

# Adult Hirschsprung's Disease; a case report of total Colonic Aganglionosis (Zuelzer-Wilson Syndrome)

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## SUMMARY

Hirschsprung's disease is rare in adults and it is thus often undiagnosed or misdiagnosed. It should be considered in young adults with a history of chronic constipation. Where mostly diseased part is confined to rectosigmoid area it may involve larger segments i.e., whole of colon or even beyond that. Barium enema and anorectal manometry play role in diagnosis but definitive diagnosis is established on histology of full thickness biopsy from the rectum showing aganglionosis. Management is always surgical involving resection of aganglionic segment and restoring normal bowel continuity. We report a case of 32 years old female with Hirschsprung's disease involving whole of colon.

**Keywords:** Adult Hirschsprung's disease, Zuelzer-Wilson syndrome, total colonic aganglionosis,

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## INTRODUCTION

Frederick Ruysch in 1691 reported the first case of a patient with Hirschsprung's disease, but it was a Danish pediatrician Harald Hirschsprung who in 1888 published the classic description of congenital aganglionic megacolon<sup>1</sup>. It is a congenital disorder characterized by the absence of myenteric and submucosal ganglion cells along a variable length of the distal gastrointestinal tract<sup>2</sup>, caused by failure of neural crest cells (precursors of enteric ganglion cells) to migrate completely during embryonic intestinal development resulting in aganglionic segment of the colon which fails to relax, causing a functional obstruction<sup>3</sup>.

The incidence of Hirschsprung's disease is approximately 1 per 5,000 live births<sup>4</sup>. The length of the aganglionic segment is variable: it is limited to the rectum and sigmoid in 75% of patients; involves the whole colon in 8%; and is rarely known to involve the small bowel. Absence of enteric nervous system in whole of colon is also called "Zuelzer-Wilson syndrome" was first reported by Zuelzer & Wilson, who suggested that aganglionosis can extend from duodenum to the rectum. Males are affected more than females by a ratio of 4:1<sup>4</sup>. However, for short-segment disease, the male-to-female ratio is 4.2-4.4 and for long-segment disease the female-to-male ratio 1.2-1.9<sup>5</sup>.

Hirschsprung's disease is associated with a chromosomal abnormality in 12% of cases (>90% trisomy 21) and with additional congenital anomalies in 18% of cases whereas in 70% patients it occurs as an isolated abnormality<sup>6</sup>.

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Although mostly diagnosed in early childhood, roughly 5% of these cases are diagnosed in the adult population. It is frequently misdiagnosed as chronic constipation in these individuals<sup>7,8</sup>, and a common presentation is with volvulus of a colonic segment. The actual frequency in adults is unknown, mainly because this disease can be overlooked and misdiagnosed in this age group [9] as was the case in our patient. Adult Hirschsprung's disease is therefore thought to be more common than previously recognized<sup>10</sup>.

## CASE REPORT

The case being reported is that of a 32 years old unmarried female who presented in surgical emergency department in July 2014 with signs and symptoms of acute intestinal obstruction. Detailed clinical evaluation revealed history of delayed passage of meconium and chronic constipation that required laxatives and enemas since early childhood. There was no history of per-rectal bleeding or recent weight loss.

She had BMI of 17. Abdominal examination showed a soft but distended abdomen without any signs of peritonitis. Bowel sounds were exaggerated. Digital rectal examination and rest of systemic examination was unremarkable. Biochemical profile was within normal range. Abdominal X-ray showed massively dilated small gut and fecal loading of large gut (Fig. 1). CT reported fecal loading of large bowel traced up to the rectum, with markedly dilated bowel loops in abdominal cavity (Fig. 2). She underwent an emergency laparotomy, operative findings were consistent with radiography so a loop ileostomy was formed to relieve obstruction and due to high suspicion of a motility disorder full thickness rectal and colonic biopsies were taken.

Patient's obstruction was relieved. Post-op barium enema showed delayed emptying of contrast media from colon. The colonic and rectal biopsies showed aganglionosis so diagnosis of Hirschsprung's disease involving entire colon was made and patient underwent an elective total colectomy (Fig. 3) and J-pouch ileo-anal anastomosis (Fig 4) with covering loop ileostomy. Per operative frozen section of full thickness biopsies from terminal ileum showed normal enteric nervous system. Histopathology report of resected specimen showed features consistent with Hirschsprung's disease with absence of myenteric and the submucosal plexus throughout the submitted specimen. Calretinin immunohistochemistry stain was used which confirmed histopathological diagnosis of aganglionosis (Fig 5 & 6).

The patient had uneventful post-operative recovery and was discharged on the seventh post-operative day. Later reversal of ileostomy was done. Since then she has been under regular follow-up and has reported significant improvement in her bowel function and quality of life.



Figure 4

Figure 1



Figure 2



Figure 3

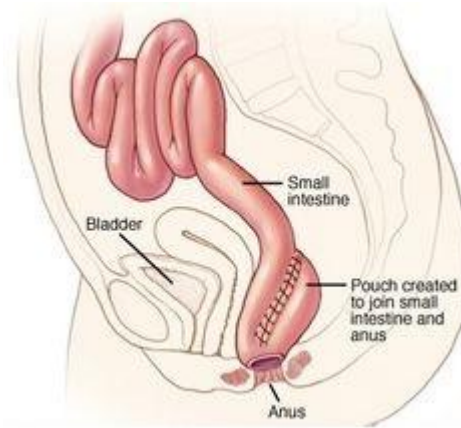
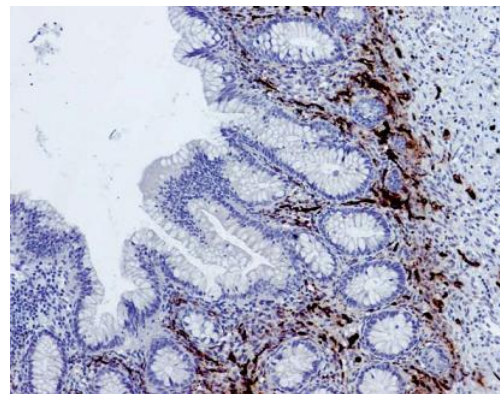
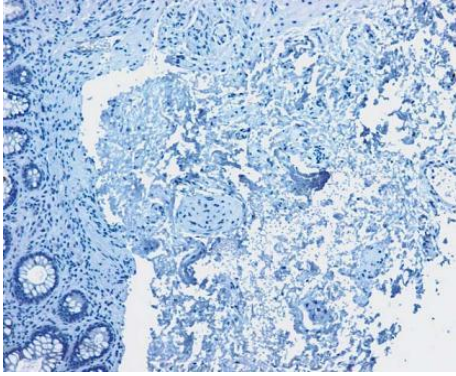


Figure 5



Calretinin immunohistochemistry in patients with normal innervation, marking positive lamina propria of the mucosa and Meissner's muscular plexus

Figure 6



Calretinin immunohistochemistry in patients with Aganglionosis. Negative in the lamina propria of the mucosa and Meissner's muscular plexus

## DISCUSSION

Hirschsprung's disease usually presents in infancy, although some patients present later in life with persistent, severe constipation<sup>11</sup>. A typical grown-up patient has history of long-standing constipation since infancy or early childhood; different manifestations may include recurrent fecal impaction, abdominal pain / discomfort and distention, failure to thrive, malnutrition etc<sup>7,8</sup>.

The primary defect in adult Hirschsprung's disease is identical to that seen in infancy<sup>12,13</sup>. The correct diagnosis is based on the proper medical history, the barium enema, anorectal manometry, but definitive diagnosis is based on rectal biopsy<sup>14</sup>, (predictive value 100%) which usually reveal absence of ganglion cells and increased acetyl cholinesterase activity of mucosa nerve fibers<sup>15</sup>.

Sensitivity and specificity of barium enema in the diagnosis of Hirschsprung's disease are reported as being 76% and 97%, respectively<sup>16</sup>, but may not be very useful in total colonic aganglionosis, with a transition zone only being accurately determined in 25% or less of all colonic aganglionosis patients<sup>17</sup>. A 24-hours-delayed x-ray obtained after a barium enema however shows retention of barium and stool in the rectum as was observed in our case. Barium enema is not as sensitive or reliable as rectal suction biopsy in ruling out Hirschsprung's disease<sup>18</sup>. Anorectal manometry records failure of reflex relaxation of the internal anal sphincter in response to inflation of a rectal balloon. It is not widely available in our setup so was not conducted in our patient.

Management always require surgical intervention. The reconstructive procedures for long-segment Hirschsprung's disease include: straight pull-through, colon patch, and J-pouch construction. Pull-through procedures using any one of the standard techniques (Swenson, Duhamel, Soave)<sup>19,20,21</sup> bring the normally innervated ileum to

just above the anal sphincter. These procedures were initially developed for pediatric disorders, but have been applied to adults with varying degrees of success. Duhamel's procedure is generally considered the best surgical procedure, provides better results in adults and is also associated with lower post-operative morbidity rates<sup>7,8,22,23,24</sup>.

No consensus exists on the optimal surgical management of total colonic aganglionosis<sup>25</sup>. We did restorative proctocolectomy with J-pouch ileoanal anastomosis in our patient which is a feasible method of reconstruction in patients requiring proctocolectomy. Its long-term functional results, e.g. continence and bowel frequency, are excellent and ensure good quality of life in majority of patients<sup>26</sup>. A J-pouch procedure involves an ileum to anal anastomosis with connected loops of folded ileum that form an internal reservoir. Another option is a colon-patch is a side-to-side anastomosis between normally innervated ileum and aganglionic colon, thus using the small bowel for motility and colon as a reservoir for storage of stool and absorption of water.

Post-operative complications in adult patients are mostly minor but anastomotic dehiscence, anastomotic stricture, pelvic abscess, anastomotic fistula, retraction or necrosis of colon may occur<sup>27</sup>. After diagnosis and surgery, the patient's family should be counselled about the importance of a high-fiber diet in light of the fact that constipation and bowel stasis are thought to increase the risk of enterocolitis<sup>28,29</sup>.

There is some hope that the use of autologous neural crest-derived enteric stem cells in future may be a treatment for Hirschsprung's disease which would mean avoidance of surgery which has associated risks<sup>30</sup>.

## CONCLUSION

Diagnosis of Hirschsprung's disease in adults is a challenging condition because it can be easily overlooked or misdiagnosed. High index of clinical suspicion should guide the physicians towards making the correct diagnosis and management plan.

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