

# Endoscopic Full-Thickness Resection As a Means to Diagnose Hirschsprung Disease



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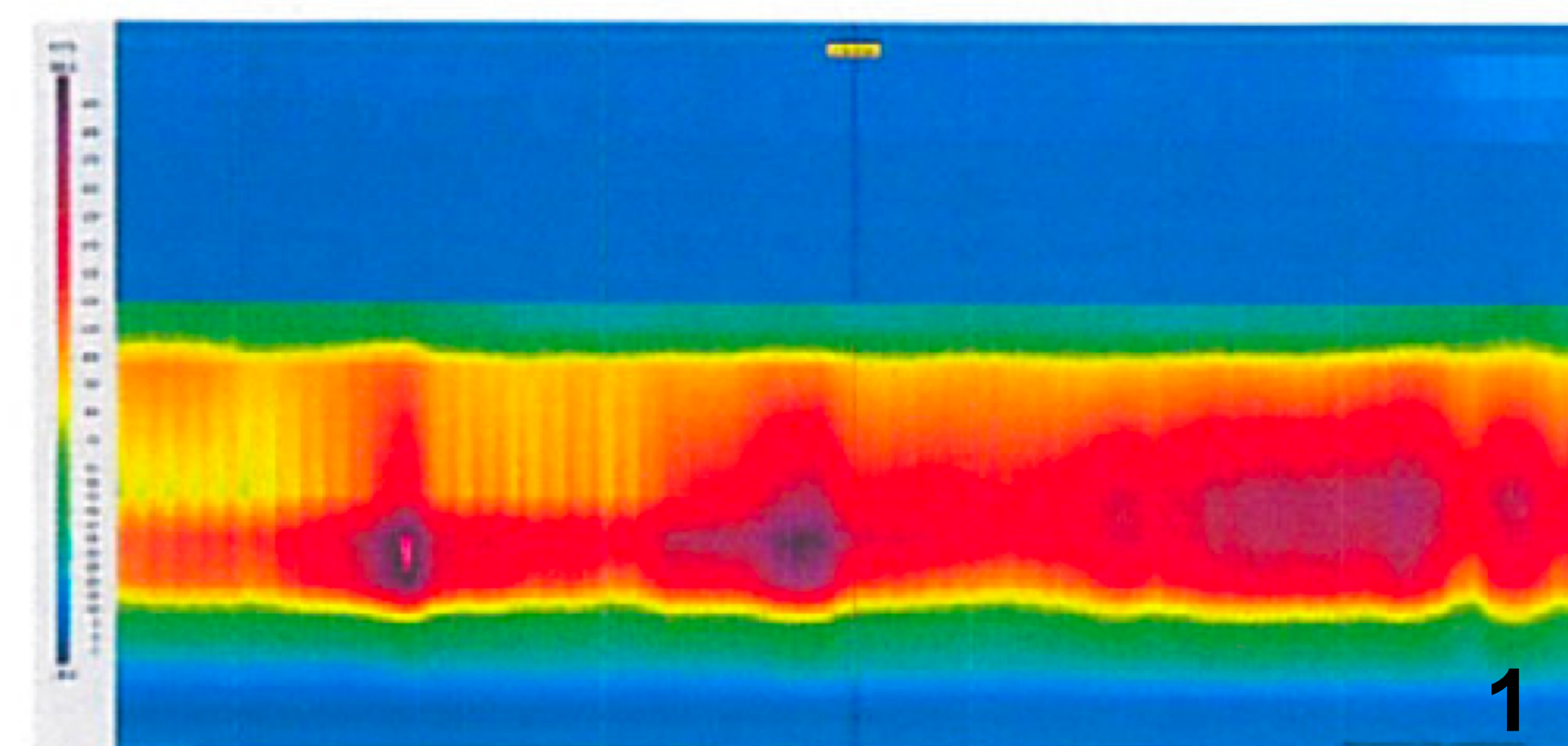
## Introduction

Hirschsprung disease (HD) is a congenital absence of ganglionic cells in the rectum and often thought of as a disease of childhood.

Confirming the diagnosis historically required a suction or surgical open rectal biopsy which is the gold standard of diagnosis.

Here we present a case of HD diagnosed via endoscopic full-thickness resection (EFTR).

## Anorectal Manometry



**Figure 1:** High sphincter pressure at rest is demonstrated

Abnormal RAIR-this was not present up to 130 mL

High resting pressure up to 190 mmHg with normal contractility-sphincter fatigue not seen during squeeze

Patient refused balloon expulsion study due to pain

Normal sensation and urge with rectal hypersensitivity noted

## Discussion

Endoscopic biopsy of the rectum is a minimally invasive technique that can provide adequate tissue samples to confirm a diagnosis.

Endoscopic rectal biopsy may be a safe and effective method to diagnose Hirschsprung Disease in the adult population.

EFTR devices are potentially able to resect the entire wall of the gastrointestinal tract.

This technique could greatly simplify the diagnostic process in that it is safe and effective, and less invasive than surgical techniques.

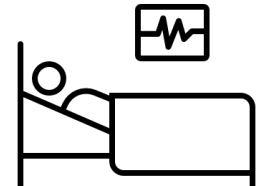
There is a paucity of data regarding comparison between endoscopic versus traditional techniques.

Further studies are needed to characterize the utility of EFTR in this specific population.

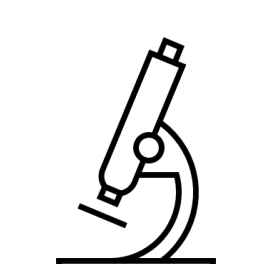
## Timeline



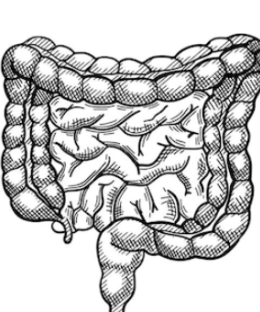
42-year-old female with a history of Hirschsprung disease diagnosed as a child. The patient had an extensive surgical history.



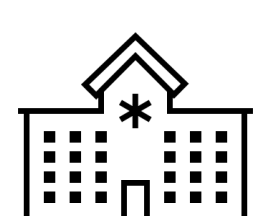
Presented with concern for stricture at previous anastomosis site causing distal obstruction



Anorectal manometry found abnormal push, high sphincter pressure at rest, and absence of rectal inhibitory reflex, findings concerning for HD.



Flexible sigmoidoscopy was performed to determine the extent of the disease. Full thickness endoscopic biopsy of the rectum using an EFTR device showed an absence of ganglionic cells.



The patient underwent a takedown of enterocutaneous fistula as well with improvement in symptoms

## Endoscopy

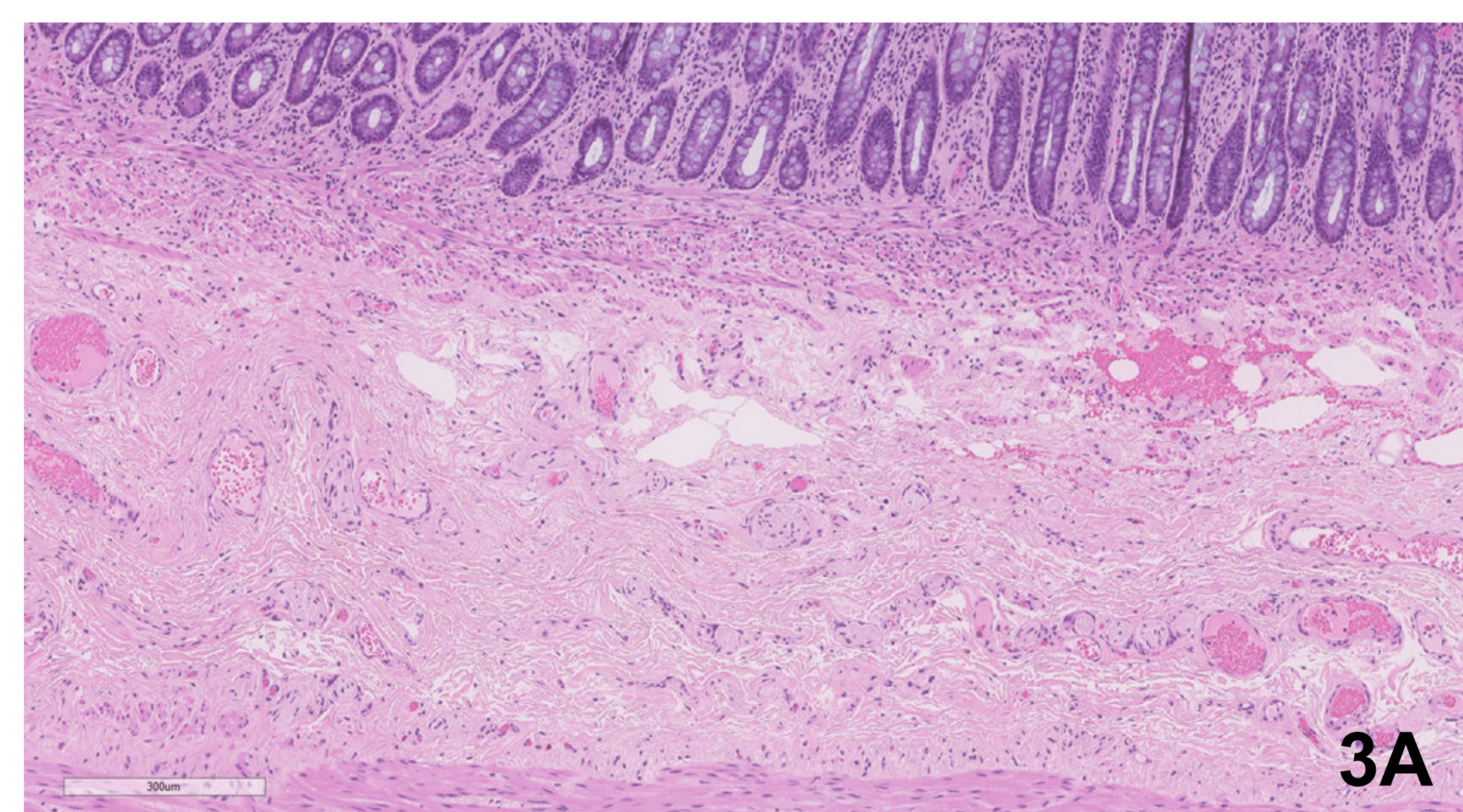


**Figure 2A:** Successful endoscopic full-thickness resection of tissue with Ovesco clip in place

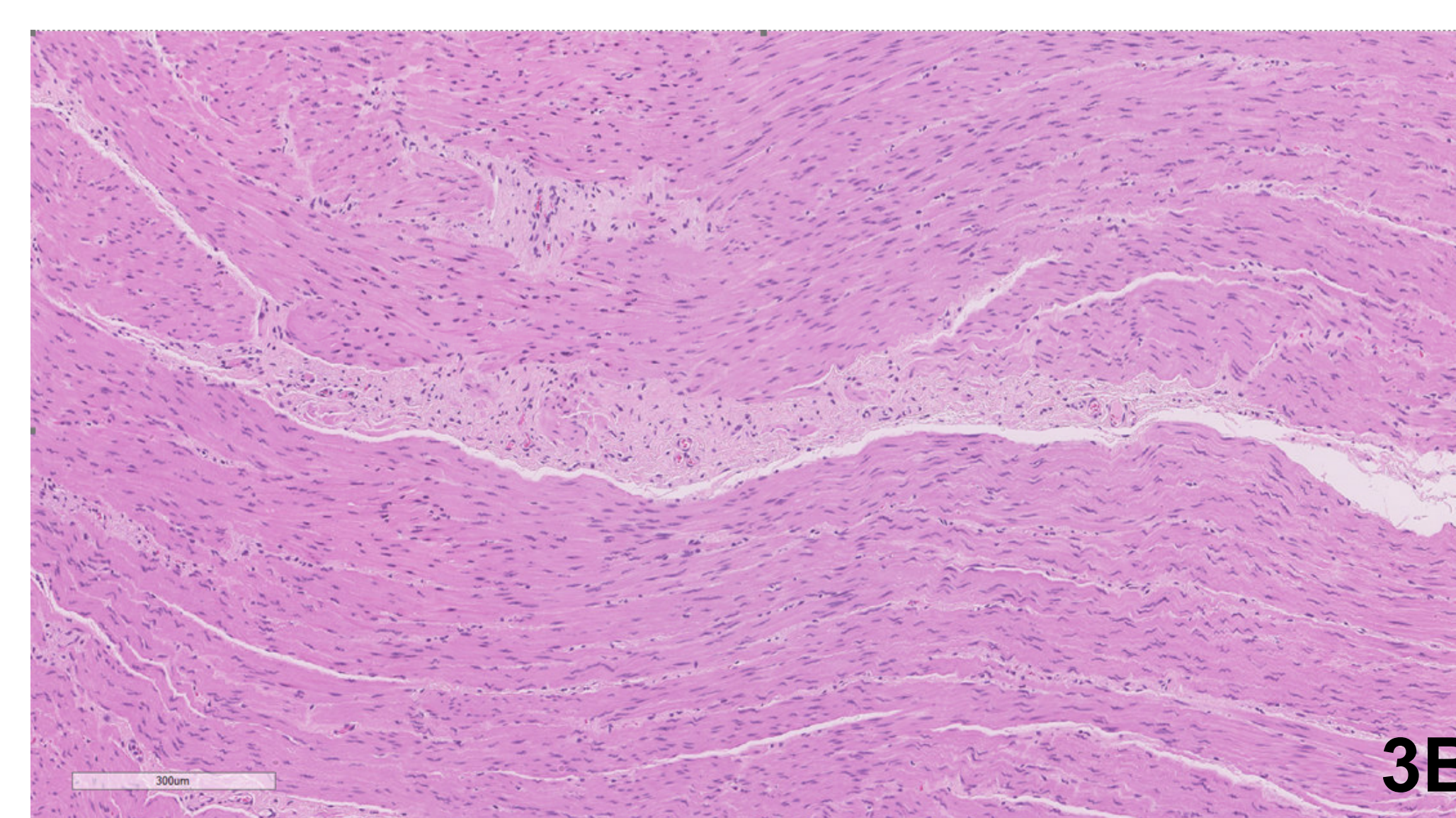


**Figure 2B:** Successful endoscopic full-thickness resection of tissue with Ovesco clip in place

## Pathology



**Figure 3A:** H&E showing absence of submucosal ganglion cells and mild submucosal nerve hypertrophy



**Figure 3B:** H&E showing absence of myenteric ganglion cells.

## References

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