

# Isolated Intestinal Neuronal Dysplasia- Type B of Ileum: a Rare Occurrence

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## Case Report

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# Abstract

**Background:** Intestinal neuronal dysplasia type B in the gastrointestinal tract is a rare occurrence and may occur alone or in combination with Hirschsprung disease. Distal colon seems to be frequent site for isolated IND-B cases, however small bowel involvement is scarcely reported.

**Case presentation:** We report a case of 9 years old boy presenting with features of intestinal pseudo-obstruction for 5 years. Exploratory laparotomy revealed narrowed distal ileum with huge proximal dilatation. Histopathology of the resected terminal ileum revealed giant submucosal ganglion, hyperplastic submucosal nerves and ectopic ganglion cells in the lamina propria suggestive of IND-B.

**Conclusions:** Although IND-B involving ileum is a rare occurrence, suspicion should be kept in cases of intestinal obstruction with minimal response to conventional treatment.

## Background

Intestinal neuronal dysplasia type-B (IND-B) is a rare congenital malformation of gastrointestinal innervation caused by dysplastic embryonal development of the enteric nervous system. The changes associated with IND-B are more common in the distal colon; however, they can affect any segment of the enteric nervous system and occur in different age groups ranging from newborns to adult, alone or in combination with Hirschsprung disease (HD) (1). Very rarely isolated ileal involvement has been reported (2). We herewith report a case of IND-type B in the ileum of a child presenting as chronic intestinal pseudo-obstruction (CIP).

## Case Presentation

A 9 years old boy presented with intermittent abdominal pain and fullness sensation since 4 years of age. He had history of poor bowel habits with encopresis. Antenatal and natal histories were uneventful. Histories of delayed passage of meconium, recurrent vomiting, retentive posturing or blood in stool were absent. Child had 2–3 episodes of febrile seizure which remitted on its own. He had received empirical anti-tubercular therapy for duration of 11 months for the presenting complaint, although there were no other systemic symptoms. There was no significant family history.

On general examination child's weight was 24.8 kg (3rd-10th centile), height 138 cm (10th – 50th centile) with failure to thrive. There was no pallor, icterus, edema or lymphadenopathy. On abdominal examination, inspection revealed distended abdomen with few prominent veins and visible peristalsis and borborygmi was present. There was no organomegaly on palpation. On per-rectal examination, there were no fissures, tags or gush of air and soft stool was palpable. Cardiovascular, respiratory and neurological examination showed no abnormalities.

On upper gastrointestinal endoscopy, oesophagus and stomach were normal. X-ray abdomen, erect posture, showed dilated small bowel loops with multiple air fluid levels. Computed tomography of

abdomen showed collapsed proximal jejunum and terminal ileum with dilated lower small bowel loops (Fig. 1). Patient was evaluated for pseudo-obstruction. Exploratory laparotomy revealed narrowed distal ileum (approximately 15 cm) with huge proximal dilatation. Differential diagnosis of intestinal pseudo-obstruction, hirschsprung disease and celiac disease were kept. Resection of distal ileum, cecum and appendix measuring 13 cm in length with end to end ileo-ascending colon anastomosis was performed. No apparent dilatation of the segment was noted. Histopathology of the terminal ileum revealed giant submucosal ganglion (average 10–14 ganglion cells per ganglion), hyperplastic submucosal nerves and ectopic ganglion cells in the lamina propria. Muscularis propria was largely unremarkable however, serosal fat revealed hypertrophic nerve fibers. Histopathology was suggestive of IND-B (Fig. 2). Cecum and appendix were unremarkable.

On follow-up at one month, bowel prolapse from distal stoma site was noted. An exploratory laparotomy revealed proximal 20 cm of ileum anastomosed to ascending colon was hugely dilated. Another ileal resection was done till the normal calibre was identified with ileo-ascending colon reanastomosis. Histopathology of the dilated part showed similar findings. At 2 months follow-up, child is now gaining weight and passing formed stools.

## Discussion And Conclusions

The patient presented with history of CIP. On laparotomy narrowed distal ileum with proximal dilatation was present. Histopathology revealed intestinal neuronal dysplasia- type B of ileum. Similar findings were also present in the ileal segment found dilated and non-functional during second surgery.

The broad spectrum of pediatric enteric neuropathy with hyperganglionosis includes ganglioneuroma and IND-B. IND-B is considered to be a malformation probably resulting from the incomplete malformation of the submucosal plexus. The hallmark feature being increased “giant” submucosal ganglia (> 8 ganglion cells per ganglion) (1). Ganglioneuroma on the other hand are mucosal polypoid or diffuse types. The diffuse ganglioneuromatosis have hyperplasia of neural and glial elements in both the submucosal and myentric plexuses. They are often associated with multiple endocrine neoplasia type 2b (MEN2b) and neurofibromatosis type 1(NF-1). For pathologists it is important to classify the anatomical different forms of intestinal pseudo-obstruction for the appropriate management and follow-up.

Isolated IND-B presents as a chronic condition with signs and symptoms similar to HD. Findings range from chronic constipation and obstructive signs including abdominal distension and vomiting. Associated anomalies like anal stenosis, intestinal malrotation and malformations have been reported in 30% of cases (3, 4). The diagnostic workup must focus to rule out organic causes of intestinal constipation, particularly HD, the most prevalent intestinal dysganglionosis. However, anorectal manometry and barium enema, established tests for HD screening, are non-specific for IND-B (5). Thus, the diagnosis of IND-B essentially relays on histopathological analyses of rectal biopsies (6).

In recent years, IND-B in adults has shown rising trends. Some of them have experienced symptoms of severe constipation since childhood while others had onset of symptoms at adulthood (2, 7, 8). Our case

was a child with symptoms since 4 years of age. Prognosis of IND-B is usually good, but symptoms may progress to serious complications like CIP, acute bowel obstruction or intestinal infarction. The intestinal resections should be the treatment for the refractory cases showing no improvement with at least 6 months of conservative management, or in patients having obstructive complications (1, 9, 10).

The present case showed classic features of IND-B with changes chiefly involving the submucosal plexi with increased density of ganglion cells. The IND-B involving only the ileum is a very rare occurrence (2).

Suspicion for IND-B involving ileum, although rare, should be kept in patients presenting with intestinal obstruction and showing minimal response to conventional treatment.

## **Abbreviations**

CIP: chronic intestinal pseudo-obstruction

HD: Hirschsprung Disease

IND-B: Intestinal Neuronal Dysplasia type-B

MEN 2b: Multiple Endocrine Neoplasia type 2b

NF-1: Neurofibromatosis type 1(NF-1)

## **Declarations**

### **Ethics approval:**

Not applicable

### **Consent to participate:**

The written informed consent was taken from the patient's parents.

### **Funding:**

None.

### **Conflicts of interest/Competing interests:**

None.

### **Availability of data and material:**

The data will be available on the reasonable request to the author.

### **Author contribution:**

**Pratishtha Sengar:** Writing and editing manuscript

**Ram Nawal Rao:** Concept and design of the study

**Conflict of interest statement:**

There is no conflict of interest and work is original.

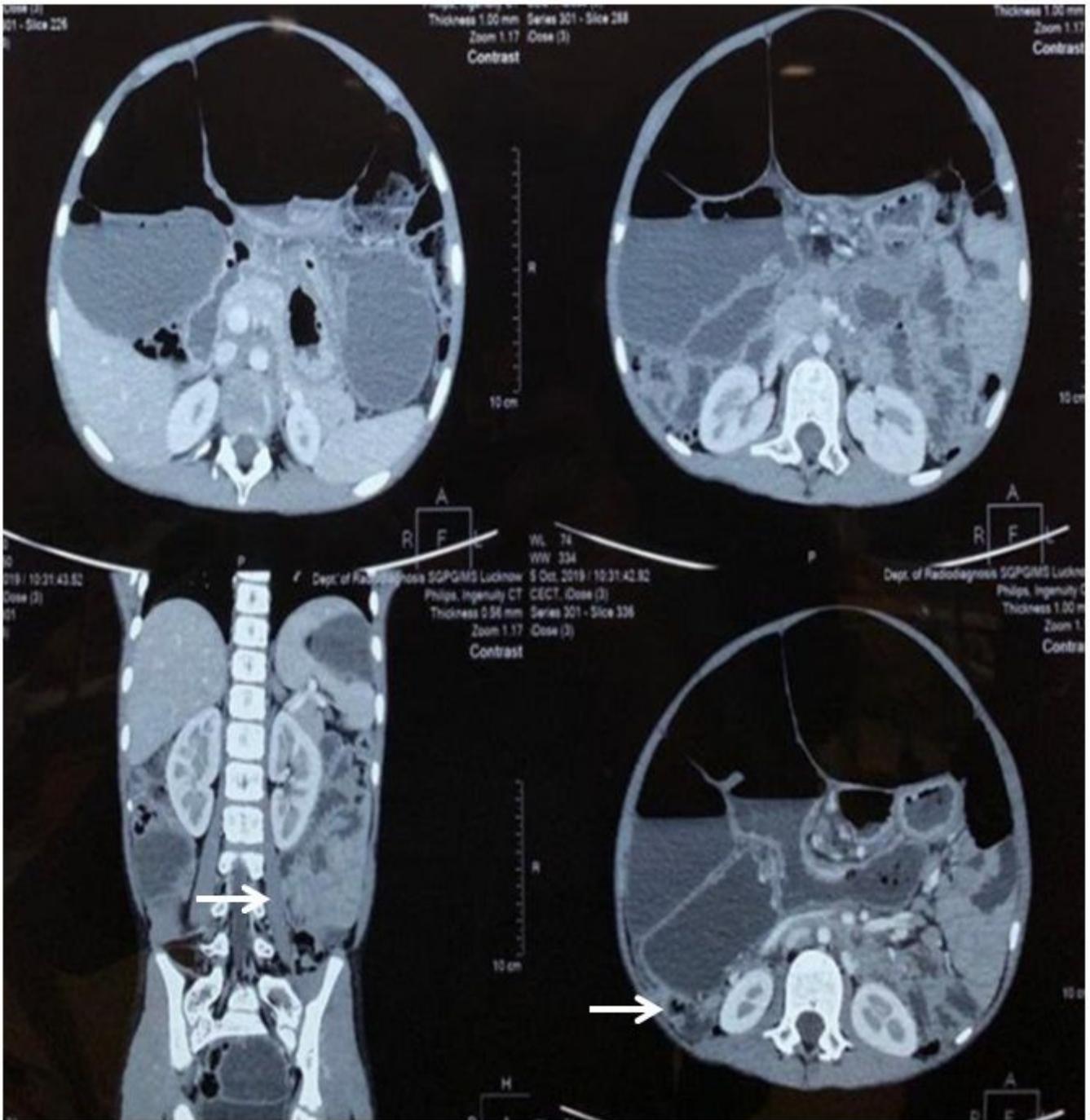
**Acknowledgement:**

We acknowledge the patient's parents for giving informed consent.

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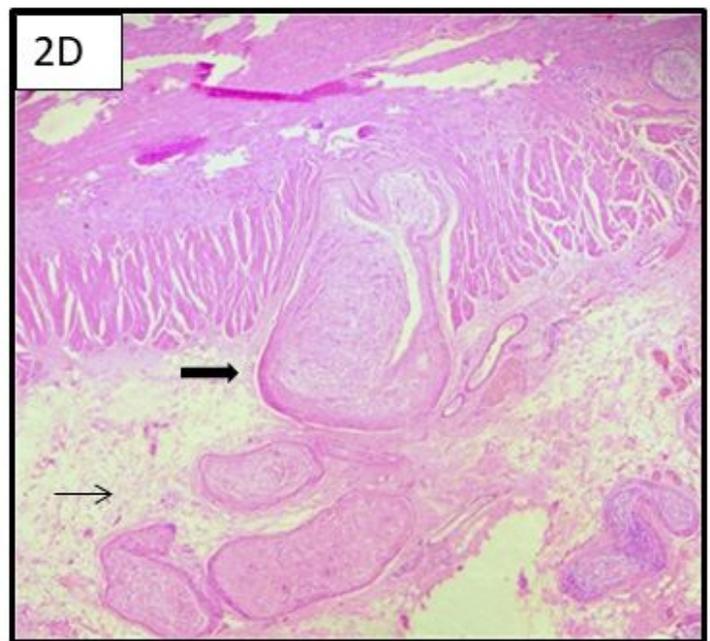
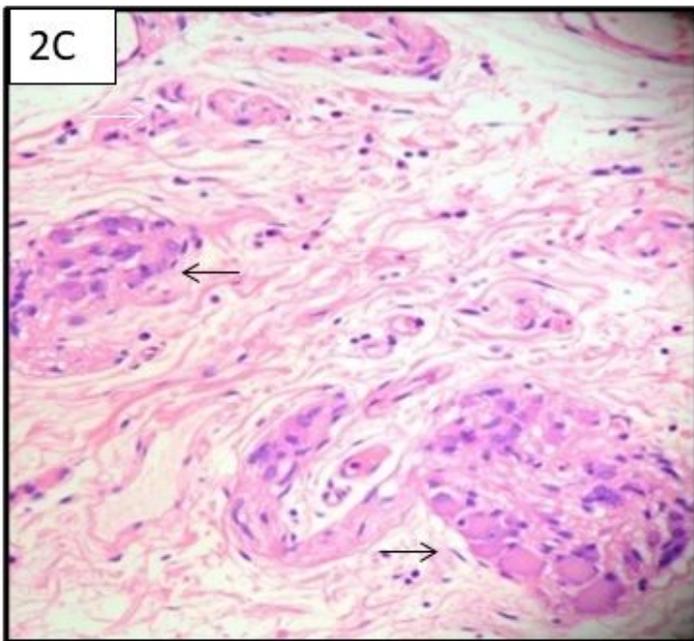
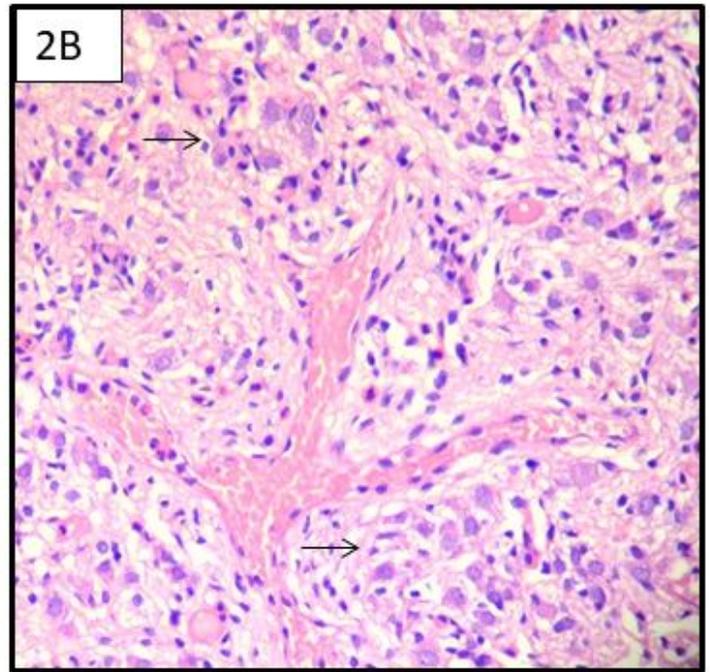
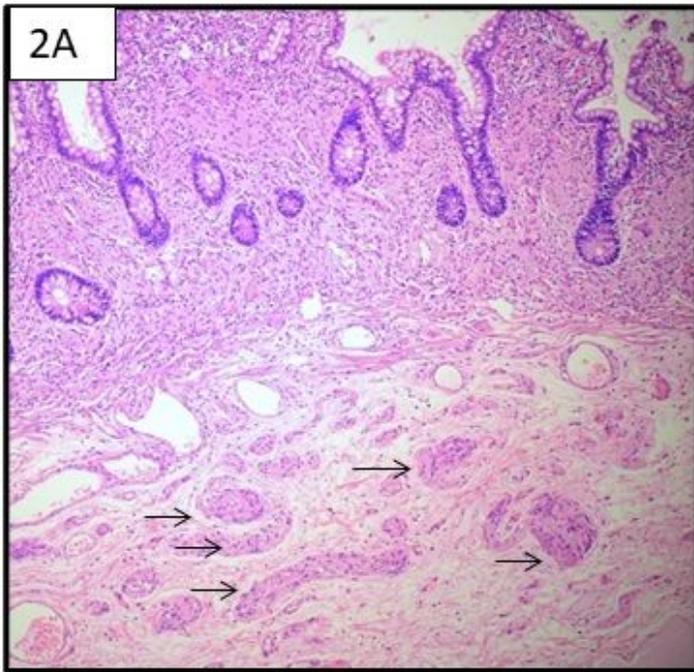
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## Figures



**Figure 1**

CT abdomen shows multifocal dilations maximum 7 cm of lower small bowel with collapsed proximal jejunum and terminal ileum (arrows).



**Figure 2**

2A) Ileum showing mucosa and submuosa with submucosal ganglion hyperplasia highlighted by arrows (H&E, 200x). 2B) Lamina propria also reveals ectopic ganglion cells (arrows) around the crypts and centered around the capillaries (H&E, 400x). 2C) Giant submucosal ganglia (H&E, 400x). 2D) Serosal fat with hypertrophic nerve fibers (thin arrow), occasional fibers seen insinuating into the muscularis propria highlighted by solid arrow (H&E, 200x).

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