

Hirschsprung's Disease: A Rare Entity in Adults

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ABSTRACT

Hirschsprung's disease (HD), a classic disease of childhood, may rarely escape diagnosis and lead to unique presentations in adults. By definition, the disease involves the rectosigmoid colon and is the result of a birth defect characterised by the absence of nerve cells responsible for the movement of the colon; thereby leading to functional obstruction. We present here a case of an 18-year girl who visited multiple doctors for the complaints of chronic constipation, abdominal distension, and failure to thrive since infancy. Based on a full-thickness rectal biopsy, she was ultimately diagnosed as a case of HD in our Unit. A defunctioning loop ileostomy for colonic decompression, followed by a Swenson Abdominoperineal Pull-Through at a six weeks interval was done, which proved curative.

Key Words: *Hirschsprung's disease, Constipation, Rectal biopsy, Surgical management.*

INTRODUCTION

About one infant out of every 5000 is known to be born with Hirschsprung's disease (HD).¹⁻³ The rarity of adult HD is attributed to the disease being diagnosed in infancy or early childhood.⁴ Rosin documented the first case of adult HD in 1950s.^{1,5,6} The term "adult HD" is used for patients presenting after 10 years of age.^{1,3-7} Approximately, 94% of cases of the disease are diagnosed before the age of 5 years,^{3,6} and only 5% are diagnosed in adulthood.^{2,8} Uptil now, approximately 550 cases of adult HD have been reported; making this disease a major cause of chronic refractory constipation in adults.^{1,4} In adults, the disease is of less severity, so it is taken for granted and is usually misdiagnosed or remains unrecognised until complications occur in the form of fecal retention and/or intestinal obstruction.^{2,5} So, for an accurate diagnosis of HD in adults, a high index of suspicion is needed.²

We report here a case of adult HD with the aim to educate all healthcare professionals who may come across such cases in the future.

CASE REPORT

An 18-year female presented in the Surgical Outpatient Department (OPD) in March, 2018 with the complaints of chronic constipation, recurrent abdominal distention and failure to thrive since infancy. She had a history of delayed passage of meconium. However, there was no history of bleeding per rectum or weight loss of recent onset. She had visited many doctors for the evaluation of chronic constipation, but no accurate diagnosis had been established. We admitted her in our Unit and did a complete evaluation of her symptoms.

She had a low body mass index (BMI) of 16.5. The rest of her general physical examination revealed no abnormal findings. Per abdomen, she was soft, mildly tender and distended with no signs of peritonitis. There were exaggerated bowel sounds on auscultation. Digital Rectal Examination (DRE) was reported normal. A thorough systemic review was also conducted, in which no abnormal findings were observed.

Her biochemical profile was normal. Plain abdominal radiograph showed grossly dilated small and large bowel. Barium enema (Figure 1) showed the same, *i.e.* a hugely dilated proximal colon with a transition zone located between the middle and upper third of the rectum (red arrow). The history, clinical features and imaging studies were all consistent with HD; however, for a confirmatory diagnosis, a full-thickness rectal biopsy was taken. The histopathology report showed segmental submucosal and intramural aganglionosis of



Figure 1: Barium enema showing reversal of recto-sigmoid ratio.

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Figure 2: Resected specimen showing the aganglionic segment of the colon.

the upper rectum with hypertrophied nerve fibers, thus confirming HD.

A de-functioning loop ileostomy was done to decompress her grossly dilated colon and at the same time four full thickness biopsies were taken from the ascending, transverse, descending and sigmoid parts of the colon to know the extent of colonic involvement. The histopathology reports were normal, confirming involvement of rectum only.

After a period of 6 weeks, Swenson Abdominoperineal Pull-Through was done. The affected segment was resected (Figure 2) and coloanal anastomosis was done; saving the anal sphincter and preserving continence. The stoma was then reversed after 8 weeks. The patient made an uneventful recovery. Her BMI had improved drastically on follow-up visit after 3 months.

DISCUSSION

In 1886, the Danish physician, Harald Hirschsprung was the first to describe Hirschsprung's disease,^{3,8,9} as a congenital disorder characterised by the absence of ganglion cells in the Auerbach and Myenteric plexuses of the colon.^{2,3,7,8,10} The aganglionic portion remains contracted, causing functional obstruction and resulting in proximal dilatation.^{7,8} Based upon the extent of colonic involvement, the disease is classified into short segment disease (involvement of sigmoid colon only), ultra-short segment disease (involvement of rectum only), long segment disease (involvement beyond sigmoid colon) and total colonic aganglionosis. It may present as a separate entity or in combination with other anomalies, e.g. Down's syndrome in 10% cases.^{2,3} Adult HD may manifest between the age of 10 to 73 years and is more common in males than females (frequency being 4:1);^{3,6,7} however, females are more affected in the adult form of the disease (3:1).⁷ The common presenting features are chronic refractory constipation, recurrent abdominal distension and discomfort since infancy and early childhood.^{2,3,5-8,10}

A full thickness rectal biopsy is considered as the gold standard for definitive diagnosis of the disease with barium enema and anorectal manometry being useful adjuncts in diagnosis.^{5,7,10} A classical rectal biopsy shows absence of ganglion cells in the affected segment along with neural hyperplasia in the submucosal and muscular layers.^{2,7,10} Barium enema exhibits reversal of the rectosigmoid ratio (normal is 1),¹¹ which is visible as a grossly dilated proximal colon and distal narrow rectum (aganglionic segment); representing a transition zone.^{1,10} To differentiate idiopathic megacolon from adult HD, anorectal manometry is being highly advised as a screening test showing absent rectoanal inhibitory reflex in affected individuals.⁸

The management of HD is surgical, in which more than half a dozen approaches have been described. However, all of them have their own pros and cons. These approaches include Soave Endorectal Pull-Through, Duhamel Retrorectal Pull-Through, Swenson Abdominoperineal Pull-Through, Posterior anorectal myectomy, Posterior anorectal myectomy with Low Anterior Resection, Low Anterior Resection (state procedure) and colectomy. Among these procedures, Anorectal myectomy combined with Low Anterior Resection shows excellent results in terms of complications and long-term outcome.^{3,10} All these procedures can be done laparoscopically as well, with promising results.⁷

The patient's clinical history, radiological reports and rectal biopsy findings were, in fact, compatible with those described in literature. She made a good recovery with Abdominoperineal Pull-Through procedure done 6 weeks after a defunctioning ileostomy. Her symptoms were relieved, and BMI had markedly improved at 3 months follow-up. As evident from the history of our patient and supported by literature, adult HD represents a misdiagnosis of the disease at an early age, which is correctable by surgery.

HD in adults poses a diagnostic challenge because it is usually misdiagnosed or may go unrecognised in childhood. A high index of clinical suspicion is, therefore, the one important tool that may aid in accurate diagnosis, and accordingly a timely intervention.

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