The Diagnostic Value of Anorectal Manometry as a Screening Test for Hirschsprung's Disease

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Abstract

The diagnostic accuracy of anorectal manometry as a screening test for Hirschsprung's disease (HD) was assessed in 46 children suspected of having HD on clinical grounds. Among 33 children in whom a rectosphincteric reflex (RSR) was not demonstrated on manometric recordings, 31 were later proven to have HD by full thickness rectal biopsy : the remaining 2 children were shown not to have HD. None of the 12 children who had a normal RSR had HD. Thus, two false positive diagnoses were observed in this study, giving an overall diagnostic accuracy by anorectal manometry of 95 per cent and diagnostic sensitivity of 100 per cent. Anorectal manometry is an accurate, non-invasive test which has a place as the screening test of choice for HD.

Key word : Anorectal Manometry, Diagnosis, Hirschsprung's Disease

To date, there has been some debate on which investigation is the most appropriate screening test for Hirschsprung's disease (HD), since neither, barium enema, rectal suction biopsy (H & E, histochemistry) or anorectal manometry have been shown to have 100 per cent diagnostic accuracy. Each of these tests has both advantages and disadvantages in terms of availability, technical difficulty, radiation exposure, and invasiveness. Consequently, the choice of screening test and order of investigations for HD differs among medical centres.

Anorectal manometry has been recommended by some investigators as an initial screening test of choice because it is safe, reliable and the least invasive, and has a physiologic basis[1,2]. By this test, an absence of rectosphincteric reflex (RSR) in HD is demonstrated. The purpose of this study is to report our experience in the use of anorectal manometry for diagnosis of HD.

PATIENTS AND METHOD

Between May 1993 - Aug 1996, 48 children in whom there was a clinical suspicion of HD under-

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went anorectal manometry at Songklanagarind Hospital. Two children were not available for follow-up after the manometric study, and therefore the series comprised 46 children.

The clinical features of 46 children were categorized into two groups based on their likelihood of having HD on clinical grounds.

Clinically typical HD (Group I) was made up of 25 children presenting with chronic faecal retention, abdominal distension, and a history of delayed passage of meconium since birth. This group was considered to have the typical clinical presentation of HD.

Clinically atypical HD (Group II) consisted of 21 children, including 11 having chronic constipation, with an obscure history of delayed passage of meconium since birth, 8 presenting with clinical features suggesting distal colonic obstruction, and 2 very young infants who had bowel movement changes from several stools per day during the early days of life to once in several days.

Following manometric study, all the patients who did not have RSR underwent barium enema and full thickness rectal biopsy. However, the decision to perform further investigations for HD in patients who had RSR was based on their clinical features of group I or group II since it is unethical to perform such invasive investigations, before considering medical treatment, in cases where RSR was present, and they also had no clinically typical HD. Investigations and management in the studied patients are summarized in the diagram. The definitive diagnosis of HD relied on an absence of ganglion cells from full thickness rectal biopsy. Patients of group II who had normal RSR on manometric study and an improvement of constipation with medical treatment did not undergo full thickness rectal biopsy. These patients were considered not to have HD (diagram).

Techniques used in anorectal manometric study : A triple-lumen manometric anal probe of 3 mm outer diameter was used. The catheter had two sideholes, 2 cm apart, radiating at 120° and a distensible latex balloon fixed to the distal end to assess reflex internal anal sphincter response to intrarectal balloon distension. The catheter was perfused with water at a constant flow rate of 0.5 ml/min through a motor drive pump (Albyn Medical, Dingwall, UK). Anal pressure was recorded by a water filled strain-gauge and displayed on a personal computer using a commercial analysis program (Albyn Medical Phoenix and Griffon system, Dingwall, UK).

Investigations were performed with the subjects in the left lateral position with flexed knees and hips. Bowel preparation with saline or polyethylenum glycol enema was performed in every patient at least 2 hours before study and the use of sedations (oral chloral hydrate or an intramuscular combination of pethidine, promethazine and chlorpromazine) was reserved for either very young or uncooperative patients[3]. The probe was introduced manually into the rectum and withdrawn at increments of 0.5 to 1.0 cm (station pull-through) until a high pressure zone of anal pressure was reached. Observations were then made of the internal anal sphincter response to rectal distension. The presence of normal rectosphincteric reflex was defined when rythmicity of internal sphincter contractility was totally inhibited by rectal distension accompanied by a simultaneous drop in internal sphincter pressure to more than 75 per cent below the base-line pressure. A child was considered to have normal rectosphincteric function when at least 3 successive normal reflexes were demonstrated in a single examination.

Manometric studies for the diagnosis of HD were assessed for accuracy, sensitivity, specificity, and positive and negative predictive values using aganglionosis on full thickness rectal biopsy as the reference point.

RESULTS

Of 46 children, a definite diagnosis of HD was made in 31. Fourteen were shown not to have HD and one had internal anal sphincter (IAS) achalasia. The boy with IAS achalasia was 6 years old and had chronic constipation with an absence of RSR on anorectal manometry but the presence of ganglion cells on full thickness rectal biopsy. He recovered completely from constipation by internal sphincter myectomy. This patient was excluded from further analysis since IAS achalasia has no relationship to HD proper and, more importantly, the diagnosis of IAS achalasia relies on the discordant result of anorectal manometry and rectal biopsy[4]. The details of demographic and clinical data of 45 children are summarized in Table 1. It is noted that the majority of HD in this study were male and up to 26 per cent (8/31) of the total had clinically atypical HD at presentation. Mean anal canal pressure of children with HD and those without was 38.7 ± 14.2 mmHg and 42.4 ± 20.9 mmHg, respectively, not significantly different ($p = 0.59$, by Student's *t*-test).

Table 1. Clinical data of 45 children with chronic constipation.

	HD (n = 31)	Non-HD (n = 14)
Male/Female	24/7	5/9
Age at manometric test (mos)		
Mean (range)	12.4 (0.2-75)	13.5 (0.7-120)
Newborn (No., %)	10 (32.2)	3 (21.4)
Clinically typical HD (Gr I)	23	2
Clinically atypical HD (Gr II)	8	12
Anal canal pressure (mmHg)*		
Mean ± SD (range)	38.7 ± 14.2 (13-64)	42.4 ± 20.9 (21-91)

* Not significantly different between HD and non-HD (p = 0.59, by Student's *t*-test).

Table 2. Anorectal manometry of children with HD and those without.

		HD (n = 31)	Non-HD (n = 14)
Rectosphincteric reflex			
Presence		0	12
Absence		31	2
Accuracy	95.5%		
Sensitivity	100%		
Specificity	85.7%		
Positive predictive value	94%		
Negative predictive value	100%		

Table 3. Barium enema of children with HD and those without.

		HD (n = 31)	Non-HD* (n = 14)
Transition zone			
Presence		21	0
Absence		10	5
Accuracy	72.2%		
Sensitivity	67.7%		
Specificity	100%		
Positive predictive value	100%		
Negative predictive value	33.3%		

*Barium enema was not performed in 9 children (see text).

Accuracy of anorectal manometry and barium enema in the diagnosis of HD : Results of anorectal manometry are shown in Table 2. Of the 31 children with HD, none had RSR. However, the false positive absence of reflex was observed in 2 who had normal rectal biopsies, thus, giving an overall diagnostic accuracy by anorectal manometry of 95 per cent and diagnostic sensitivity of 100 per cent. The two patients with false-positive manometric results consisted of one premature infant (36th gestational week) who was considered to have HD on initial clinical presentation but was later proven to have congenital hypothyroidism and a 1.5 month-old infant having transient delayed intestinal transit. Our results also demonstrated that the presence of RSR on manometric study could exclude HD with 100 per cent confidence, as there were no false-negative results observed in children with HD.

Table 3 shows the results of the barium enema. It was found that a roentgenographic transition zone, a hallmark radiographic diagnosis of HD, was not demonstrated in up to 30 per cent (10/31) of children with HD. Of 14 non-HD, 9 were the children in whom the RSR was observed, as well as having an improvement of constipation with medical treatment on follow-up. Barium enemas were not performed in these children. The remaining 5 underwent a barium enema during which a transition zone was not demonstrated. When we considered the diagnostic efficacy, it is noteworthy that the accuracy and sensitivity for the transition zone of barium enemas to detect HD was very low when compared to anorectal manometry, namely, 67.7 per cent *vs* 100 per cent and 72.2 per cent *vs* 95.5 per cent, respectively. Assuming that the total 14 non-HD who underwent barium enema all showed normal findings, the diagnostic efficacy of barium enema remained unsatisfactory (77% accuracy, 67.7% sensitivity, 100% specificity, 100 per cent positive predictive value and 58.3 per cent negative predictive value).

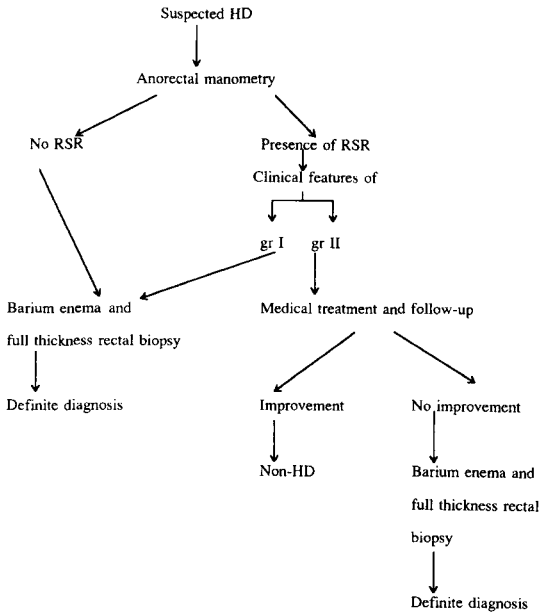

Fig. 1. Diagnostic approach in patients supected of having Hirschsprung's disease.

DISCUSSION

Anorectal manometry, a physiologic evaluation of anorectal function, is a well established technique for diagnosis of HD. To date, the diagnostic accuracy of this procedure has been reported as varying from 67 to 90 per cent[5-7]. In our experience, a high of 95.5 per cent accuracy in detecting HD by anorectal manometry was reached. In addition, the presence of RSR by this test was very reliable in excluding HD (Table 2). Although it may be considered that the number of subjects in this study were relatively small, the manometric results described herein could be reasonably applied to clinical practice because all subjects recruited in this study were strongly suspected of having HD, and a diagnostic investigation was indicated.

The accuracy of anorectal manometry depends on factors such as the examination technique, the cooperation of the child during study, and the age group of the patient. We believe that the high degree of diagnostic accuracy of this study results from the elimination of possible sources of error related to technical problems. Firstly, we used sedation in all uncooperative children as restlessness or crying at examination may easily introduce a false recording due to a displacement of the probe from the relaxation zone of the anal canal. Moreover, voluntary contraction of the external sphincter affects anal pressure and becomes superimposed on internal anal sphincter activity. On the other hand, sedation has been suggested as having no effect on the physiology of anorectal function[3,8]. Secondly, we gave cleansing enemas to the patients before the study. The presence of faeces in the rectum probably does not allow a symmetrical distension of the rectal wall, and in such cases the distorted balloon inflation may induce a displacement of the probe side hole from the sphincteric zone, but return to the correct position during balloon deflation. Such motion produces a pressure drop and recovery on the manometric recording that is identical to the RSR and could be misinterpreted.

Varying types of manometric techniques, namely, an open fluid perfused catheter, an anal balloon filled with either air or with water, and a microtransducer catheter, have been used for evaluating anorectal function. To date, the question of whether the use of different manometric techniques may explain these different results has yet to be answered since there has been no study to compare the diagnostic accuracy of those different devices. Some investigators have suggested that microtransducer catheters offer more advantages than other techniques, due to the less physiologic disturbance to anorectum and higher sensitivity[8,9]. However, our results indicate that an open fluid perfused manometric system is accurate and reliable for diagnosing HD.

In the neonate, the accuracy of manometric diagnosis for HD appears to be doubtful. Menieur et al reported the manometric diagnosis was wrong in 26 per cent of their neonate subjects[10]. Davies et al showed that an absence of RSR was demonstrated in only 43 per cent of the neonates with aganglinosis[5]. These results contrasted to those of Verder et al, who found that anorectal manometry was accurate in 21 of the 22 neonates studied[11]. The diagnostic uncertainty in the neonate is thought to be the result of physiologic immaturity of anorectal function[12], a relatively short anal canal which may result in loss of contact of the anal probe with the high pressure zone during examination, and varying manometric techniques[9,10]. In the present study, we were accurate in 12 of the 13 neonates studied, giving an accuracy of 92 per cent.

It is well accepted that the early diagnosis and treatment of HD decreases the risk of morbi-

dity and mortality from enterocolitis. However, the definite diagnosis of HD requires full thickness rectal biopsy procedure which is invasive and performed under general anesthesia. Thus, a non invasive and reliable screening test is very helpful in providing additional information to a physician in determining the cause of constipation before considering full thickness rectal biopsy. Traditionally, the barium enema has been widely used as an initial test in children suspected of HD but it is fraught with inaccuracies. As we have demonstrated in this study, the sensitivity and accuracy of barium enema was only 67.7 per cent and 72.7 per cent, respectively, and 10 out of 31 children with HD would have been missed if diagnostic screening had stopped with barium enema. Our results were in agreement with those of Taxman et al[13]. Their study included 58 infants and reported that 80 per cent of HD had a transition zone, while 20 per cent did not. Despite its inaccuracy, barium enema is still helpful for surgeons to estimate the length of aganglionic segment, which helps by determining the appropriate operation. Rectal suction biopsy is another recommended screening test for HD. This procedure requires no anesthesia and can be conveniently performed on an outpatient basis. However, inadequacy of sampling tissue biopsy has rendered routine H & E preparations difficult for evaluating submucous ganglions. It failed to reveal ganglion cells in 39 per cent of children without HD[14]. There has been an effort to solve the problem of inadequate specimen harvesting by the use of acetylcholinesterase histochemistry technique in which only the lamina propria mucosa is required. Using this technique, HD can be diagnosed from increased submucosal nerve plexus and does not require the absence of ganglion cells. Therefore, it seems that the problems related to inadequate rectal biopsy can be eliminated. However, discordant results are still obtained. Hamoudi et al[15] described up to 29 per cent of false-negative reactions in children with HD, whereas Lake et al [16] and Ikawa et al[14] reported that a nearly 100 per cent diagnostic accuracy was achieved with acetylcholinesterase histochemistry. Possible sources of discrepancies are the variability in the biopsy site, the size and quality of the biopsy material, the technical variations in performance of the stain, and the experience of individual pathologists. Additionally, technical difficulty is another potential disadvantage of the acetylcholinesterase staining method.

In our experience, anorectal manometry is a highly accurate screening test when carefully performed with patient cooperation. Recently, some investigators have suggested an order of evaluation of constipated children: First by anorectal manometry, and following that with rectal suction biopsy in cases where RSR was not present, before considering surgery[7,14]. Our studies support these guidelines. However, our results also suggest that a rectal suction biopsy may be unnecessary in the screening plan since most children in whom the RSR was not demonstrated in the initial manometric test were later proven to have HD by full thickness rectal biopsy. Moreover, the presence of RSR by anorectal manometry was 100 per cent reliable in excluding HD. In summary, anorectal manometry is simple and safe, has a high accuracy and should be recommended as the screening test of choice for HD.

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ความแม่นยำของ Anorectal Mamometry ในการวินิจฉัยโรค Hirschsprung

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จากประสบการณ์การใช้ anorectal manometry เพื่อตรวจหา rectosphincteric reflex (RSR) ในผู้ป่วยเด็กซึ่งสงสัยเป็นโรค Hirschsprung จำนวน 46 ราย พบว่าในผู้ป่วย 33 ราย ซึ่งตรวจไม่พบ RSR 31 ราย ได้รับการวินิจฉัยยืนยันเป็นโรค Hirschsprung จากพยาธิสภาพ (full thichness rectal biopsy) และ 2 รายที่เหลือไม่เป็นโรค Hirschsprung ในผู้ป่วย 12 ราย ซึ่งพบ RSR ทุกรายไม่เป็นโรค Hirschsprung

การใช้ anorectal manometry ให้ความแม่นยำ (diagnostic accuracy) ถึงร้อยละ 95 และมีความไว (sensitivity) ร้อยละ 100 ดังนั้นวิธีดังกล่าวจึงเหมาะสมมากสำหรับการตรวจกรองในผู้ป่วยที่สงสัยเป็นโรค Hirschsprung เพราะมีความแม่นยำสูงและมีความปลอดภัย (non-invasive)

คำสำคัญ : Anorectal Manometry, การวินิจฉัย, โรค Hirschsprung

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