Diagnostic conundrums of hypoganglionosis that may delay timely, optimal
treatment

Authors:  Adam Alzubedi, Roman Styliński, Paweł Polski, Monika Kusz, Przemysław Matras, Sławomir Rudzki

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Title: Diagnostic conundrums of hypoganglionosis that may delay timely, optimal treatment

Short title: Hypoganglionosis in an adult

Adam Alzubedi¹, Roman Styliński¹, Paweł Polski¹, Monika Kusz², Przemysław Matras¹, Slawomir Rudzki¹

1. I Chair and Department of General and Transplant Surgery and Nutritional Treatment, Medical University of Lublin, Lublin, Poland

2. II Chair of Pediatrics, Department of Pediatric Nephrology, Medical University of Lublin, Lublin, Poland

Correspondence to: Adam Alzubedi, MD, I Chair and Department of General and Transplant Surgery and Nutritional Treatment, Medical University of Lublin, ul. Jaczewskiego 8, 20-954 Lublin, Poland, phone: +48 81 724 48 29, email: adam.alzubedi@gmail.com

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Hypoganglionosis is a very rare disease that presents with severe chronic constipation resembling clinical features of Hirschsprung’s disease and may give rise to megacolon. The median age at diagnosis is considerably higher in this group of patients due to difficulties in diagnosis that is based in general on the result of full-thickness biopsy that may indicate but often cannot prove hypoganglionosis and a lack of consensus in diagnostic criteria[1]. Delayed diagnosis leads to chronic constipation poorly treated with enemas and laxatives, lowers the quality of life, and may have fatal complications. A systematic review in 2010 of 92 cases presented over 11 publications revealed that only 32% of cases were diagnosed in newborns[2].

We present a case of a 19-year-old patient with chronic severe constipation who was diagnosed since early childhood for Hirschsprung’s disease. The patient underwent three rectal biopsies, which were negative for aganglionosis. He was diagnosed with chronic idiopathic constipation. After seeking professional help patient self-medicated to attenuate symptoms with chronic use of laxatives, enemas, cigarette smoking, and food restriction, which led to poor health-related quality of life. The patient was admitted with symptoms of acute obstruction, distended, painful abdomen, and vomiting. Laboratory findings were unremarkable. Abdominal X-ray (Figure 1A) showed fecal impaction, Computed Tomography scans (Figure 1B-1D) showed megacolon with compression on both abdominal and chest organs. The clinical and radiological presentation was typical for Hirschsprung’s disease in adults[3]. Apart from acute symptoms, the patient was also malnourished (Body Mass Index – 18.1; weight – 62 kg after manual disimpaction) and in poor mental condition. Before the planned surgical treatment patient was on total parenteral nutrition and underwent two sessions of manual disimpaction under general anesthesia as a mechanical bowel preparation. The patient displayed a high degree of emotional lability and did not consent for a restorative proctocolectomy with ileal pouch-anal anastomosis. Instead, he insisted on total
proctocolectomy with a definitive ileostomy formation. Due to 19 years of suffering chronic
constipation patients life quality was low, and he was not willing to bear the risk of ileal
pouchitis or higher frequency of bowel movement. Surgery started with a laparoscopic
approach, but due to lack of working space, it was converted to laparotomy. Intraoperatively
an enormously distended, thick-walled megacolon with a maximum diameter of 20 cm was
visualized; total proctocolectomy with definitive ileostomy formation was performed as
planned. After the initial surgery patient presented with a prolonged postoperative ileus and
was, therefore, qualified for revision laparotomy during which stoma reformation was
performed. To our surprise, histopathology once again revealed sparse myenteric ganglia with
nerve plexuses proliferation with reduction of nerve cells toward the distal resection line
without aganglionosis in the specimen leading to the final diagnosis of isolated
hypoganglionosis.

The purpose of this case report is to highlight the radiological and clinical similarity of
isolated hypoganglionosis to Hirschsprung’s disease, which makes it difficult to diagnose in
the early period with rectal biopsies that can be false negative, thus leading to delayed
diagnosis and treatment with subsequent poor quality of life.
References


FIGURE 1 Megacolon. (A) Abdominal X-Ray shows massive fecal impaction. (B) Computed tomography image taken in the axial plane just below the level of the trachea bifurcation shows ascending colon filled with stool causing compression of the right lung and pushing the heart to the left side. (C) Computed Tomography image taken in the sagittal plane shows megacolon compressing liver and reaching high into the chest. (D) The image taken in the coronal plane shows abdominal distension and right lung compression. (E) Intraoperative photograph. (F) Resected specimen after decompression.