Journal of Coloproctology

www.jcol.org.br

Case Report

Late diagnosis of Hirschsprung's disease

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ARTICLE INFO

Article history:
Received 11 December 2014
Accepted 20 February 2015
Available online 27 May 2015

Keywords:
Hirschsprung's disease
Congenital megacolon
Fecal incontinence

ABSTRACT

Hirschsprung's disease is a condition characterized by the absence of ganglion cells in a variable segment of the large intestine, mainly producing the symptom of constipation and being usually diagnosed in the first year of life. With diagnostic methods already established in the literature, the sole treatment is surgery. The objective of this study is to report a case of late diagnosis of the disease at age 13, with symptoms of fecal incontinence in its evolution.

Diagnóstico tardio da doença de Hirschsprung

Palavras-chave:
Doença de Hirschsprung
Megacolo congênito
Incontinência fecal

RESUMO

A Doença de Hirschsprung é uma patologia caracterizada pela ausência de células ganglionares em um segmento variável do intestino grosso, produzindo principalmente o sintoma de constipação, sendo normalmente diagnosticada até o primeiro ano de vida. Com métodos diagnósticos já consagrados na literatura, o tratamento é exclusivamente cirúrgico. O objetivo deste trabalho é relatar um caso de diagnóstico tardio da doença, aos 13 anos, com sintomatologia de incontinência fecal na evolução do quadro.

Introduction

Hirschsprung's Disease (HD), also known as congenital aganglionic megacolon, is an anomaly characterized by an absence of ganglion cells in the myenteric and submucosal plexuses in a variable bowel segment.[1,2]

The absence of ganglion cells results in permanent contraction of the affected segment, preventing the passage of fecal content through that region.[3] The distal functional

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http://dx.doi.org/10.1016/j.jcol.2015.02.009

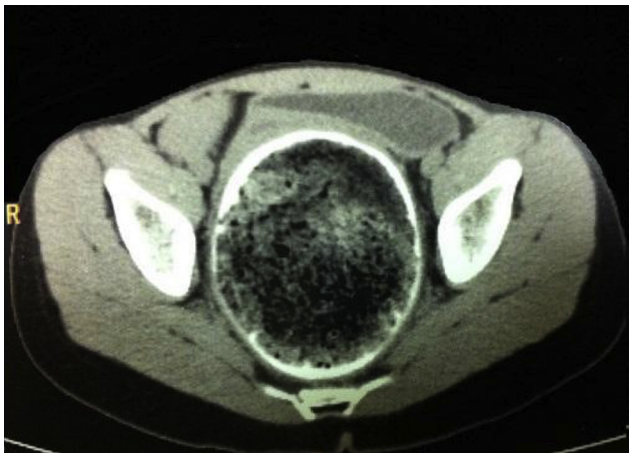

Fig. 1 – Computed tomography (axial plane) showing rectal fecal impaction with megarectum.

obstruction leads to inefficient peristaltic contractions, with consequent dilatation of proximal intestine, producing megacolon.[1]

HD presents itself by symptoms of constipation, such as a greater than 48-h delay in elimination of meconium, abdominal distention and vomiting. In 80% of cases, this disease is diagnosed in the first year of life, being uncommon in adolescence and adulthood; such cases usually appear in the form of an ultrashort segment disease.[4]

The objective of this paper is to report a case of HD in a 13-old patient, with treatment and outpatient follow-up at the Hospital Regional de Mato Grosso do Sul Rosa Pedrossian – HRMS. The infrequency of this diagnosis in this age and the clinical course of this patient underscore the uniqueness of this case.

Clinical case

Female patient, 13 years old, coming from Campo Grande – MS, reports that since birth had intestinal constipation, with mean bowel movements at every 3–5 days with hardened feces, being followed-up by a pediatrician and in treatment for functional constipation. The girl refers onset of fecal incontinence at the age of 12.

Tests ordered: given the unavailability of barium enema, a computed tomography (CT) study was obtained, revealing the presence of rectal fecaloma (Figs. 1 and 2).

A hypothesis of HD was proposed; thus an anorectal manometry was asked, but its result was inconclusive due to patient's lack of cooperation.

Then a rectal biopsy was performed; the pathologist report confirmed the absence of ganglion cells with neurotization of myenteric and submucosal plexuses in a short rectal segment (Fig. 3), confirming an ultra-short form of aganglionosis.

Serology for Chagas' disease was carried out in order to exclude a diagnosis of Chagas' Disease megacolon, with negative result. Thus, the diagnosis of HD was confirmed. Surgical treatment was performed laparoscopically, according to Duhamel-Haddad technique modified with protective

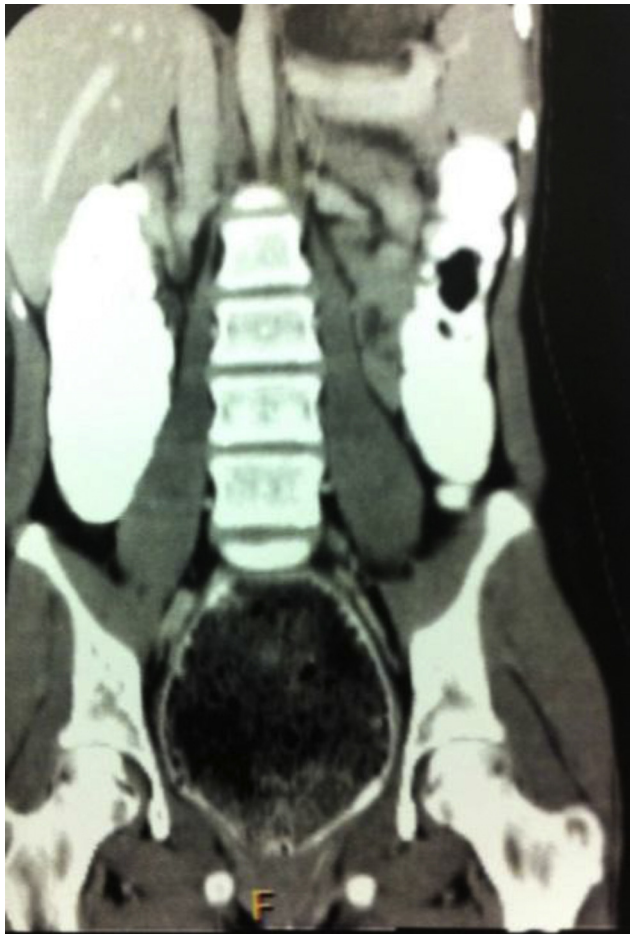

Fig. 2 – Computed tomography (coronal plane) showing rectal fecal impaction with megarectum.

loop ileostomy, with good clinical progression. The girl was discharged on the 5th day after surgery.

Discussion

HD is a congenital anomaly that occurs due to a discontinuation of the cranial–caudal migration of neural crest cells, which are responsible for innervation of the colon, or when the ganglion cells undergo premature death between 5th and 12th weeks of pregnancy.[1–3] It was also suggested that changes in the extracellular matrix in the human embryo gut can interrupt the migration of neural crest-derived cells, producing aganglionosis.[2]

According to the extent of colon involvement, HD is classified into short-segment disease (80% of cases), when the aganglionic segment does not exceed the sigmoid colon; or into long-segment disease, when it outruns the sigmoid colon. In this latter case, HD may affect the entire colon and even the small intestine.[5] HD is still classified into ultrashort-segment disease when it involves only the distal part of the rectum, occurring in 2–3% (in some reports, with estimates of up to 8%) of the cases.[2,6]

HD occurs in approximately 1 in 5000 live births. It can occur alone or in combination with other development