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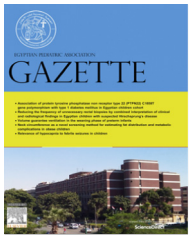

Reducing the frequency of unnecessary rectal biopsies by combined interpretation of clinical and radiological findings in Egyptian children with suspected Hirschsprung's disease

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KEYWORDS

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Abstract *Introduction:* Hirschsprung's disease (HD) should be considered in children with neonatal-onset constipation. Clinical differentiation between HD and idiopathic constipation (IC) is difficult in late presenting infants. Consequently, paediatric surgical centres receive numerous referrals for rectal biopsies, requiring admissions and GA, particularly if suction biopsy is unavailable, and in older children.

Methods: Forty-two cases referred for rectal biopsy, were studied for clinical features, single contrast enema, as compared to rectal biopsy findings, to determine the statistical reliability towards achieving a diagnosis.

Results: The mean age at presentation was 106 days in HD patients, and 172 days in IC. Significant neonatal clinical features were present in 54%. Delayed passage of meconium was present in 86% of HD, compared to 14% of IC ($p = 0.001$). Rectal examination found a tight segment in 90% of HD, and a distended anorectum in 64% of IC ($p = 0.005$). The sensitivity of contrast enema was 86%,

Abbreviations: HD, Hirschsprung's disease; IC, idiopathic constipation.

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and the specificity was 90%. The histological analysis of strip rectal biopsy was sensitive in 93%, and inconclusive in 7%.

Conclusion: This audit generated a checklist of 6 clinical and 3 radiological criteria, to differentiate HD from ID, including *clinically* (1) neonatal onset; (2) male sex; (3) congenital anomalies, dysmorphic features and/or family history of HD; (4) delayed meconium passage; (5) enterocolitis or significant bowel obstruction/impaction; (6) tight segment on rectal examination; and *radiologically* (7) funnelled transition zone or a reversed rectosigmoid index (<1); (8) delayed evacuation of contrast after 24 h; and (9) absent distension of the anorectum with contrast, absent mucosal irregularities, and absent sigmoid looping.

Introduction

Hirschsprung's disease (HD) remains a challenging diagnosis to the paediatric surgeon. It should be considered in any early-onset constipation and difficult passage of stools. It is generally an isolated disorder of full-term otherwise healthy infants.[1]

The absence of ganglion cells from the distal intestine is the hallmark of the disease. Ganglion cells are missed from both the submucosal and inter-muscular nerve plexuses. In addition, there is a marked increase in the nerve fibres extending into the submucosa. Aganglionosis typically extends up to the recto-sigmoid region in approximately 80% of cases.[2]

It has been reported that 90% of HD patients could be diagnosed before the age of one month, and if not during the neonatal period, mostly by the age of one year.[3,4] Classic rectosigmoid segment HD has a male to female ratio of 4:1,[5] but this male to female predilection declines in the more proximal forms, compared to the classic pattern.[6]

Most HD cases present with delayed passage of meconium. Some reports stated a wide range of the incidence of this finding, varying between 60% and 94%.[4] It may surprisingly fail to differentiate HD from idiopathic constipation (IC), as some reports found it in only 65% of HD patients, versus 13% in IC patients.[3]

Chronic constipation, dependency on laxative suppositories, straining during defecation, noticeable abdominal distension, hard stools, and occasionally blood-streaked stools are features of both HD and IC, and it could be difficult at times to differentiate clinically between both groups without further investigation. It could be generally stated that infants presenting beyond the neonatal period have a higher incidence of not being HD, hence their dietary and medical management should be attempted thoroughly.[7]

An early diagnosis of HD is important as there is a present consensus of operating on HD during the neonatal period or shortly after, to avoid the myriad of complications that occur in untreated cases, as well as the general concept that the operation could be technically easier in neonates.[9]

The rectal biopsy, traditionally a full-thickness biopsy, is the gold standard for the diagnosis of HD. Suction rectal biopsy has been widely adopted, and is reported to have an accuracy of more than 90%.[9] Yet, when the diagnosis cannot be made by suction rectal biopsy, a full-thickness posterior rectal wall biopsy should be performed in the operating room. The pathologic evaluation of a suction biopsy for the diagnosis of HD is considerably more difficult than that of full thickness biopsy and needs a highly skilled pathologist.[10]

There have been some attempts at studying the sensitivity and specificity of the various clinical data and noninvasive investigations aiming at reducing the performance of unnecessary rectal biopsies, which are believed not to be without complications. This hypothesis was based on reports that only 12–17% of biopsies performed on constipation patients yielded a positive result of aganglionosis and that up to 80% of biopsies could be unnecessary.[10,11]

As contrast enema is a non-invasive diagnostic tool, the presence of suggestive enema findings associated with a classic clinical presentation is pathognomonic, and warrants a surgical referral and involvement. It is the opposite situation that may need a suggested algorithm, or a scoring system, when both the clinical and contrast enema findings are equivocal.

This current work analysed the statistical significance of the available clinical and radiological parameters, and investigated the reliability of these diagnostic tools. We aim not at substituting absence of ganglion cells in a rectal biopsy, as the sole indication to do a pull-through operation, but rather to properly select the patients that need a rectal biopsy within the context of management of resources for very busy paediatric surgical services in the developing world.

Methods

Forty-two patients presenting with chronic persistent constipation, referred to the paediatric surgeons for a rectal biopsy, after having a contrast enema, were retrospectively studied. The patients were enrolled over a period of 6 months from January 2013 to June 2013.

We statistically analysed the clinical assessment of the enrolled patients including the age at presentation, neonatal onset of symptoms, consanguinity, maturity at birth, antenatal history, associated congenital anomalies, delayed passage of meconium, nutritional status, vomiting, feeding difficulties, features of enterocolitis or significant bowel obstruction, abdominal distension, and the digital per-rectal examination.

Similarly we statistically analysed the radiological features of the contrast enema including the presence of a funnel-shaped transition zone, recto-sigmoid index (ratio of the rectal diameter to sigmoid diameter, normally $\geqslant 1$, and considered reversed and thus suggestive of HD if <1), residual contrast in the colon after 24 h, distension of the anorectum with contrast, abnormal colonic contractions or spasms, mucosal irregularities, serrations, sigmoid looping and redundancy. The interpretation of the contrast enema was done by both an