Original Article

Rectal Punch Biopsy for Hirschsprung's Disease: Diagnosis and Complications

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Abstract

Background: *Hirschsprung disease (HD) is a congenital disorder characterized by the absence of ganglion cells at the Meissner's plexus (submucosa) and Auerbach's plexus (muscularis) of the terminal rectum that extends in a variable distance proximally. This leads to a lack of gut motility, stool stasis and constipation. Full thickness biopsy is the gold standard for the diagnosis of HD. This study aims to evaluate the efficacy of rectal punch biopsy in the diagnosis of Hirschsprung's disease.* ***Materials and Methods:*** *This cross-sectional observational study was carried out in the department of pediatric surgery, Mymensingh Medical College Hospital, Mymensingh from January 2018 to April 2019. Sixty (60) cases of clinically, radiologically consistent with Hirschsprung's disease not in acute stage, chronic constipation with assisted defecation in older children and children who came with colostomy for subsequent management without tissue diagnosis were purposively included in the study. Patients' information's were obtained in predesigned questionnaire after obtaining written consent of the parents/guardians in the consent form. All the patients underwent rectal punch biopsy. Study results were expressed as mean ± SD, frequency and percentage.* ***Results:*** *The age range of the study group was 3 days to 7 years with a mean age of 1.20±1.90 years. Most of the patients were in neonatal period (25, 41.66%) followed by infants (18, 30%). Regarding punch biopsy, the adequate sample was 59 (98.31%) and the inadequate sample was only 1 (1.69%). Among adequate tissue material HD 47 cases, non-HD 11 cases and suspicious cases were in 1 case. Only 1 (1.66%) case was presented with hemorrhage after punch biopsy which was later managed. Sensitivity, specificity, Positive predictive value, Negative predictive value and efficiency of the rectal biopsy were 100%, 96.66%, 97.8%, 100% and 98.2% respectively.* ***Conclusion:*** *Rectal punch biopsy is a simple, safe and effective procedure for diagnosing Hirschsprung's disease, offering high accuracy and a low risk of complications.*

Keywords: Hirschsprung's disease, Rectal Biopsy, Complications.
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Introduction

Hirschsprung's disease (HD) is a developmental disorder of the enteric nervous system characterized by absence of ganglionic cell in the myenteric and submucosal plexus along a variable portion of distal intestine and aganglionosis process is nearly always continuous and interrupted started at the anus[1]. Within the first 24 hours of birth, 98.5% of full-term newborn infants pass their first stool and the remaining infants do so within the next 24 hours. If the newborn infant did not pass meconium within 48 hours of birth, typically they present Hirschsprung's disease with presence of a normal looking anus[2]. Hirschsprung's disease can manifest in neonates as intestinal obstruction or in older children as chronic constipation. In neonate early diagnosis, appropriate treatments are necessary to prevent morbidity and mortality from various complications. In older children a simple, safe and reliable method is desirable to distinguish those with Hirschsprung's disease from a much larger number with functional constipation[3].

Hirschsprung's disease (HD) was first described in 1691 by Frederik Ruysch, a Dutch anatomist and surgeon, as a phenomenon associated with very dilated colon disorders[4,5]. Nonetheless, at the German Pediatric Society conference in Berlin in 1886, the disorder was given its name in honor of Harald Hirschsprung[5,6]. HD, another name for congenital aganglionic megacolon, which produces a functional obstruction and proximal dilatation of the affected segment[7,8]. It is categorized into 2 types

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according to the extent of innervation. The short segment, which only affects the rectosigmoid region of the gut, is the most common and is seen in 80-85% of patients with HD. In up to 20% of long segment disease cases, aganglionosis extends proximally to the sigmoid colon. Aganglionosis of the entire colon is less common, affecting only 3%-8% of HD patients[9]. Another rare variant is ultra-short segment disease, affecting only the distal rectum (≤2 cm)[9-11].

Abdominal distension, vomiting, and delayed meconium passage are the traditional triad of symptoms associated to HD. This symptom is observed in 80% to 90% of patients affected by this condition[12]. A digital examination may reveal a tight anal sphincter and an explosive bowel movement. Many diagnostic modalities are now available for the evaluation of Hirschsprung's disease in paediatric surgical practice. Diagnostic workup should be based on clinical presentation and include only those modalities that will provide pertinent information and will occasion the least discomfort. These include imaging studies and biopsies[13]. The critical diagnostic step in Hirschsprung's disease is obtaining proof of its criteria. No logical treatment plan can be formulated until an accurate diagnosis is made and this often requires biopsy[14]. The biopsy ideally should be performed at a facility that can provide complete patient care. The type of biopsy performed must be tailored to the lesion so that adequate tissue is obtained without prematurely removing important structures or deforming the patient[15].

A full-thickness rectal biopsy is regarded as the gold standard for diagnosing Hirschsprung's disease. Full thickness biopsy includes mucosa, sub mucosa and muscle layers[15]. Fisher and Gherardi first described the method of full thickness biopsy[16,17]. The advantage of this procedure is that it provides sufficient tissue for histopathological examination and allows for the diagnosis of other neurocristopathies. Disadvantages are bleeding, scarring and need for general anaesthesia[18]. Rectal suction biopsy is another method for diagnosis of HD. Dobbins and Bill 1965 and in 1969 Noblett introduced rectal mucosa including sub mucosa for histopathological analysis. It is easy, safe, with no need for anaesthesia and done at bedside[19]. Rectal punch biopsy (RPB) was first described in 1972[20]. It is simple, safe, with no need for anaesthesia, done on bedside with a simple biopsy forceps or laryngeal biopsy forceps with the help of proctoscope, nasal speculum or handmade biopsy tube. In punch biopsy only rectal submucosa is needed and obtained easily[14].

Diagnostic accuracy of the biopsy specimens depends on the adequacy and number of the specimens, levels at which they are obtained, and a skilled pathological team including the technician. When all these criteria are met diagnostic accuracy is as high as 99.78%. In fulfillment of these criteria, punch biopsy is considered the most effective method for diagnosing Hirschsprung's disease[21]. This type of study was done previously in our context. This study aims to assess the effectiveness of rectal punch biopsy in diagnosing Hirschsprung's disease and compares its findings with previous studies in terms of variation, consistency, and reinforcement.

Materials and Methods

This cross-sectional study was conducted in the Department of Pediatric Surgery at Mymensingh Medical College Hospital, Mymensingh, between January 2018 and April 2019. Sixty (60) cases of suspected HD were purposively selected for this study fulfilling the exclusion & inclusion criteria. Ethical approval of the study was taken from the Ethical Review Board (ERB) of MMCH (ref: MMC/IRB/2018/70). Clinically, radiologically consistent with HD not in acute stage, chronic constipation with assisted defecation in older children and children who came with colostomy for subsequent management without tissue diagnosis were included in the study. Patients with clinically HD with acute intestinal obstruction who needed immediate surgical intervention and whose legal guardian will not provide valid consent were excluded from the study. For each case, patient information was collected using a predesigned questionnaire after obtaining written consent from the parents or guardians. All the patients underwent rectal punch biopsy and specimen were sent to histopathological examinations. The biopsy was performed in the lithotomy position without anesthesia. A biopsy tube was inserted into the rectum, with the side hole directed toward the posterior wall of the rectum. Post biopsy we follow up every patient for any per-rectal bleeding, abdominal distension, temperature or any local infection. All patients' guardians were advised not to use any suppository or enema for 48 hours.

All data were processed and analyzed using Microsoft Excel and IBM-SPSS v23.0 for Windows. Statistical inference was based on 95% confidence interval and p-value <0.05 was considered statistically significant. Variables were presented as mean ± standard deviation (SD), frequency, and percentage. The summarized data were presented in the form of tables and figures.

Results

In this cross-sectional observational study, a total of 60 patients who were clinically and radiologically consistent with Hirschsprung's disease (40 males and 20 females) and not in an acute condition were

included. The age range of the study group was 3 days to 7 years with a mean age of 1.20 ± 1.90 years. Among the 60 patients' maximum patients were in neonatal period 25 (41.66%) and 18 (30%) were infants and 17 (28.33%) were older children (Figure-1). Table-I shows adequacy of punch biopsy in study population, total adequate sample were 59 (98.31%) and inadequate sample was only 1 (1.69%). Among adequate tissue material HD 47 cases, non-HD 11 cases and suspicious cases were in 1 case. In the neonatal period 22 (36.66%) were HD, 2 (3.33%) were non-HD, 1 (1.66%) was suspicious case. In infant period 13 (21.66%) were HD, 5 (8.33%) were non-HD. In child period 12 (20%) were HD, 4 (6.66%) were non-HD and 1 (1.66%) was inadequate case (Table-II). Table-III shows complication of rectal punch biopsy in study population where only 1 (1.66%) case was presented with hemorrhage after punch biopsy. Table-IV showed the accuracy of punch biopsy in study population. Among the 60 cases true positive was 46 and true negative was 11 cases. Besides false positive result was found in 1 case and there was no false negative result. Sensitivity, specificity, Positive predictive value, Negative predictive value and efficiency of the test were 100%, 96.66%, 97.8%, 100% and 98.2% respectively.

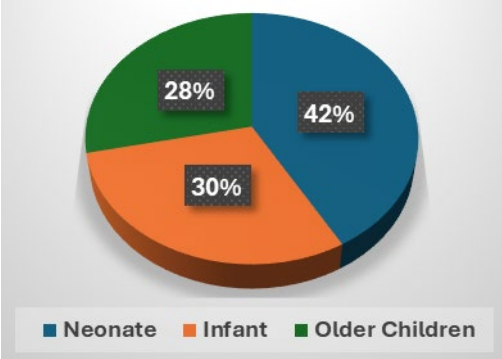

Figure-1: Age distribution of the study cases (n=60)

Table-I: Adequacy of punch biopsy in study population (n = 60)

Adequacy (n)	HD	Non-HD	Suspicious
Adequate (59)	47 (78.33%)	11 (18.33%)	1 (1.66%)
Inadequate (1)	1 (1.69%)		

Table-II: Result of punch biopsy among study population in relation to age of the patient (n=58)

Age Group	HD	Non-HD	p-value
Neonate	22 (36.66%)	2 (3.33%)	p=0.001 (significant)
Infant	13 (21.66%)	5 (8.33%)	
Older Children	12 (20%)	4 (6.66%)	

Table-III: Complication of the rectal punch biopsy in the study population (n = 60)

Types of Complication	Haemorrhage	Infection	Perforation	Pelvic Haematoma
Number of Patients	1 (1.66%)	00	00	00

Table-IV: Accuracy of punch biopsy in the study population exclude suspicious and inadequate specimen (n = 58)

Accuracy		Distal Rectal Tissue	Full Thickness Rectal Tissue
		+	-
Punch Biopsy	+	46	11
	-	1	0

Discussion

Hirschsprung's Disease (HD) is a challenging surgical condition in children, particularly in neonates, as it leads to neonatal intestinal obstruction. Definitive diagnosis and appropriate treatment are mandatory. Surgery is considered the definitive management for patients with Hirschsprung's disease[22]. Treatment formulated by tissue diagnosis prior to definitive surgery. In this regard punch biopsy is designed as first line tissue diagnosis in this study. Present Study shows a total of 60 patients were undergoing punch biopsy who were clinically and radiologically consistent with HD. In each case we took multiple biopsy samples that were stained by Hematoxylin and Eosin for histopathological examination. The main challenge in interpreting biopsy samples for diagnosing HD is the presence of negative histological findings. It needs an expert technician for tissue processing and expert pathologist to interpret and for that the search for a positive parameter needs to the use of immunohistochemical markers to demonstrate abnormal innervation and abnormal diameter nerve trunks. In our institution it is still not available, so we still absolutely depend on Hematoxylin and Eosin.

All 47-punch biopsy proven HD cases performed definitive procedure for reconfirmation of diagnosis. A total of 46 patient diagnosis as HD after definitive procedure. During the definitive procedure distal rectal tissue was taken and sent for histopathological examination. One patient diagnosed as HD after punch biopsy procedure but after the definitive procedure diagnosed as non-HD. This may be due to pathological error or ganglion cell morphological difference. In our study, one (1) patient was diagnosed with HD after laparotomy and labelling biopsy with transverse colostomy. Total 11 (eleven)

punch biopsy not proven HD underwent full thickness rectal biopsy to see accuracy of punch biopsy and diagnosed as non-HD. After punch biopsy procedure, total inadequate sample is one (1) due to absence of submucosa. The patient underwent a full-thickness rectal biopsy, which confirmed the diagnosis of Hirschsprung's disease. After punch biopsy procedure suspicious case was one (1) who was later confirmed as non-HD in full thickness biopsy. In this patient histology showed presents of few ganglion cells thickened nerve fiber. In a study by Pease, et al.[23] showed that 35 patients were diagnosed with HD and 23 patients were diagnosed as non-HD. Based on the pathological findings from the study by Alehossein et al.[24], 36 children (65.4%) were diagnosed with HD, while 19 (34.6%) were found to have non-HD. Besides, Yoshimaru, et al.[25] in their study showed 289 patients were diagnosed with HD and 569 patients diagnosed as non-HD due to larger sample size and long duration. This is generally consistent with the findings of our study.

Punch biopsy obtaining tissue in this study was adequate in 98.02% and inadequate in 1.69%. Cases with insufficient biopsy specimens underwent a full-thickness rectal biopsy, which confirmed the diagnosis of HD. Inadequate sample taken to anyone that was little, or no submucosa or biopsy was like stratified squamous or columnar epithelium. In this study, one (1) case had little submucosa. In Hiroshe, et al.[14], study inadequate sample was 11.2% which is more in older children due to difficult in obtaining an accurate biopsy specimen, because of morphological change such as an increase in the thickness of the rectal wall, mucosal oedema and fibrous tissue, who received chronic enema. This is not in line with our study. After punch biopsy procedure complications occurred in one (1) case only. This case developed post biopsy continues per rectal oozing. Then oozing was stopped by application of soaked gauze rectal pack and blood transfusion. Another study found that two (2) patients experienced complications after the rectal biopsy: one had hemorrhage, while the other developed pelvic hematoma[23]. In this study, no additional complications such as perforation, infection, or abscess were observed. Hiroshe, et al[14]. also found no complications in their study.

The accuracy of punch biopsy in this study was measured by sensitivity, specificity, positive predictive value (PPV), negative predictive value (NPV) and efficiency of the test. Sensitivity, specificity, PPV, NPV and efficiency of the test were 100%, 96.66%, 97.8%, 100% and 98.2% respectively. In another study, the sensitivity and specificity of diagnostic scoring system were 88.9% and 84.2% respectively. Moreover, the positive and negative predictive values were 91.4% and 80%, respectively[24]. Tang et al.[26] found that rectal section biopsy was effective in confirming Hirschsprung's disease, showing a sensitivity of 89%, specificity of 83%, and both positive and negative predictive values of 89% and 83%, respectively. These findings are nearly consistent with our study. Several factors such as age, sex and disease duration may also be important in the prediction of sensitivity and specificity of rectal biopsy in patients with older ages.

Limitations of the Study

There were some limitations to this study, including its small sample size, short study duration and single institutional design.

Conclusion

Rectal punch biopsy is a simple, safe and effective procedure for diagnosing Hirschsprung's disease, offering high accuracy and a low risk of complications. Early detection and diagnosis of the disease are expected to improve the chances of timely surgical intervention and reduce the risk of severe complications in children.

Recommendation

To strengthen future research, studies should include a larger sample size, extend the duration of observation, and involve multiple institutions to improve the generalizability and reliability of the findings.

Conflict of interest

The authors declared that they have no conflicts of interest.

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