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Diagnosis of Hirschsprung's Disease : Accuracy of Barium Enema Findings[1]

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Purpose : To determine the relative accuracy of barium enema findings of Hirschsprung's disease (HD) and to calculate a level of probability for three signs combined.

Materials and Methods : Barium enema findings in 45 patients who had undergone rectal biopsy to prove or exclude the diagnosis of HD were retrospectively analyzed by evaluating the presence of a transition zone, irregular contractions and delayed evacuation of barium. Seventeen were neonates (group 1) and the other 28 were infants and children (group 2). The sensitivity, specificity, and positive and negative predictive values of the findings were compared.

Results : In visualization of a transition zone, sensitivity, specificity and positive predictive value were 76.5%, 72.7% and 89.7%, respectively. Sensitivity for irregular contractions and delayed evacuation of barium was 76.5% and 91.7%, respectively, whereas for specificity, the corresponding values were 63.6% and 40%. Sensitivities for radiologic signs were higher in group 1 than in group 2, but, the specificities were lower. If two or three findings were positive, the level of probability was 85-100%. If two findings were negative, however, the corresponding value was 30%.

Conclusion : We conclude that the most reliable HD finding is the presence of a transition zone. Irregular contractions and the delayed evacuation of barium are not specific. Two or three positive findings may suggest a higher probability of HD than any single positive finding alone.

Index Words : Children, gastrointestinal tract
Colon, aganglionosis
Infants, newborn, gastrointestinal tract

Since Hirschsprung's disease (HD) in neonates and infants is often lethal with high mortality from enterocolitis, early diagnosis is essential(1). Traditionally, a barium enema can serve as the initial diagnostic procedure, both to establish diagnosis and to elicit useful information about the cause of poor evacuation (2). It has been shown that a radiologic study alone is not sensitive enough to exclude HD.

The purposes of this study were to evaluate the reliability of individual findings of the barium enema examination and to determine the probability of HD when three findings are combined.

Materials and Methods

We retrospectively studied the medical records and barium enema radiographs of all neonates, infants and children referred for the radiologic evaluation of an evacuation problem. The barium enema was performed without prior bowel preparation or rectal examination. A soft rubber catheter was placed with the tip just proximal to the anorectal junction. In order to detect a distal transition zone, the examination was begun in lateral projection. Barium diluted with isotonic saline was introduced slowly, with careful moni-

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toring of the filling rate. To delinate intermittent occurrence of irregular contractions or narrowing in the aganglionic segment and the distal transition zone, multiple initial spot films were obtained. After the transition had been identified, no additional barium was injected. When the barium enema radiographs were available, 45 patients from 3 days to 8 years old (median, 2 months) were included in our series ; to rule out HD, rectal biopsy was performed in each patient. Seventeen of these patients were neonates (group 1), and the remaining 28 were infants and children (group 2). One pediatric and two gastrointestinal radiologists working together reviewed all roentgenograms for the presence of a transition zone, irregular contractions at the aganglionic segment and delayed evacuation of barium, as seen on film taken at 24 hours. At the time of this study they were not aware of the final diagnosis. A transition zone was considered to be present when an abrupt change in bowel width caused by a narrowed aganglionic segment distal to dilated normal colon was noted. When a definite transition zone was not obvious, the rectosigmoid index (< 1) was measured. Irregular contraction of the aganglionic segment was defined as "spastic narrowing", and its serrated appearance as "saw-tooth" or "corrugated". The evacuation of barium was considered to be delayed if barium remained proximal to the sigmoid colon at 24 hrs. after examination. Delayed radiographs of 29 patients, including 14 neonates and 15 infants and children were available.

Data relating to the 17 patients in group 1, the 28 in group 2, and to the 45 collectively, was statistically analysed.

Each radiologic sign was evaluated for sensitivity, specificity, positive predictive value (PPV) and negative predictive value (NPV). PPV demonstrates the percentage of patients with each sign who actually had HD and represents the true positive rate for each sign ; NPV is the percentage of patients without each sign who were disease free.

In the second part of the analysis, we obtained summary scores. The absence of a radiologic finding was scored as 1, the presence as 2. The range of total scores varied between 3 and 6. For each summary score, we calculated a level of diagnostic probability of HD.

Results

Table 1 shows the predictive value of radiologic findings in each group.

Thirty-four of these cases were confirmed as HD and eleven patients were proved to be HD-free. The final diagnoses in this latter group were considered to be meconium plug syndrome($n=2$) and necrotizing enterocolitis($n=2$) in group 1, and functional megacolon ($n=5$) and pseudo-obstruction ($n=2$) in group 2.

Radiographs of 29 patients were obtained at 24 hours and the results of these are shown in Table 2.

A transition zone was seen in 26 of 34 patients (76.5%) with HD (Fig. 1), and was more often seen in group 1 (92.3%) than in group 2 (66.7%). This zone could not be identified in one neonate with total colonic aganglionosis whose barium enema showed loss of redundancy of the sigmoid colon and reversed propulsion of barium into the distal ileum on 24-hour delayed film. On rectal biopsy, however, three cases were seen to have normal ganglionic cells, despite the presence of a radiologic transition zone.

Transition zone sensitivity and specificity in group 1 (and group 2) were 92.3%(66.7%) and 50%(85.7%), respectively. Overall sensitivity and specificity were 76.5% and 72.7%, while PPV was 89.7% ; 26 of 29 patients with a radiologic transition zone were, in other words, correctly identified by the presence of a

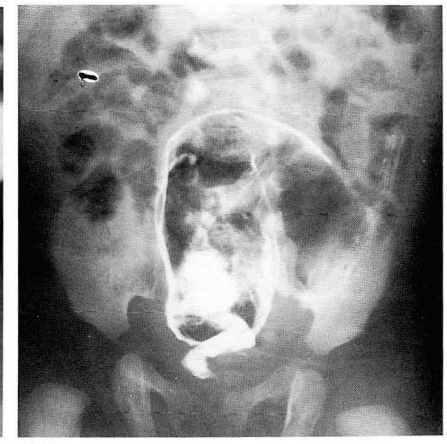
Table 1. Comparing the Predictive Power of Radiologic Findings in 45 Patients

Group Radiologic Findings	% Sensitivity	% Specificity	% Positive Predictive Value	% Negative Predictive Value
Neonates				
TZ	92.3	50.0	85.7	75.0
IC	84.6	25.0	92.9	33.3
DE*	90.9	33.3	83.3	50.0
Infants & Children				
TZ	66.7	85.7	93.3	46.2
IC	71.4	85.7	93.8	50.0
DE*	92.3	50.0	92.3	50.0

TZ : transition zone. IC : irregular contractions in the aganglionic segment.
DE : delayed evacuation of barium on 24-hour radiograph.
* Data for 29 patients who had a 24-hour delayed film available.

1

2A



2B

Fig. 1. Barium enema in 1-week-old male neonate shows a transition zone from the narrow rectum to the dilated sigmoid (open arrows). Note associated irregular contractions and mucosal irregularity in the narrow rectum (filled arrow).
Fig. 2. Barium enema in 1-month-old male infant show short-segmental spastic narrowing of distal rectum(**A**). On 24 hours delayed radiograph, retained barium is not proximal to the sigmoid, but in the distal rectum as boli (**B**). This infant had normal ganglionic cells in rectal biopsy.

Table 2. Summary of Radiologic Findings in Predicting HD in 29 Patients

Radiologic findings	% Sensitivity	% Specificity	% Positive Predictive Value	% Negative Predictive Value
TZ	76.5	72.7	89.7	50.0
IC	76.5	63.6	86.7	46.7
DE	91.7	40.0	88.0	50.0

TZ : transition zone. IC : irregular contractions in the aganglionic segment.
DE : delayed evacuation of barium on 24-hour radiograph.

sign as having HD.

Twenty-six of 34 patients (76.5%) with HD had irregular contractions in the aganglionic segment, as seen on fluoroscopy or overhead radiographs. Of the total of 30 patients with irregular contractions, saw-tooth or corrugated narrowing was noted in 22 (Fig. 1) and spastic narrowing in eight (Fig. 2). There were four cases of false positive findings of irregular contractions, including three neonates (Fig. 3) and one child ; PPV was therefore 86.7%. Overall sensitivity and specificity for irregular contractions were 76.5% and 63.6%.

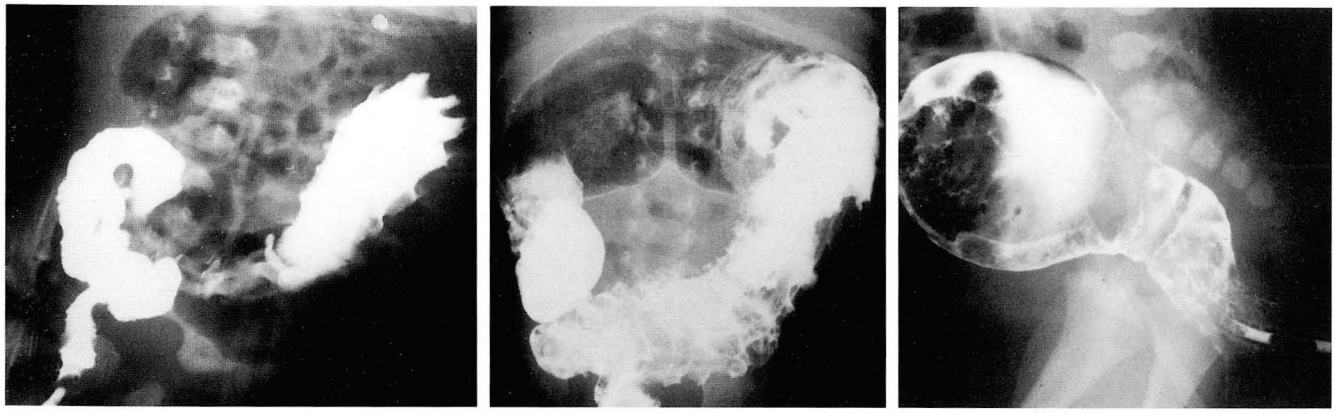
Two patients in whom evacuation of barium was normal were subsequently found to have HD. Ten of 11 neonates with HD(90.9%) were seen on film taken at 24 hours to have retained barium. A total of three cases in which barium was retained were found to be false-positive, and so 88% of patients with delayed evacuation of barium had HD(PPV). Sensitivity for delayed evacuation was 91.7%, while specificity was 40.0%.

Two or three positive findings (scores of 5-6) suggest a high probability of HD (85-100%). If, however, two findings are negative (scores of 4), there is a low probability of HD (30%).

Discussion

Barium enema has been used as an initial screening procedure in the evaluation of infants and children with constipation suggestive of HD(3). Although submucosal rectal biopsy is deep enough for findings to confirm or rule out HD, radiographic or manometric evidence of HD may help the pediatric surgeon decide whether to perform a rectal biopsy(4). Despite the accumulation of radiologic findings of HD, the diagnosis of disease remains a major problem for clinicians and radiologists.

The transition zone was believed to be the single most reliable finding for diagnosis of HD (5). Many authors have commented, however, that this finding was not always present in HD patients. Visualization of the transition zone was reported in 15 of 19 HD patients by Berman(6) and in 10 of 16 HD patients by McDonald and Evans(7). According to Rosenfield's report (2), 19 of 32 HD patients (59%) had a transition



A B 4

Fig. 3. Barium enema in 3-day-old male neonate shows mucosal cobble stone pattern in descending and sigmoid colon and saw-tooth or corrugated appearance of the rectum. Transition zone is near the rectosigmoid junction (**A** & **B**). This neonate had normal ganglionic cells in rectal biopsy. There was clinical evidence of necrotizing enterocolitis at that time.

Fig. 4. Barium enema in 2-month-old male infant shows a rectum of normal diameter and a disproportionate dilatation of sigmoid with large bolus of fecal material lodged at the rectosigmoid. Pathologic examination reveals aganglionosis extending throughout the entire rectum and sigmoid. This case shows impacted stool may passively dilate the aganglionic segment.

zone.

Many authors have mentioned that the diagnosis of HD becomes easier as the child gets older, since a transition zone is more often visualized in the older child.

Our data suggest that the proportion of false-negatives is higher in group 2 than in group 1 ; it is possible that in group 2, a distal aganglionic segment passively dilated by impacted stool or previously repeated cleansing enema may obscure the transition zone.

As noted by Bodian et al. (8), the transition zone seen radiologically and seen at surgery may differ from the true pathologic transition seen on microscopic study. Peristalsis and fecal masses can dilate the proximal aganglionic bowel as well as the normal ganglionic segment. Except in some cases, we were able to predict relatively accurately the location of the transition zone ; in the cases in which prediction was not accurate, appararent rectosigmoid levels of caliber transition seen just above the rectosigmoid junction were found to be sigmoid colon with microscopic transition (Fig. 4). The pseudotransition zone of a narrow descending colon has been described to functional immaturity of the large bowel, so called meconium plug syndrome or neonatal small left colon syndrome(9, 10). This, with a transition at the splenic flexure, was seen in two of our neonates.

The finding of a transition zone as the diagnostic sign showed high sensitivity, specificity and positive predictive value.

Irregular contractions in the aganglionic segment are apparently the result of denervation hypersensitivity of the smooth muscle(11). In the absence of a transitional zone or confusing dilated aganglionic segment, these irregular contractions revealed the true extent of aganglionosis(4). The absence of these bizzare, irregular contractions certainly militates against the presence of HD (12).

Irregular contractions were noted in four patients in our series whose biopsy results were normal. On radiographs taken at 24 hours, it was seen that in two of these, barium had been well-expelled, and this raises the possibility of enterocolitis. The sensitivity of this sign as high as that of the transition zone, but the specificity was not high.

Delayed radiograph obtained at 24 hours after barium enema to document poor emptying of the colon have also been considered important in the diagnosis of HD in neonates and infants because of their normally increased stool frequency compared with older children(1).

The lowest sensitivity and specificity found in infants, 57% and 27% respectively, was reported by Taxman et al. (13). They noted that the delayed evacuation of barium is commonly present in infants with constipation and is not a reliable predictor of HD. The highest sensitivity recorded for infants, 84% was reported in the findings of Rosenfield et al. (2). It has been suggested that in our group, the high sensitivity (91.7%) for the total group and increased value for group 2 were representative of only a small percentage of available roentgenograms taken at 24 hours.

Two of 24 patients who did not undergo cleansing enema or rectal manipulation, but whose 24 hr. radiographs showed they had expelled barium, whereas suffering from HD. The reasons for this remain unclear, but some authors have suggested that the cause of emptying is related to underlying enterocolitis(14).

According to Rosenfield et al. (2), a comparison of 24-hr. and 48-hr. radiographs may be helpful, but our data not include findings based on film taken at 48 hrs.

In this study, we conclude that the presence of a transition zone, with high sensitivity and specificity, is the finding which most reliably indicates the presence of HD. Irregular contractions in the aganglionic segment and delayed evacuation showed sensitivities indicative of HD, but, their specificities are low. Although in our series there are high false-negative rates for barium enema findings alone, two or three such findings which are positive may suggest a higher probability of HD than any single radiologic finding alone.

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Hirschsprung's Disease의 바륨대장검사소견상 진단의 정확도[1]

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목 적 : Hirschsprung's disease (HD)의 대장조영검사 소견들에 대한 진단의 정확도를 알아보고자 한다.

대상 및 방법 : HD로 의심되어 대장조영검사를 실시하고 병리조직학적으로 확인된 45명의 환아들을 대상으로 하였으며 연령에 따라 신생아군(1군)과 영유아군(2군)으로 분류하였다.

술 전 대장조영검사소견으로는 이행대 (transition zone)의 출현, 불규칙한 대장 수축 (irregular contraction) 및 배설시간 지연 (delayed evacuation of barium)의 유무를 조사하였고 이를 후향적으로 비교분석하였다.

결 과 : HD를 예측하는데 있어 이행대의 소견은 76.5%의 예민도와 72.7%의 특이도를 보였다. 불규칙한 대장수축 및 배설시간지연 소견의 예민도는 각각 76.5%와 91.7%로 높았으나 특이도는 63.6%와 40%로 낮았다. 연령별로 볼 때 대장조영검사 소견들의 예민도는 신생아군에서 영유아군보다 높았으나 그 특이도는 낮았다. 또한 이들 소견중 두가지이상이 있는 경우 HD진단의 확률수준은 85-100%인 반면, 어떠한 한가지 소견만 있을 경우에서는 30%로 낮은 수준이었다.

결 론 : 대장조영검사상 HD를 진단하는데 있어 가장 신뢰도가 높은 소견은 이행대이었고 불규칙한 대장수축 및 배설시간지연 소견의 특이도는 낮은 수준이었다. 그리고 이들 소견중 두가지이상이 있는 경우 한가지 소견만 있는 군보다 HD진단의 확률수준이 월등히 높았다. 따라서 HD의 진단의 정확도를 높이는데 있어 이러한 대장검사소견들의 병합분석이 중요하리라 생각된다.