CASE REPORT

Evaluating single incision laparoscopy-assisted extracorporeal biopsy as an alternative to frozen sections in the management of Hirschsprung disease

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SUMMARY

Hirschsprung disease (HD) is a congenital condition characterized by the absence of ganglion cells in the distal intestine, leading to bowel obstruction. While the use of frozen sections during biopsy is common practice, discrepancies with immunohistochemistry results and the unavailability of frozen section technology in certain pediatric surgical facilities in Indonesia highlight the need for alternative diagnostic approaches. This study evaluates the effectiveness of Single Incision Laparoscopy-Assisted Extracorporeal (SI-ECo) leveling biopsy with immunohistochemistry as a reliable alternative to frozen sections in the preoperative management of Hirschsprung disease, especially in facilities lacking frozen section capabilities. We present three cases of pediatric patients diagnosed with HD confirmed through rectal biopsy. Each patient underwent an SI-ECo leveling biopsy, successfully identifying the ganglionic zone by locating ganglion cells in the distal sigmoid. Based on these findings, subsequent transanal endorectal pull-through (TEPT) procedures were performed. The results demonstrated that SI-ECo leveling biopsy effectively identifies the ganglionic zone, providing a less invasive and precise method for preoperative planning. Leveling biopsy with SILS offers an effective method for identifying the ganglionic zone in Hirschsprung disease. SI-ECo reduces diagnostic discrepancies and provides higher specificity for detecting ganglion cells compared to frozen sections before the pull-through procedure. SI-ECo leveling biopsy with immunohistochemistry offers a practical, accurate, and less invasive alternative for diagnosing and managing Hirschsprung disease. It reduces the risk of discrepancies observed with frozen sections, making it a viable option for facilities without access to frozen section technology.

KEYWORDS:
Hirschsprung disease, Single Incision Laparoscopy, Extracorporeal Biopsy, Frozen Section Alternative, Pediatric Surgery

INTRODUCTION

Hirschsprung disease is a congenital disorder characterized by the absence of enteric ganglion cells in the distal intestine, resulting in functional obstruction.[1] The gold standard for diagnosis remains rectal biopsy with histopathological examination to confirm the absence of ganglion cells.[2] Traditionally, intraoperative frozen section biopsies have been used to determine the level of aganglionosis and guide surgical resection.[1] However, frozen sections have limitations, including potential discrepancies with permanent section results and the need for specialized pathology services that may not be available in all settings.[1]

Single-incision laparoscopic surgery (SILS) has emerged as a minimally invasive technique in pediatric surgery, offering benefits such as reduced postoperative pain and improved cosmesis.[3] The application of SILS for extracorporeal biopsies presents a potential alternative to frozen sections in managing HD. This approach allows for full-thickness biopsies to be obtained and processed for immunohistochemistry, which has shown higher sensitivity and specificity for detecting ganglion cells than frozen sections.

In Indonesia and other resource-limited settings, the unavailability of frozen section technology in many pediatric surgical facilities poses a significant challenge in the optimal management of HD. This highlights the need for alternative diagnostic approaches that are accurate and feasible in diverse healthcare environments.

This study aims to evaluate the effectiveness of Single Incision Laparoscopy-Assisted Extracorporeal (SI-ECo) leveling biopsy with immunohistochemistry as a reliable alternative to frozen sections in the preoperative management of Hirschsprung disease. By presenting a series of cases utilizing this technique, we seek to demonstrate its potential as a less invasive and more precise method for determining the level of aganglionosis before definitive surgery.

CASE PRESENTATION

We present three cases of pediatric patients diagnosed with HD who underwent Single Incision Laparoscopy-Assisted Extracorporeal (SI-ECo) leveling biopsy as an alternative to frozen sections. All patients were initially diagnosed with HD through rectal suction biopsy, which remains the gold standard for diagnosis.[4]

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Case 1
A 3-month-old male infant presented with a history of delayed passage of meconium, abdominal distension, and constipation since birth. The initial contrast enema showed a transition zone at the rectosigmoid junction, suggesting short-segment HD. The patient underwent an SI-ECo leveling biopsy under general anesthesia. A 10 mm incision was made at the umbilicus for camera port insertion with Single incision laparoscopic surgery (SILS).

Full-thickness biopsies were taken at 5 cm intervals, starting from the rectosigmoid junction and moving proximally. The biopsies were sent for immunohistochemistry (IHC) staining using calretinin, Bcl2, and S100 which has shown high sensitivity and specificity for detecting ganglion cells. The results confirmed the presence of ganglion cells in the distal sigmoid colon, 5 cm above the peritoneal reflection. A transanal endorectal pull-through (TEPT) procedure was performed based on these findings. The patient's postoperative course was uneventful, with regular bowel movements achieved by the 6-month follow-up.

Case 2
A 2-year-old female child presented with chronic constipation and failure to thrive. SI-ECo leveling biopsy was performed as described in Case 1. IHC staining with calretinin, Bcl2, and S100 antibodies, which have shown improved accuracy in detecting ganglion cells compared to conventional H&E staining, revealed the presence of ganglion cells in the mid-sigmoid colon.

A transanal endorectal pull-through (TEPT) procedure was subsequently performed. At the 1-year follow-up, the patient had normal bowel function and showed catch-up growth.

Case 3
A 6-month-old male infant, previously diagnosed with Down syndrome, presented with recurrent episodes of abdominal distension and enterocolitis. Rectal suction biopsy confirmed the diagnosis of HD. Given the higher risk of extended aganglionosis in patients with Down syndrome, SI-ECo leveling biopsy was crucial for the accurate determination of the transition zone.

The procedure was performed as in the previous cases, with biopsies taken at closer intervals (3 cm) due to the potential for skip lesions. IHC staining using a panel of neuronal markers, including calretinin, Bcl2, and S100, identified ganglion cells in the proximal sigmoid colon. At the 1 month follow-up, the patient had satisfactory bowel function with three to four daily bowel movements.

In all three cases, the SI-ECo leveling biopsy technique accurately identified the ganglionic zone, allowing for precise planning of the definitive pull-through procedure. IHC staining on full-thickness biopsies offered superior diagnostic accuracy compared to traditional frozen sections.

Operative times for the SI-ECo leveling biopsy ranged from 45 to 60 minutes, with minimal blood loss (<10 mL) in all cases. No intraoperative complications were observed. The single-incision approach resulted in excellent cosmetic outcomes, with the umbilical scar barely visible at follow-up visits.

The definitive pull-through procedures were performed within 1-2 weeks of the leveling biopsies. Histopathological examination of the resected specimens confirmed the accuracy of the transition zone identification in all cases, with no instances of retained aganglionosis.

This case series demonstrates that SI-ECo leveling biopsy with immunohistochemistry is a safe and effective alternative to frozen sections in the preoperative management of Hirschsprung disease. The technique allows for accurately determining the transition zone, facilitating precise surgical planning, and potentially reducing the risk of postoperative complications related to retained aganglionosis. Moreover, the single-incision approach offers the benefits of minimally invasive surgery, including decreased postoperative pain and improved cosmesis.

DISCUSSION

The management of HD has evolved significantly over the past decade, with a trend towards minimally invasive approaches and single-stage procedures. Our study evaluates the effectiveness of Single Incision Laparoscopy-Assisted Extracorporeal (SI-ECo) leveling biopsy with immunohistochemistry as an alternative to frozen sections in the preoperative management of HD. The results demonstrate that SI-ECo leveling biopsy effectively identifies the ganglionic zone, providing a less invasive and precise method for preoperative planning.

The accuracy of intraoperative diagnosis is crucial for the successful management of HD. Traditionally, frozen sections have been used to determine the level of aganglionosis during surgery. However, several studies have reported discrepancies between frozen section results and permanent section findings.[2] In our case series, SI-ECo leveling biopsy with immunohistochemistry provided consistent and reliable results, aligning with the final histopathological diagnosis in all cases.

The use of immunohistochemistry (IHC) markers has significantly improved the accuracy of HD diagnosis. Calretinin, in particular, has emerged as a highly sensitive and specific marker for the presence of ganglion cells.[5] Our study utilized a panel of IHC markers, including calretinin, CD56, and S-100, showing high sensitivity and specificity in detecting ganglion cells. This approach allows for a more precise identification of the transition zone compared to traditional hematoxylin and eosin staining.

Single-incision laparoscopic surgery (SILS) has gained popularity in pediatric surgery due to its minimally invasive nature and improved cosmetic outcomes. The SI-ECo technique combines the benefits of SILS with the accuracy of extracorporeal biopsy and IHC analysis. This approach offers several advantages over conventional multi-port laparoscopy, including reduced postoperative pain, faster recovery, and better cosmesis.[1]

One of the critical benefits of SI-ECo leveling biopsy is the ability to obtain full-thickness biopsies, which are essential for the accurate diagnosis of HD. This is particularly important in ultrashort- segment HD or when dealing with the transition zone, where partial-thickness biopsies may lead to false-negative results.[1]

Using suspension sutures in our SI-ECo technique, as described by Tran et al., facilitates better exposure and simplifies the procedure. This modification addresses some technical challenges of single-incision laparoscopy in infants and small children, making the procedure more feasible and reproducible.

In resource-limited settings, where frozen section capabilities may not be available, SI-ECo leveling biopsy with IHC offers a practical alternative. This approach allows for accurate preoperative planning without requiring specialized intraoperative pathology services. Moreover, it potentially reduces the risk of retained aganglionosis, which can occur when relying solely on frozen sections.

The long-term outcomes of patients undergoing SI-ECo leveling biopsy followed by transanal endorectal pull-through (TEPT) are encouraging. Our case series demonstrated good functional results, with no anastomotic stricture or enterocolitis during the follow-up period. These findings are consistent with other studies reporting favorable outcomes for laparoscopic-assisted approaches in HD management.

Despite the promising results, our study has limitations. The small sample size and relatively short follow-up period necessitate further investigation with larger cohorts and longer-term follow-up. Additionally, a direct comparison with frozen section outcomes in a randomized controlled trial would provide more robust evidence for the superiority of SI-ECo leveling biopsy.

Future research should focus on standardizing the IHC panel for HD diagnosis and exploring the potential of novel markers to further improve diagnostic accuracy. The role of advanced imaging techniques, such as confocal laser endomicroscopy, in conjunction with SI-ECo biopsy, may also be worth investigating.

CONCLUSIONS

In conclusion, SI-ECo leveling biopsy with immunohistochemistry offers a practical, accurate, and less invasive alternative for diagnosing and managing Hirschsprung disease. It reduces the risk of discrepancies observed with frozen sections, making it a viable option for facilities without access to frozen section technology. As minimally invasive techniques evolve, this approach may become part of the standard of care in the preoperative management of HD, particularly in resource-limited settings.

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CONSENT STATEMENT

All patients involved in this study provided informed written consent. Their identities have been kept confidential throughout the study.

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