sensitivity
A	×	×	×	×	✓	×	×	×	✓	×	✓	27.2%
B	×	×	×	×	✓	×	✓	✓	✓	×	✓	45.5%
C	×	×	✓	×	✓	×	O	✓	✓	×	✓	50.0%

✓= spastic colon present (correct diagnosis) ; × = spastic colon absent (incorrect diagnosis); O = equivocal

Table 2b Histological confirmed non-HD patients: spastic colon sign

	1	2	3	4	5	6	7	8	9	10	11	12	Specificity
A	✓	✓	✓	✓	✓	✓	✓	✓	×	O	×	✓	81.8%
B	✓	✓	✓	✓	✓	✓	✓	✓	✓	×	×	✓	83.3%
C	✓	✓	✓	✓	O	✓	O	✓	✓	✓	×	O	88.9%

✓ = spastic colon absent (correct diagnosis); × = spastic colon present (incorrect diagnosis); O = equivocal

Table 3a Histological confirmed HD patients: reversed recto-sigmoid index

	1	2	3	4	5	6	7	8	9	10	11	Sensitivity
A	×	✓	×	×	✓	×	✓	✓	O	✓	×	50.0%
B	×	×	×	×	✓	×	✓	×	×	×	✓	27.3%
C	O	×	✓	×	✓	×	✓	✓	×	×	✓	50.0%

✓ = reversed recto-sigmoid index present (correct diagnosis); × = reversed recto-sigmoid index absent (incorrect diagnosis); O = equivocal

Table 3b Histological confirmed non-HD patients: reversed recto-sigmoid index

	1	2	3	4	5	6	7	8	9	10	11	12	Specificity
A	✓	✓	×	✓	✓	✓	✓	✓	×	✓	✓	✓	83.3%
B	✓	✓	✓	✓	✓	✓	✓	✓	✓	×	×	×	75.0%
C	✓	✓	✓	✓	O	✓	O	✓	O	✓	×	O	87.5%

✓= reversed recto-sigmoid absent (correct diagnosis); × = reversed recto-sigmoid present (incorrect diagnosis); O = equivocal

Table 4a Histological confirmed HD patients: overall impression

	1	2	3	4	5	6	7	8	9	10	11	Sensitivity
A	O	O	×	×	✓	×	✓	O	✓	O	✓	57.1%
B	×	×	×	×	✓	O	✓	✓	✓	×	✓	50.0%
C	O	×	✓	×	✓	×	✓	✓	✓	×	✓	60.0%

✓ = diagnostic of HD (correct diagnosis); × = not diagnostic of HD (incorrect diagnosis); O = equivocal

Table 4b Histological confirmed non-HD patients: overall impression

	1	2	3	4	5	6	7	8	9	10	11	12	Specificity
A	✓	✓	✓	✓	✓	✓	✓	✓	×	✓	×	✓	83.3%
B	✓	✓	✓	✓	✓	✓	✓	✓	O	×	×	×	72.7%
C	✓	✓	✓	✓	O	✓	O	✓	O	✓	×	O	87.5%

✓ = not diagnostic of HD (correct diagnosis); × = diagnostic of HD (incorrect diagnosis); O = equivocal

histological confirmed non-HD patients are 76.9%, 84.7%, 81.9% and 81.2% respectively. This would in turn give an overall mean specificity rate of 81.2% in successfully excluding HD with the above mentioned radiological signs from the contrast enema.

The correct concordant rate for the correct radiological diagnosis among the three radiologists for the transition zone, spastic colon, reversed recto-sigmoid index and the overall impression are 52.2%, 39.1%, 30.4% and 43.5% respectively. This would in turn give an overall mean concordant rate of 41.3% among the three independent paediatric radiologists in the interpretation of the above mentioned radiological signs from the contrast enema.

Discussion

Our findings demonstrated that radiological signs in contrast enema in the diagnosis of HD have a low mean sensitivity rate of 49.6% and a moderately low mean specificity rate of 81.2%; implying that there is high false positive and moderately high false negative rates in the diagnosis of HD.

Moreover, there is significant discordance among the radiologists with a low mean concordant rate of 41.3%; implying that there is a significant inter-observer difference in the interpretation of radiological signs from the contrast enema among the three independent paediatric radiologists.

Previous studies have shown the transition zone and reversed recto-sigmoid index as the most common radiological signs in CE.[4,5] The sensitivity and specificity of CE in the diagnosis of HD have shown a wide range, between 60%-100%.[6] This could be due to the difference in patients selection: patients of different age and extent of disease; and difference in the skill levels of the radiologists.[5] In our study, we tried to standardise and only reviewed the CE films done on patients that were aged six months and younger, and none of the 11 patients with histological confirmed HD had aganglionosis involving more proximal than the mid transverse colon. Moreover, only paediatric radiologists with similar years of clinical experience were recruited.

However, one of the potential bias in our study is that due to the retrospective nature of reviewing the CE films, only non-dynamic print out of the CE films were used, this could lead to potential selection bias if the films chosen were not the most representative of the patients'. Furthermore, no real time interpretation of the actual contrast enema procedure was possible, which may also affect the sensitivity of detecting some potentially dynamic radiological signs that involve interpreting real time movement of the colon during the procedure.

We were surprised to see the low concordant rate among the different paediatric radiologists and we postulated that this could be due to the "subjective" nature in the interpretation of these radiological signs. We chose the four radiological signs: transition zone, spastic colon, reversed recto-sigmoid index and the overall impression of the CE films because these were the most commonly described features that had been reported in the contrast

enema films in our centre. As compared to another commonly used radiological sign, such as the 24 hours post contrast enema evacuation sign, these were more "subjective" in nature and theoretically would be more observer dependent.

In conclusion, our results had shown that the use of contrast enema in the diagnosis of HD had high false positive and high false negative rates, and that there was significant discordance among different radiologists in reading CE films. Therefore we suggest that the diagnosis of HD should not be relied on contrast enema alone.

Declaration of Interest

None.

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