PROSPECTIVE SURGICAL AND RADIOLOGICAL ASSESSMENT IN THE DIAGNOSIS OF PEDIATRIC HIRSCHSPRUNG DISEASE.

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ABSTRACT:-

Background: Hirschsprung disease (HD) is a developmental disorder characterized by absence of ganglia in the distal colon, resulting in a functional obstruction. It is a common cause of pediatric intestinal obstruction. Objective of present study was to evaluate a checklist of radiologic and clinical signs to determine the probability of HD in suspicious patients.

Methods:

In a diagnostic accuracy study, 19 children with clinical manifestations of HD attended pediatric OPD in a tertiary care teaching hospital, SSIMS, BHILAI from January 2022 to December 2024 were assessed. A checklist was used to evaluate the items proposed by contrast enema (CE), based on six subscales, including transitional zone, rectosigmoid index (RSI), irregular contractions in aganglionic region, cobblestone appearance, filling defect due to fecaloid materials and lack of meconium defecation during the first 48 hours after birth. The patients were classified as high score and low score. Sensitivity and specificity were calculated for identifying HD, in comparison with pathologically proved or ruled out HD.

Results:

Of the 19 patients, 11 (57.89%) cases had HD and 08 (42.11%) cases were without HD. The mean age was 2.793 ± 4.21 months. Abdominal distension, lack of meconium defecation, and constipation were the most common clinical symptoms with frequencies of 15 (78.9%), 11 (57.8%), and 14 (73.68%), respectively. 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 59.09%, 49.99%, 59.09% and 56.06% respectively.

Conclusion:-

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 histological confirmed non-HD patients are 68.75%, 81.25%, 87.5% and 79.17% respectively. This would in turn give an overall mean specificity rate of 79.17% in successfully excluding HD with the above mentioned radiological signs from the contrast enema.

INTRODUCTION:-

Hirschsprung disease (HD) is a congenital disorder characterized by the absence of ganglion cells in the distal bowel, resulting in functional intestinal obstruction. It most commonly presents in the neonatal period but may also be diagnosed later in infancy or early childhood, especially in milder forms. Early and accurate diagnosis is critical prevent in milder forms. Early and accurate diagnosis is critical to prevent complications such as enterocolitis, failure to thrive, and chronic constipation. The diagnosis of HD traditionally relies on a combination of clinical presentation, radiological imaging, and histopathological confirmation via rectal biopsy. Radiological methods, particularly contrast enema studies, play a pivotal role in the initial evaluation by identifying key features such as a transition zone between the aganglionic and normally innervated bowel. However, imaging findings can sometimes be inconclusive or operator-depende., making Surgical assessment, including rectal suction biopsy or full-thickness biopsy, remains the gold standard for definitive diagnosis, allowing direct visualization of ganglion cells and acetylcholinesterase activity. The integration of both radiological and surgical approaches is therefore essential to ensure timely and accurate diagnosis. This prospective observational study aims to evaluate the diagnostic utility and correlation of radiological findings with surgical and histopathological outcomes in children presenting with clinical features suggestive of Hirschsprung disee. By assessing diagnosis. This prospective observational study aims to evaluate the diagnostic utility and correlation of radiological findings with surgical and histopathological outcomes in children presenting with clinical features suggestive of Hirschsprung disease. By assessing the effectiveness of these complementary diagnostic tools, the study seeks to support evidence- based diagnostic protocols and improve early detection and management of HD.

METHODS

From January 2022 to December 2024, neonates and infants, and children with clinical suspicion of Hirschsprung's disease were enrolled in this prospective observational study that was approved by the institutional ethics committee of a tertiary care teaching hospital, SSIMS Junwni Bhilai

Inclusion criteria

- Delayed passage of meconium (beyond 48 hours) ·
- Patients with defecation problems since birth and abdominal distension.

Exclusion criteria

- The modified Bell staging criteria in which a composite of clinical signs and symptoms (eg, abdominal distention, bloody stools, or hypotension), biochemical parameters (eg., thrombocytopenia or neutropenia), and radiographic signs (eg, pneumatosis or pneumoperitoneum) was used to grade the severity of NEC.29
- Abdominal radiograph showing multiple air fluid levels.

Inclusion criteria were having clinical presentation highly suspicious for HD, performing a CE exam and full thickness biopsy. Written and verbal informed consent was taken from the parents who satisfied the inclusion criteria to undergo further investigations. Data on gestational age and first passage of meconium after birth

were collected. Prior to per rectal examination, all patients underwent a plain abdominal radiograph and a contrast enema.

Patients were divided into three age groups, as follows: <1 month, 1-12 months, >12 months.

Contrast enema

Radiologists performed the contrast enema with the support of a pediatrician in a routine manner using standard CE techniques. Dilute barium sulfate was administered rectally using a # 6 infant feeding tube placed just within the rectum No balloon catheters were used. All CE images were read by the same radiologist. The classical finding of a transition zone (CETZ) was considered being a positive result.

Rectal biopsy

The final diagnosis of HD was made by the absence of ganglion cells in a full thickness biopsy (FTB). Biopsy specimens were obtained at 2 cm above the dental line, posteriorly. These specimens were examined for ganglion cells with and staining hematoxylin-eosin was determined as acetylcholinesterase activity a previously described by Kamovsky and Roots.30 A biopsy was positive when the acetylcholinesterase activity was elevated in combination with an absence of ganglion cells.

A checklist was used to evaluate the items proposed by CE as radiologic signs, including six subscales: 1) transitional zone (TZ), defined as significant change in intestinal diameter from non-dilated to dilated section during CE; 2) RSI, ratio of largest rectal diameter to largest sigmoid diameter (among proximal, distal, and loop sections), is considered abnormal if it is <1; 3) bizarre large irregular contractions in aganglionic region, with saw teeth appearance, due to dysrhythmia, 4) cobblestone appearance or mucosal irregularity or proximal colon spasm; 5) filling defect due to fecaloid materials; 6) lack of meconium defecation during the first 48 hours after birth. For subscales 1 and 2, if they were positive, we would consider scoring = 2, and, if they were negative, we would consider scoring = 0. For the other subscales, the positive results had scoring = 1 and negative results had score = 0. Furthermore, the patients were evaluated based on scoring system, as follows: high (5-8), and low (0-4). Data analysis was performed with the SPSS version 20.0 software (IBM, USA). Chi-Square and independent t tests were used for analysis and were considered statistically significant at $P < 0.05$.

RESULTS

Nineteen patients (10 neonates, 08 infants and 01 aged for than 1 year) were included in the study, of these, 11 (57.89%) patients had histological confirmed diagnosis of HD and 08 (42.11%) patients had HD excluded by rectal biopsy. The mean age was 2.793+4.21 months, ranging of the 19 patients, 11 (57.89%) cases had HD and 08 (42.11%) cases were without HD. The mean age was 2.793 ± 4.21 months. Abdominal distension, lack of meconium defecation, and constipation were the most common clinical symptoms with frequencies of 15 (78.9%), 11 (57.8%), and 14 (73.68%), respectively. 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 59.09%, 49.99%, 59.09% and 56.06% respectively.

DISCUSSION

This prospective study evaluated the diagnostic effectiveness of radiological and surgical methods in children suspected of having Hirschsprung disease (HD), highlighting the importance of a multidisciplinary approach for timely and accurate diagnosis. Our findings reinforce the role of contrast enema as a valuable, non-invasive initial tool, while emphasizing the necessity of histopathological confirmation via biopsy for definitive diagnosis. The contrast enema demonstrated characteristic radiological features such as a transition zone, delayed evacuation of contrast, and a narrowed distal segment in a significant number of patients. These findings were consistent with those reported in previous studies, where sensitivity ranges from 70% to 90%. However, the presence of false negatives and operator-dependent variability limits the reliability of radiology alone as a standalone diagnostic tool. Surgical biopsy, whether suction or full-thickness, remains the gold standard due to its high specificity in confirming agangliosis. In our study, Standalone diagnostic tooI. Surgical biopsy, whether suction or full-thickness, remains the gold standard due to its high specificity in confirming aganglionosis. In our study, all cases confirmed histologically had corresponding radiological suspicion, but a subset of patients with inconclusive imaging still showed positive biopsy results. This underscores the limitations of relyingsolely on imaging, particularly in neonates or patients with short- segment disease where the transitionzone may be subtle or absent.Moreover, we observed that earlycombined assessement reduceddiagnostic delays and minimized the risk of complications such as enterocolitis. Prompt diagnosis also facilitated timely surgical intervention, leading to better postoperative outcomes and shorter hospital stays.Our study supports the integration of clinical judgment with radiological and surgical tools, especially in cases with ambiguous imaging. While imaging remains a vital first-line investigation, it should always be followed by biopsy in cases of persistent clinical suspicion.

LIMITATIONS

Limitations of this study include the single-center setting and a relatively small sample size, which may affect the generalizability of the results. Future multicenter studies with larger cohorts may provide more robust data to guide standardized diagnostic algorithms.

CONCLUSION

A combined surgical and radiological approach enhances diagnosticaccuracy in paediatric Hirschsprung disease. While contrast enema remains a useful initial tool, definitive diagnosis must rely on histopathological confirmation. A high index of clinical suspicion, even in face of normal imaging, remains essential for early and effective management.

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