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Hirschsprung Disease in a Female Infant: A Case Report

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Abstract

Hirschsprung's disease (HD) is a congenital disease characterized by the absence of ganglion cells in the intestinal muscularis nerve plexus. The segment most affected was the rectosigmoid colon (80%). The clinical manifestations are non-specific; the common signs include vomiting, abdominal distention, and defecation alterations in early-life another feasible alteration is anemia. Anorectal manometry and contrast enemas are also highly useful. We present the case of a 9-month-old female with refractory constipation who was diagnosed with HD. Some theories explain that there is dysregulation of the microecological balance and intestinal mucosa. Imaging diagnostic methods are useful tools for screening HD. The mortality rate of these conditions is between 2% and 5%; therefore, a group of qualified professionals is necessary for treatment and postsurgical care.

Keywords: Hirschsprung disease, constipation, children

Introduction

Hirschsprung's disease (HD) is a congenital disease characterized by the absence of ganglion cells (GC) in the nerve plexus of the intestinal muscularis.[1] The prevalence of this condition ranges from 1 to 1.63 per 10,000 births, with a higher occurrence in males than in females at a ratio of 5 to 1.[2] The most affected segment is the rectosigmoid colon (80%), followed by the sigmoid colon and the entire colon (15% and 5%, respectively).[1]

The clinical manifestations are nonspecific, making diagnosis challenging. Common signs of defecation include vomiting, abdominal distention, and alterations in defecation in early life.[3] Anorectal manometry and contrast enemas are highly useful diagnostic tools, avoiding the necessity of more invasive methods, such as biopsy. Histopathological examinations of rectal biopsies provide a definitive diagnosis, but are not indicated for all patients.[1]

Resection of the abnormal intestinal segment is the preferred treatment; endorectal pull-through and Duhamel pull-through are the most common techniques performed.[4] We describe the case of a 9-month-old female patient with refractory constipation who was diagnosed with HD.

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Case Report

A 9-month-old female with a history of hip dysplasia consulted the emergency room with her mother for 15 days because of absence of stool, vomiting on two occasions, and subjective fever. The mother administered laxatives to her, but they did not produce the desired effects. On physical examination, her vital signs were as follows: heart rate, 159 bpm; respiratory rate: 24 bpm; temperature: 38.5°C; oxygen saturation: 95%; weight: 10 kg; height: 69 cm. Additionally, the abdomen had intestinal sounds, a palpable mass on the hypogastric and anal regions without lesions, and abundant dry depositions.

Initial labs reported mild microcytic hypochromic anemia [hemoglobin 10.3 g/dL, normal range (NR): 11.3-14.1, hematocrit: 32.2%, NR: 31-41%, mean corpuscular volume: 73.3 um^3, NR: 71-88 um^3, mean corpuscular hemoglobin: 23.5 pg, NR: 24-30 pg]; the rest of the laboratories were within NR. Imaging studies revealed simple abdominal radiography with abundant fecal matter and no air-fluid levels (**Figure 1A**). Abdominal ultrasound showing distended intestinal loops with liquid fluid in the lower abdomen.

Pediatricians initially considered fecal impaction, and the patient was treated with enemas and oral laxatives. Her first stool was three days after admission; it was abundant, liquid, and fetid, without blood. Control of simple abdominal radiography using fecal matter in smaller quantities without air-fluid levels **(Figures 1B and 1C)**. The patient continued with refractory constipation due to amplified laboratory findings on suspicion of aganglionic colon; thyroid function was normal, and a barium enema revealed sigmoid elongation with a difference in the maximum transverse diameter recto-sigmoid of 3 cm and slight distal haustration (**Figure 2**). The patient was transferred to another hospital to continue his in-hospital stay at a high-level complex institution. Consent from the patient's mother was obtained before the inclusion of the child for reporting the case in accordance with the institutional ethics guidelines.

Discussion

The first documented cases of these intestinal conditions appeared in the Hindu literature; it was many years until 1691, when Frederick Ruysch reported the first case of congenital megacolon. However, an extensive disease description was realized 200 years later by Harald Hirschprung.[2,5] Since then, many theories have been developed regarding the etiology and physiopathological mechanisms of these uncommon diseases. Okamoto and Ueda in 1967 proposed a theory regarding alterations in the migration of cranial-caudal intestinal GCs, and it was not until the 1990s that the genes involved began to be studied.[5]

Clinical manifestations vary according to the aganglionic segment and age at presentation. In the case of newborns, there are predominantly symptoms that mimic an intestinal obstruction; later, in infants, children, and adults, the symptoms become more unspecific, including vomiting, abdominal distension, and refractory constipation to medications and rectal therapies.[6] Furthermore, the absence of glucocorticoid causes intestinal overactivity with an elevated production of acetylcholine because the affected colon segment has a persistent contraction with a subsequently progressive to secondary dilatation of the non-affected proximal colon.[1]

Anemia is another possible complication of HD. Some theories explain that there is dysregulation in the microecology balance and intestinal mucosa; these microbes regulate the expression of iron transporter genes and stimulate hepcidin, which inhibits the release of iron into plasma.[3] Our patient had a clinical manifestation suggesting constipation although normal treatment was ineffective, which raised suspicion of an aganglionic intestine. Additionally, she experienced mild, low-volume anemia characteristic of iron deficiency anemia. Although it was not possible to determine the complete iron deficiency profile, this alterations may have been associated with her colon involvement.

Imaging diagnostic methods are useful tools and work as screening tests in HD. There are two imaging options

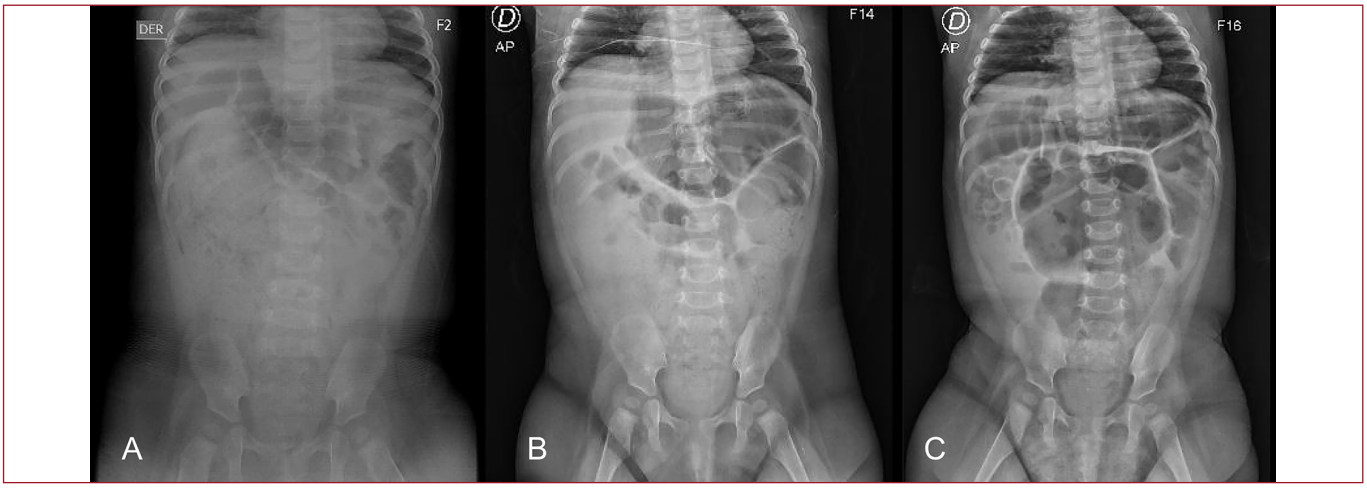

Figure 1. Simple abdominal radiography: A) abundant fecal matter and no air-fluid levels; B and C) control at days three and four showing fecal matter in smaller quantities and without air-fluid levels

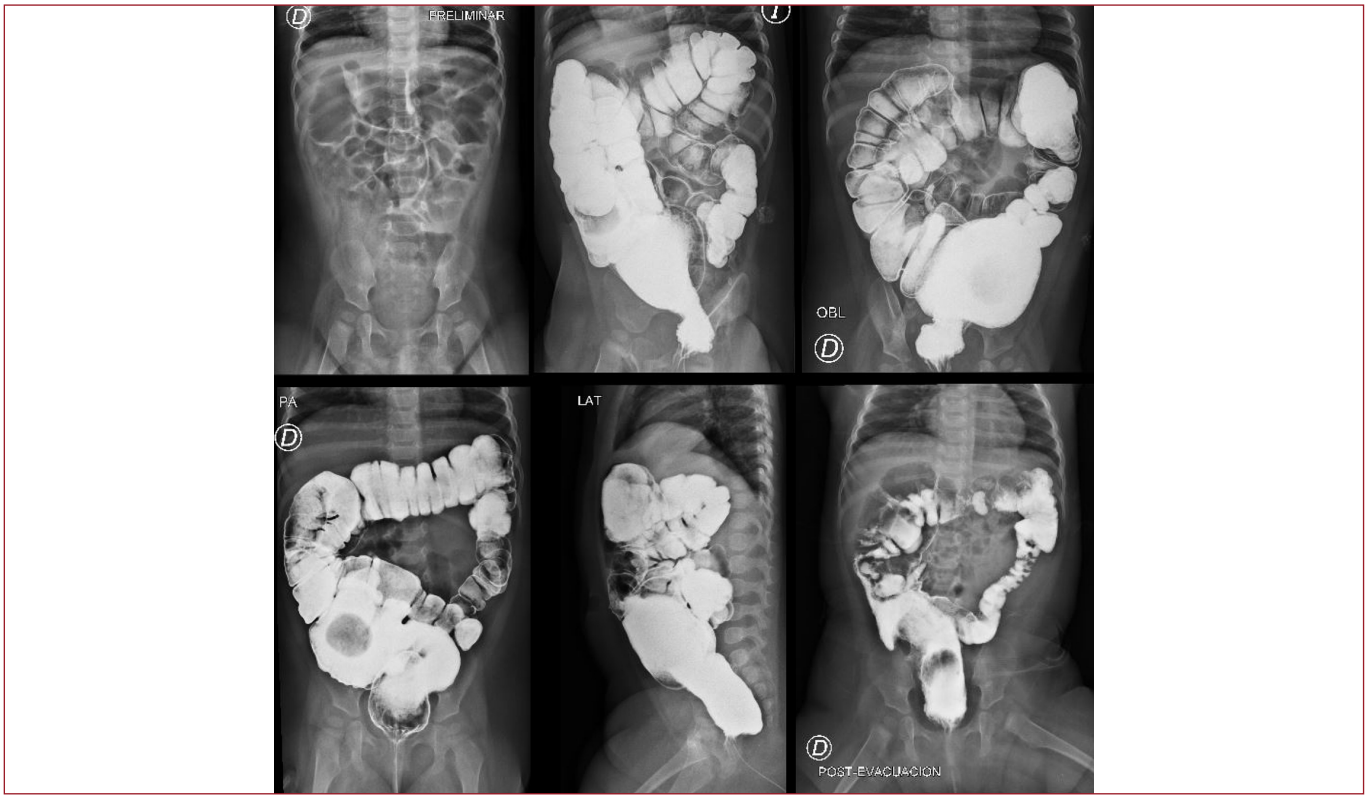

Figure 2. Enema contrast test showing sigmoid elongation with a difference in the maximum transverse diameter recto-sigmoid of 3 cm and slight distal haustration

that are especially useful in HD: Contrast enema and anorectal manometry.[6,7] The contrast enema is a widely available test in which a water-soluble contrast is administered into the colon by a catheter placed inside the anus. After contrast instillation, live images using fluoroscopy are taken.[6] Contrast enema is pathological when it reports a radiographic transition zone with a dilated proximal bowel, contrast retention, irregular mucosa and bowel, and reverse rectosigmoid ratio.[6,7] Notably, normally, the rectum is larger than the sigmoid colon because it is a reservoir and the rectosigmoid ratio is >1. When HD is present, the ratio is inverted because of the impossibility of the rectum to distend it; in contrast, the sigmoid colon starts to distend proximally to store the stool.[7]

Anorectal manometry is the second choice of test for imaging diagnosis of HD; it evaluates different measures of anorectal function and voluntary and involuntary anorectal properties. The procedure is performed using a flexible catheter with sensors introduced into the rectum to allow continuous measurement of pressures.[7] It is considered indicative of HD when the recto anal inhibitory reflex is absent.[6] According to Meinds et al.[8] anorectal manometry is a useful tool for excluding HD diagnosis and avoiding unnecessary rectal biopsy procedures.

Other imaging methods, such as hydrocolonic sonography, can be valuable tools with high sensitivity and specificity for HD diagnosis. This type of study allows for a morphological and vascular assessment of the colon.[9] In addition, histopathological studies using rectal biopsies, when available, are especially useful for definitive diagnosis in cases in which imaging methods are not conclusive or in cases in which there is a specific necessity to perform an invasive procedure such as rectal biopsy. In order to obtain a good specimen for studies, the biopsy should be taken 2 cm above the dentate line and should have a minimum of 3 cm of specimen, of which one-third should be submucosa.[4]

Currently, there are approximately 24 genes associated with this entity; however, the rearranged during transfection gene is the main one involved.[1,2] Approximately 30% of children with HD have other chromosomal and/or congenital anomalies, with Down syndrome being the most commonly associated. However, it has been studied for malformations in different systems, including limbs.[6] The patient's case has a history of hip dysplasia, an entity that has been correlated with multiple gene alteration[10], even though there are no studies that precise a correlation between these entities. Despite advances in medicine, the mortality rate of these conditions is between 2 to 5%.[6] For this reason, a group of qualified professionals is necessary for the treatment and postsurgical care of these patients to obtain better results and prevent complications.[1,4]

Conclusion

HD is a complex entity that has a lower incidence and is predominant in males. This approach is a clinical challenge due to clinical variability, although it should be considered in children with constipation refractory to treatment. Imaging tests are crucial for diagnosis, while histopathological studies provide diagnostic confirmation when available.

Ethics

Informed Consent: Consent from the patient's mother was obtained before the inclusion of the child for reporting the case in accordance with the institutional ethics guidelines.

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Footnotes

Author Contributions: Rojas-Urrea A: Concept, Desing, Analsis or Interpretation, Writing.; Arias-Mariño D: Concept, Desing, Analsis or Interpretation, Writing.; Landines-Peña FE: Desing, Data Collection or Processing, Literature Search, Writing; García-Agudelo L: Concept, Desing, Analsis or Interpretation, Literature Search; Niño-Patarroyo BA: Desing, Data Collection or Processing, Literature Search.

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