

Adult Onset Hirschsprung's Disease: A Case Report

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Abstract: ***Background:** Hirschsprung's disease is commonly diagnosed in children shortly after birth. It is an uncommon diagnosis in adults and often misdiagnosed. **Case:** A 29-year-old woman presented with chronic constipation and distended abdomen for over 2 months. After several examinations she was diagnosed with adult Hirschsprung's Disease. Surgery was performed and a dilated colon was obtained and resected. Biopsy of the dilated segment revealed aganglionic and hypoganglionic cells consistent with the diagnosis of Hirschsprung's Disease. **Conclusion:** Although uncommon, Hirschsprung's Disease should be considered in adults. Early diagnosis and prompt management can provide a good outcome.*

Keywords: Adult Hirschsprung's disease, chronic constipation, colostomy, biopsy

1. Introduction

Hirschsprung's disease is a congenital malformation which is commonly diagnosed shortly after birth. This disease is characterized by the absence of ganglion cells (aganglionosis) in the distal bowel, resulting in functional intestinal obstruction.

Hirschsprung's disease occurs in approximately 1 in 5000 live births and more common in male, with over 80% of patients being diagnosed in the neonatal period or during infancy [1]. However, if the symptoms are mild, this disease may not be diagnosed until adulthood. According to literature, if diagnosed above 10 years of age, it can be termed as adult Hirschsprung's Disease [2].

Adult presentation usually comes with recurrent constipation in most cases, requiring enema for relief of the symptoms right from childhood, but some of the cases may present with symptoms for the first time in adulthood [3]. Diagnosis is confirmed when the aganglionosis parts of the colon was found in rectal biopsy. Staged surgical resection and subsequent re-anastomosis provide a good outcome.

2. Case Report

A 29-year-old woman presented with symptoms of colicky abdominal pain, abdominal distention and chronic constipation which she had been experiencing over 2 months. She experienced nausea and anorexia. She felt comfortable in a lying position and felt discomfort when sitting due to her distended abdomen.

She had no hematochezia, had not undergone any previous surgery and had no family history of Hirschsprung's Disease. However, she had similar symptoms at 5 years of age. At that time the patient's parents only gave her medications and her complaints improved.

Clinical examination showed a distended abdomen, normal bowel sound, palpable intra-abdominal mass and slight

tenderness. Digital rectal examination revealed good anal sphincter tone and high rectal fecal load.

Routine laboratory evaluation showed an elevated white blood cells count (25.400/uL) with elevated neutrophil count (81, 3%) and slight anemia (11,0 g/dL). Electrolyte evaluation showed a slight elevated chloride level (112 mmol/L). Other laboratory evaluations were all within normal limits.

Plain abdominal radiograph showed fecal mass and gas distribution in the intestinal lumen down to the distal.

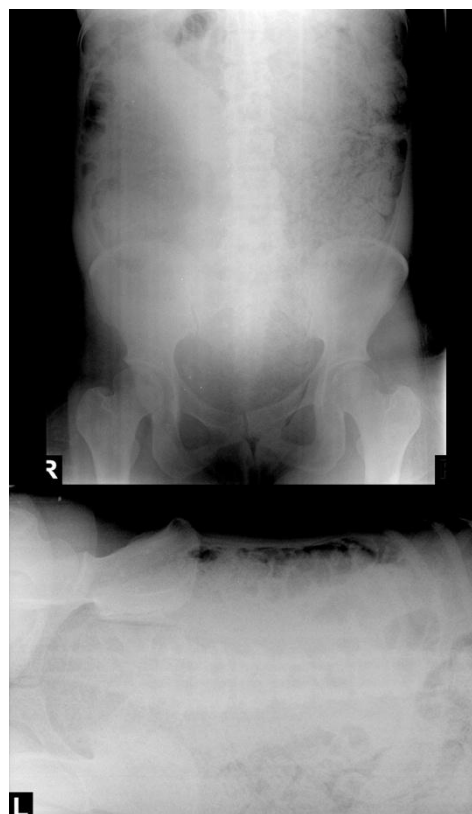


Figure 1: Fecal mass on plain abdominal radiograph

Computed tomography (CT) of the abdomen showed dilated and distended sigmoid colon and descending colon up to T8-9 with large quantities of fecal mass in the lumen.

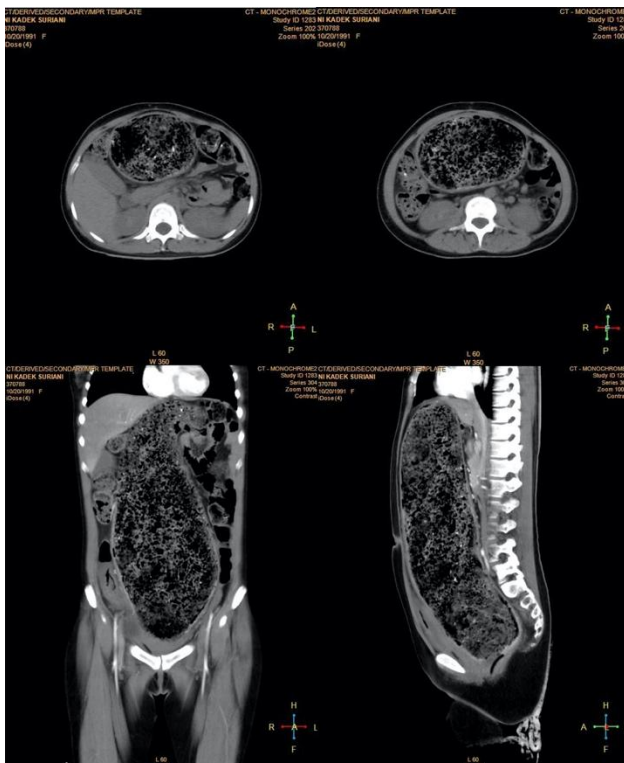


Figure 2: Severely dilated colon with fecal impaction.

As the initial management, decompression was performed using a nasogastric tube. Laxatives and enema were given but only resulted in minor improvements.

After a few days without significant changes, the patient underwent surgery. Exploratory laparotomy was performed with a midline incision. The dilated sigmoid colon and descending colon were found to occupy most of the abdominal cavity and filled with fecal matter. The dilated segment (approximately 40 cm) was resected and sent to the pathological laboratory for further examination. Furthermore, a colostomy was made.

After the surgery, the patient's symptoms have completely subsided after a few days of post-operative care. Furthermore, the patient is planned to undergo further surgery for colorectal anastomosis whenever possible.

Histological examination of the resected segment revealed aganglionic and hypoganglionic cells consistent with the diagnosis of Hirschsprung's Disease.

3. Discussion

Hirschsprung's Disease (HD) is a congenital anomaly that occurs due to a discontinuation of the cranio-caudal migration of neural crest cells, which are responsible for innervation of the colon, or when ganglion cells develop at 5th to 12th weeks of pregnancy [4].

The first documented case of adult HD was described by Rosin et al, in 1950. While the incidence of adult HD is not

known at this moment presume because this disease is often overlooked in the adult population. According to Miyamoto et al, adult HD is more common in male over women 133 to 42 with the patient age ranging from 10 to 73 years old and the average of 24,1 years (half of the population are under 30) [5].



Figure 3: Intraoperative findings

Symptoms of adult HD include chronic recurrent constipation, since birth, infancy, or childhood in most cases, and since adulthood (>50 years) in some cases. On clinical examination various degrees of abdominal distension and pain are common (83-86%), usually with associated palpable fecal mass (50-56%) or fecal impaction (25-26%), abdominal pain, and tenderness (40-80%). Cathartics and/or enemas were used regularly to achieve bowel movement (73-92%).⁶ Related to this case, the patient was found with chronic constipation, abdominal colic pain, and distended abdomen for over two months. For the radiologic examination plain abdomen radiographs typically show massive distention at the proximal sites of the aganglionic segment of the colon, with narrowing distal

segment. Therefore, for the diagnosis of adult HD we need an accurate history of medical history, barium enema test, and most importantly full thickness rectal biopsy showing aganglionosis in the myenteric plexus and hypertrophied nerve endings as the gold standard to make a definitive diagnosis [5]-[7].

After HD is diagnosed, surgery is usually needed. Prior to surgery, conservative approaches such as serial rectal irrigation can be performed to decompress the bowel and prevent enterocolitis. In addition, other actions can also be taken such as, *nil per oral*, nasogastric tube insertion, intravenous broad-spectrum antibiotics, intravenous fluids for maintenance and correction of dehydration, routine blood investigations and also correction of acid base and electrolyte imbalance if any [3].

The surgical procedure in adult HD cases is a modified form of the surgical procedure applied to HD cases in children. Rectolectomy with colorectal anastomosis (Rebentzen procedure) or total colectomy with ileorectal anastomosis (modified Martin-Duhamel procedure) are some of the procedures commonly used in adult HD cases. Unlike in children HD cases, the surgical procedure in adult HD uses a one-stage approach due to relatively healthier nutritional status, without performing a temporary protecting colostomy. If the patient has HD related enterocolitis or a significantly dilated colon, a colostomy may be performed for several months while the patient recovers and anastomosis procedure is usually performed four to six months after colostomy placement. In this case a temporary colostomy was performed due to the significant difference in diameter of size of the proximal and distal segments so that anastomosis could not be performed.

Intraoperative frozen section biopsy examination may be needed to identify the distal margin of the bowel with ganglion cells [2].

Most patients with HD who receive prompt treatment do not experience complications. However, complications such as constipation, fecal incontinence, enterocolitis and colonic rupture may occur.

After diagnosis and treatment, the patient needs to consume foods high in fiber to prevent constipation.

4. Conclusion

As a conclusion, although uncommon, adult Hirschsprung's Disease should be considered as one of the differential diagnosis in patients presenting with chronic constipation accompanied by dilated colon without evidence of other causes as obstruction. Prompt diagnosis and management can provide satisfactory outcomes.

5. Acknowledgement

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6. Disclosure

The authors state no conflict of interest in this manuscript.

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