

Normal Habit, Abnormal Distention: Abnormal presentation of Hirschsprung disease

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Introduction

Hirschsprung's disease (HSCR) is characterized by a lack of enteric nervous system ganglion cells (aganglionosis) in a variable extent of distal bowel^{2,3}. It is a developmental disorder in the distal colon resulting in a functional obstruction. Hirschsprung's disease usually can be detected in days after birth as neonate cannot have their bowel opening in the first 24 to 48 hours of life. However, those with short segment that lack normal nerve cells may not exhibit prominent symptoms for several months to years. We present such case that presented at our centre.

Case description

A 15 years old Orang Asli girl, complained chronic abdominal distension for the past 3 years. Initially, she sought treatment at a Health Clinic and was told normal. This time, she was brought in by her school teacher in view of reduced effort tolerance and abdominal distention. The bowel habit was normal. Blood investigations was relatively normal, with urine pregnancy negative. Plain radiography abdominal X-ray was massively faecal-loaded with possible mass. Computed tomography (CT) abdomen revealed very distended large bowel with faecal loaded bowel suggesting undiagnosed Hirschsprung disease.

Discussion

Hirschsprung disease is a developmental disorder of the intrinsic component of the enteric nervous system that is characterized by the absence of ganglion cells in the myenteric and submucosal plexuses of the distal intestine¹. Because these cells are responsible for normal peristalsis, patients with Hirschsprung disease present with functional intestinal obstruction at the level of aganglionosis. Incidence of Hirschsprung disease is approximately 1 in 5000 live-born infants¹. In this report, older children may be presented later with abdominal symptoms. The complications that may arises if being untreated can be life threatening, and results in high mortality rate as high as 80%.

Conclusion

This report highlights that Hirschsprung's disease may present as chronic abdominal distention. This case also shows that the disease could occur despite normal bowel habit. Doctors managing such case should be aware of this possibility despite the relatively late age group and preservation of bowel habit.

Keywords

“Hirschsprung's disease”, “abdominal distension”, “ganglion cells”

References:

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