*Case Report*

# Hirschsprung's Disease in an Adult with a 37-year History of Colostomy

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***Key Words***
Hirschsprung's disease;
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In adults Hirschsprung's disease is a rare and frequently misdiagnosed cause of longstanding refractory constipation. A 37-year-old man visited our emergency department because he had been experiencing dull abdominal pain for days. The patient had received a colostomy for intestinal obstruction shortly after birth. He received takedown of the colostomy when he was 7-year-old, but the colostomy was performed again due to persistent intestinal obstruction. However he continued to suffer from similar symptoms thereafter. Physical examination revealed a colostomy stoma on the left upper quadrant of his abdomen and moderate tenderness to palpation in both the right and left quadrants. Abdominal plain film revealed fecal and barium impaction. A diagnosis of Hirschsprung's disease was made when a rectal biopsy was performed and no ganglion cells were noted. The patient underwent a modified Duhamel's pull-through operation and a protective colostomy. No abdominal discomfort was noted before he discharged or during followed up. Adult Hirschsprung's disease is a rare entity. Although several reports have mentioned adult Hirschsprung's disease, there are no reports describing a case with a colostomy after birth. We report this case and discuss the history of management of Hirschsprung's disease to help explain the reason for delayed treatment in this patient.
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Adult diagnosis of Hirschsprung's disease (HD) is a rare and frequently misdiagnosed cause of long-standing refractory constipation.[1,2] In most cases, patients have a mild form of the disease and as a result go undiagnosed early in their lives. A mild form of the disease may occur because the proximal innervated colon is hypertrophied and thus compensates for the distal obstructed aganglionic rectum.[3] These patients often try to relieve the constipation by taking cathartics and using enemas. Here we investigate the treatment of a 37-year-old patient who underwent one colostomy just after birth that was reversed followed by a second colostomy.

## Case report

A 37-year-old man with a multi-year history of intermittent abdominal pain was admitted to our emergency department with severe abdominal pain. He had previously relieved his symptoms by using an enema. The patient had undergone a colostomy for intestinal

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obstruction shortly after birth. Takedown of the colostomy was performed at 6 years of age, however, a second colostomy was performed due to the development of life-threatening intestinal obstruction and sepsis. Our physical examination revealed a colostomy stoma on his left abdomen and moderate tenderness to palpation was noted in the right and left lower quadrants in the absence of rebounding pain. His anal tone was very tight. All laboratory data were within normal limits. Figure one illustrates the abdominal plain film examination showing fecal and barium impaction (Fig. 1). Histological analysis of the rectal biopsy specimen revealed a lack of ganglion cells that is consistent with Hirschsprung's disease (HD).

In our hospital the patient underwent a modified Duhamel's pull-through operation and a dilated sigmoid colon and partial descending colon were found and resected (Fig. 2). We also excised the loop-colostomy so that it would not negatively affect the pull-through procedure and a new protective colostomy was performed. The resection colon was 41.6 cm in length (including 11.6 cm below the previous colostomy). Pathological examination revealed segmental absence of ganglion cells in the myenteric plexus and submucosal plexus with proliferation of nerve bundles measuring 4 cm in length and focal transitional segments with presence of immature ganglion cells (Fig. 3). The patient did not report any abdominal discomfort after the operation and elected not to undergo takedown of the new colostomy.

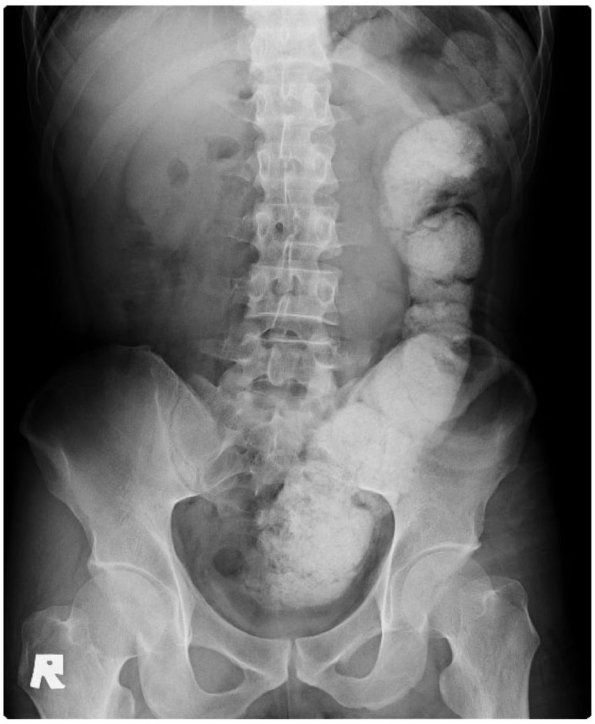


**Fig. 1.** Abdominal plain film reveals fecal and barium impaction in the sigmoid and descending colon.

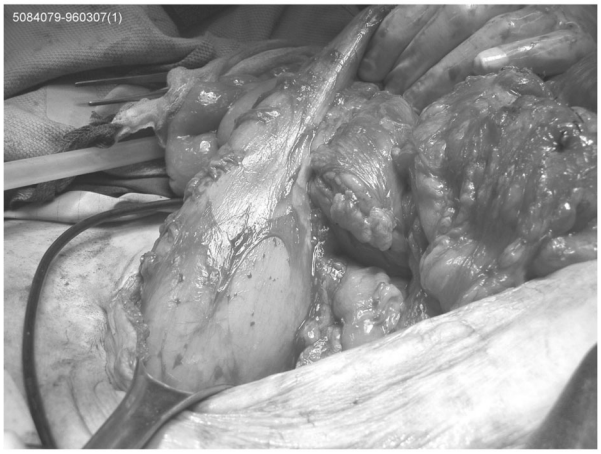


**Fig. 2.** Dilated sigmoid and descending colons were found.

## Discussion

The majority of cases of HD are diagnosed and treated in the neonatal period making persistence of the disease into adulthood rare. Most patients with adult HD will survive, however the disease substan-

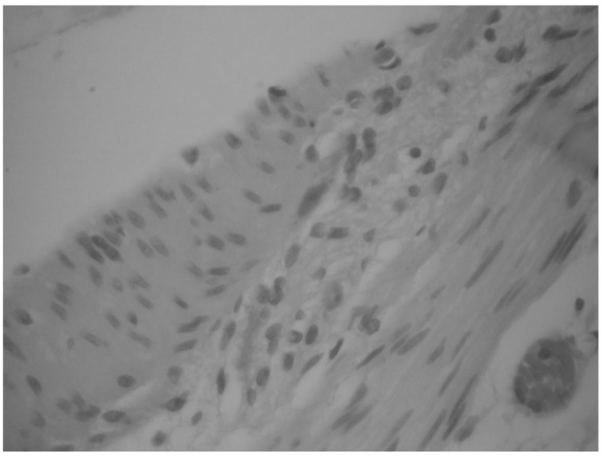

**Fig. 3.** Abnormal immature ganglion cells were found in muscular layer (H & E 400X).

tially affects their quality of life. Although our patient did not suffer from constipation, the colostomy caused him to suffer from depression.

Swenson performed the first surgical treatment of HD, an abdominoperitoneal pull-through procedure, in 1948.[4] In 1960, Duhamel described a retrorectal pull-through procedure,[5] and in 1964, Soave described an endorectal pull-through procedure.[6] Although adults with HD are rare, previous studies have found surgical results to be uneventful.[1-3] In 1965, Dr. Hung was the first to treat a case of HD in Taiwan using the modified Duhamel's procedure. Since this time most pediatric surgeons in Taiwan opted to perform the same procedure for treatment of HD.[7] After 1996 Dr. Wang began to treat HD using the laparoscopy-assisted endorectal pull-through method.[8]

Our patient was born in 1970 and underwent a colostomy for intestinal obstruction shortly after birth. It is not clear from the patient's medical records why the takedown of the colostomy was performed in 1976. However, the result was the development of life-threatening intestinal obstruction and sepsis causing doctors to perform a second colostomy. Likely the low socioeconomical level of his parents contributed to their lack of knowledge of the disease and treatment of their son. Although the second colostomy relieved some pain, the patient continued to suffer from intermittent abdominal pain and frequently required an enema to relieve his symptoms. Nevertheless, his body weight and height was largely normal (body mass index value was 20), he married, and had three healthy children.

In conclusion, a colostomy appears to markedly improve the survival rate of patients with HD, however the presence of a colostomy affects the patient's quality of life. In addition, patients with a colostomy still frequently use enemas to relieve symptoms as do patients who have not received a colostomy.

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**病例報告**

# 一個成人型赫司朋氏病病患伴隨人工肛門 37 年：病例報告

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一個 37 歲男性病患因反覆性腹部疼痛而來院求治。理學檢查發現在左側腹壁上有一個人工肛門。根據病患自己的描述，他於出生後 (1970 年) 不久即因腸阻塞而接受人工肛門手術。雖然於 7 歲時接受人工肛門關閉手術，但隨即因腹脹併發敗血症而再次接受人工肛門手術。經直腸切片檢查後，確認爲赫司朋氏病。病患於接受改良型 Duhamel 直腸拉出手術後，症狀明顯改善。台灣外科先驅洪文宗教授於 1960 年代開始施行赫司朋氏病的直腸拉出手術，一直到 1970 年代才開始廣泛施行於整個台灣。在這之前的赫司朋氏病病患如何治療呢?雖然人工肛門可用以維持病患的生命，但並無法緩解病患腹部的不適。我們報告這一個病例並回顧相關的文獻及台灣治療赫司朋氏病的歷史。

**關鍵詞** 成人、赫司朋氏病、人工肛門。