Remembering Childhood Diseases: A Case of Hirschsprung Disease in an Adult with Chronic Constipation

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Background

Hirschsprung disease (HD) is an acquired genetic disorder commonly presenting in the first 24 to 48 hours of life with clinical manifestations occurring before the age of five years in 90% of cases (1).

“Adult HD” is termed for those who develop HD at 10 years of age or older. Typically the age range seen is between 10 to 73 years with an increased incidence before the age of 30 (1).

HD is due to the absence of the ganglion cells of Meissner's plexus in the submucosa and those of the Auerbach plexus in the muscularis.

Case Description

A 24 year old male with chronic constipation presented to the emergency department with intractable severe abdominal pain.

He had a history of intermittent constipation, only liquid bowel movements, no solid stools, and no relief with laxatives or enemas.

Vital signs were within normal limits on arrival. Physical examination showed decreased bowel sounds through the abdomen and left lower quadrant tenderness.

A contrast tomography of abdomen and pelvis showed massive rectosigmoid impaction of feces in the proximal colon, with colon distension up to 18 cm (Figure 1).

Gastroenterology was consulted for colonoscopy completed a week later after intensive bowel preparation with 11 doses of polyethylene glycol, 12 doses of oral magnesium citrate, 7 doses of bisacodyl enema, and 9 doses of oral Bisacodyl.

A rectal biopsy confirmed HD, colorectal surgery recommended a Duhamel with temporary colostomy, which was scheduled and has not been performed yet.

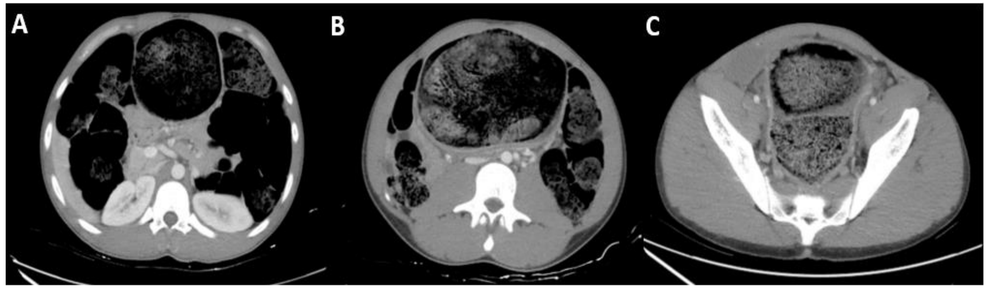


Figure 1. Axial cuts from the patient’s CT abdomen and pelvis showing massive colonic distension with stool burden (Panel A and B) and stool burden in the rectum (Panel C).

Technique	Description	Advantages	Disadvantages
Duhamel Procedure (most commonly used)	Creates a side-to-side anastomosis between normal bowel and a retained rectal stump	Preserves pelvic nerves; lower risk of stricture	Retained rectal stump may cause fecal stasis; requires stapling or suturing
Swenson Procedure	Removes the aganglionic segment and directly anastomoses healthy bowel to rectum	Straightforward and definitive	Risk of injury to pelvic nerves; potential for anastomotic stricture
Soave Procedure	Pull-through of healthy bowel through a cuff of rectal muscle (submucosal dissection)	Reduces risk of nerve damage	Technical complexity; risk of cuff-related complications (infection, obstruction)

Table 1. Displays the three surgical techniques used for treatment of Hirschsprung disease.

Discussion

HD in adults presents delayed because the aganglionic segment is ultrashort or short making it difficult to diagnose (2). Diagnosis in adulthood is usually due to lack of follow-up or ignoring symptoms.

Generally a contrast enema and anorectal manometry are performed rather than suction biopsy as the initial diagnostic option. On manometry a clear transitional zone on barium enema is noted.

Absence of ganglion cells will be seen on biopsy, but also hypertrophic nerve fibers, increased acetylcholinesterase activity in the muscularis mucosa, and decreased or absent immunoreactive fibers in the lamina propria can be present (3).

Mainstay treatment is surgery is resection of the aganglionic segment followed by restoration of continuity between the two healthy segments.

Conclusions

HD in adults is rare, considered if chronic constipation occurs, but it is important to remember that structural causes and medications should be ruled out prior. The diagnosis is based on combination of clinical, manometric, and histological evidence.

References

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