Rev Paul Pediatr. 2016;34(3):388–392

REVISTA PAULISTA DE PEDIATRIA

www.rpped.com.br

CASE REPORT

Hirschsprung's disease – Postsurgical intestinal dysmotility

Mariana Tresoldi das Neves Romaneli, Antonio Fernando Ribeiro, Joaquim Murray Bustorff-Silva, Rita Barbosa de Carvalho, Elizete Aparecida Lomazi*

Faculdade de Ciências Médicas da Universidade Estadual de Campinas (Unicamp), Campinas, SP, Brazil

Received 13 August 2015; accepted 22 December 2015
Available online 12 May 2016

KEYWORDS
Infant;
Hirschsprung's disease;
Gastrointestinal motility

Abstract
Objective: To describe the case of an infant with Hirschsprung's disease presenting as total colonic aganglionosis, which, after surgical resection of the aganglionic segment persisted with irreversible functional intestinal obstruction; discuss the difficulties in managing this form of congenital aganglionosis and discuss a plausible pathogenetic mechanism for this case.
Case description: The diagnosis of Hirschsprung's disease presenting as total colonic aganglionosis was established in a two-month-old infant, after an episode of enterocolitis, hypovolemic shock and severe malnutrition. After colonic resection, the patient did not recover intestinal motor function that would allow enteral feeding. Postoperative examination of remnant ileum showed the presence of ganglionic plexus and a reduced number of interstitial cells of Cajal in the proximal bowel segments. At 12 months, the patient remains dependent on total parenteral nutrition.
Comments: Hirschsprung's disease presenting as total colonic aganglionosis has clinical and surgical characteristics that differentiate it from the classic forms, complicating the diagnosis and the clinical and surgical management. The postoperative course may be associated with permanent morbidity due to intestinal dysmotility. The numerical reduction or alteration of neural connections in the interstitial cells of Cajal may represent a possible physiopathological basis for the condition.

* Corresponding author.
E-mails: elizete.apl@gmail.com, fernando.anferi@gmail.com (E.A. Lomazi).

http://dx.doi.org/10.1016/j.rppede.2016.05.001

PALAVRAS-CHAVE
Lactente;
Doença de Hirschsprung;
Motilidade gastrointestinal

Doença de Hirschsprung – Dismotilidade intestinal pós-cirúrgica

Resumo

Objetivo: Descrever o caso de um lactente portador de doença de Hirschsprung na forma aganglionose colônica total que, após a ressecção cirúrgica do segmento agangliônico, manteve quadro irreversível de obstrução intestinal funcional; discutir as dificuldades no manejo dessa forma da aganglionose congênita e apontar um mecanismo patogenético plausível para o caso.
Descrição do caso: O diagnóstico de doença de Hirschsprung na forma aganglionose colônica total foi definido em lactente aos dois meses de vida, após episódio de enterocolite, choque hipovolêmico e desnutrição grave. Após ressecção colônica, o paciente não recuperou a função motora intestinal que possibilitasse a alimentação via enteral. O exame do íleo remanescente pós-operatório mostrou presença de plexos ganglionares e redução numérica das células intersticiais de Cajal em segmentos proximais do intestino. Aos 12 meses de vida, o paciente mantém-se dependente de nutrição parenteral total.
Comentários: A doença de Hirschsprung na forma aganglionose colônica total tem particularidades clínico-cirúrgicas que a diferenciam das formas clássicas e dificultam o diagnóstico e o manejo clínico-cirúrgico. A evolução pós-operatória pode associar-se à morbidade permanente decorrente de dismotilidade intestinal. A redução numérica ou as alterações das conexões neurais das células intersticiais de Cajal podem representar uma possível base fisiopatológica para a condição.

Introduction

Hirschsprung's disease (HD) is the most prevalent cause of functional bowel obstruction in infants, with an incidence of 1:5000 live births.[1] It is genetically determined and characterized by a defect in the migration of embryonic cells from the neural crest, generating an aganglionic segment at the distal end of the intestines.[2] The gold standard diagnostic method is a rectal biopsy showing absence of ganglion cells and increased number of acetylcholinesterase-positive nerve fibers.[3]

The anatomical location of the transition between the distal aganglionic segment and the proximal ganglionic segment allows for the classification of HD as follows: classic – when the aganglionic segment extends to the proximal sigmoid; with long segment – when aganglionosis reaches the splenic flexure or the transverse colon; or total colonic aganglionosis (HDTCA) – when the aganglionic segment extends from the anus up to at most 50cm proximal to the ileocecal valve. HDTCA presents clinical, histological, and genetic differences in relation to the other types of HD, and is associated with diagnostic and management difficulties.[4] The classic form of HD is observed in 7–88.8% of cases; the long form, in 3.9–23.7%; and HDTCA, in up to 12.6% of patients.[5]

Surgical therapy in HD minimizes the complications of intestinal obstruction when the aganglionic segment is completely resected. In some patients, postoperative intestinal dysmotility persists, most often manifested as chronic constipation and recurrent episodes of enterocolitis. Different histopathologic findings can be identified in these cases, such as incomplete resection of the aganglionic segment, hypoganglionosis, and intestinal neuronal dysplasia juxtaposed to the aganglionic zone.[4]

The present article reports a case in which difficulties of diagnosis, therapy, and prognosis were observed. The publication of this case report was approved by the Institutional Review Board (IRB) of the State University of Campinas, IRB Opinion/Article No°012/2015 from July 28, 2015.

Case description

A black male patient was referred to a tertiary hospital at age 2 months, with a diagnosis of intractable diarrhea and vomiting for 22 days. He evolved with hypovolemic shock and refractory metabolic acidosis. The patient had undergone a simple abdominal radiograph, which showed widespread bowel distension (Fig. 1), and a CT scan showing lack of progression of the enteral contrast to the distal colonic segments.

History of prenatal ultrasound showing distended fetal bowel loops. The mother denied delay (>24h) in the passage of meconium at birth and complaints compatible with intestinal obstruction. The child was born vaginally, at 39 weeks of gestational age, discharged from the hospital on the third day of life, with birth weight of 2950g. Neonatal screenings for phenylketonuria, hypothyroidism, and cystic fibrosis were negative. The child had been in exclusive breastfeeding since birth; the mother denied constipation, but reported poor weight gain. After 38 days of life, the child had diarrhea that required hospitalization in the city of origin. After ten days, he was transferred to a tertiary hospital for intractable diarrhea and metabolic acidosis.

Upon admission to the tertiary hospital, weighting 2860g, he presented generalized muscle atrophy, sparse subcutaneous tissue, and distended abdomen tympanic to