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Intestinal Neuronal Dysplasia presenting at adulthood: A Case Report

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Authors' contributions

This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Intestinal Neuronal Dysplasia (IND) is a rare and complex gastrointestinal disorder that affects the enteric nervous system. It is described as a congenital malformation of gastrointestinal innervation and is caused by dysplastic embryonal development of the enteric nervous system. IND is potentially associated with mild and chronic gastrointestinal motility disturbances and is rarely reported in adults.

We present the case of a 31-year-old female, who presented with gross distension of abdomen along with abdominal pain, constipation and difficulty in breathing. The initial management of the patient included immediately intubation and needle decompression as the increased intra-abdominal pressure was severely restricting lung expansion on Chest X-ray. A CT done subsequently revealed an extensive dilatation of small and large bowel loops proximal to the distal sigmoid colon with partial rotation of mesentery seen in distal sigmoid. A total colectomy with Hartmann's procedure was performed with an ileostomy in the RIF. Microscopic examination

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revealed features that were consistent with Intestinal Neuronal Dysplasia: completely ganglionated plexuses with very subtle alterations in morphology, size and number of ganglia. A few crowded miniatured ganglia were seen along with “giant ganglia”; a characteristic microscopic finding for IND. Hence, a diagnosis of Intestinal Neuronal Dysplasia was established.
This case highlights the challenges in the diagnosis and management of IND, and the need for further research to better understand the pathophysiology of this disorder and to develop evidence-based guidelines for its diagnosis and management.

Keywords: Intestinal neuronal dysplasia type B; enteric nervous system; ganglion cells; myenteric plexus; surgery.

1. INTRODUCTION

Intestinal Neuronal Dysplasia is a rare and complex gastrointestinal disorder that affects the enteric nervous system (ENS) [1]. The ENS is a network of nerve cells that regulates the movement of food and waste products through the digestive system [2]. In individuals with IND, the ENS is affected by abnormal development of the intestinal neurons, leading to a variety of symptoms including chronic constipation, abdominal pain, vomiting, and intestinal obstruction [3,4].

IND can affect individuals of any age, but it is most commonly diagnosed in infants and young children [5]. The exact cause of IND is not well understood, but it is believed to be related to genetic or environmental factors that disrupt normal development of the ENS [6,7].

IND can be divided into two distinct subtypes: IND type A and IND type B IND type A, also known as hypoganglionosis or aganglionosis, presents in the neonatal period and comprises of 5% of all cases and is characterized by congenital aplasia or hypoplasia of the sympathetic innervation. This leads to a loss of peristaltic activity and subsequent accumulation of fecal matter, causing chronic constipation and abdominal distension.

IND type B, also known as hyperganglionosis or ganglioneuromatosis, comprises of 95% of all cases of IND [8] and is characterized by an excess of ganglion cells in the myenteric plexus of the ENS .It is caused by the malformation of the parasympathetic submucosal and myenteric plexus and is found in adults as well as in children [8]. IND type B leads to abnormal peristaltic activity and causes abdominal pain and malabsorption. It's worth noting that some cases of IND may have features of both type A and type B dysplasia, making the classification of IND more complex. In addition to these two main types, IND can also be classified based on the location of the affected segment of the intestine. For example, IND can affect the entire length of the intestine (total colonic IND), or it may be restricted to a specific segment of the intestine (segmental IND).

Overall, the classification of IND is based on a combination of histopathological features and clinical presentation, and can be challenging due to the complexity of the disorder.

Diagnosis of IND can be again challenging, as symptoms can be nonspecific and may mimic other gastrointestinal disorders. Diagnosis is typically made based on a combination of clinical presentation, histopathological analysis of a biopsy specimen, and exclusion of other potential causes of gastrointestinal symptoms.

The management of IND is primarily symptomatic and varies depending on the severity of symptoms. Treatment options include dietary modifications, laxatives, enemas, and surgical intervention. However, the efficacy of these treatments is variable, and there is no consensus on the optimal management strategy for IND.

Despite being first described over five decades ago, the pathophysiology of this disorder remains poorly understood, and the diagnosis and management of IND remain a challenge for clinicians. In this research paper, we aim to describe a case of a patient presenting with Intestinal Neuronal Dysplasia at adulthood.

2. CASE PRESENTATION

A 31-year-old female presented to the Emergency Department with complaints of gross distension of abdomen along with abdominal pain, constipation and difficulty in breathing. General clinical examination revealed a poorly built and poorly nourished female with cyanosis.

Abdominal Examination revealed gross abdominal distension with widespread tympany, no evidence of bowel sounds and a tense abdominal wall. An ABG revealed severe acidosis and electrolyte imbalance with inability to maintain oxygen saturation. Initial management of patient included immediate intubation and needle decompression, as the increased intra-abdominal pressure was severely restricting lung expansion on Chest X-ray.

A subsequent CT scan revealed extensive dilatation of small and large bowel loops proximal to the distal sigmoid colon and a partial rotation of the mesentery of distal sigmoid colon was also noted. The stomach and the duodenum appeared compressed and Pneumobilia was also identified.

CT scan was followed by blood investigations, which yielded normal results except for anemia and mild leukocytosis.

A decision to do a laparotomy was taken, in view of the clinical condition and CT scan findings. During Laparotomy, gross dilatation of large bowel was noted up to the rectosigmoid junction with a sigmoid volvulus.

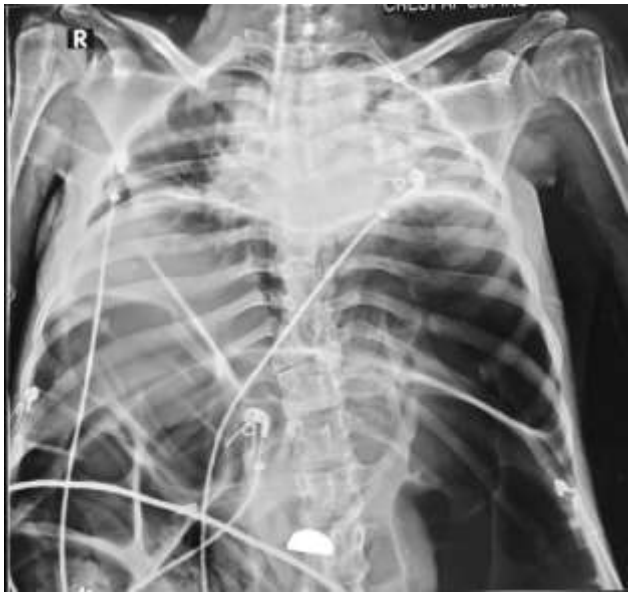
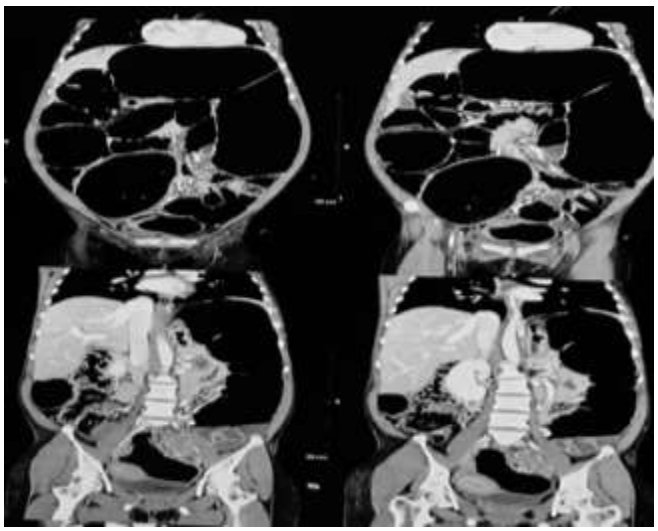


Fig. 1. Pre-operative Chest X-ray showing massive dilated large bowel (megacolon) pushing the diaphragm up, causing poor lung expansion



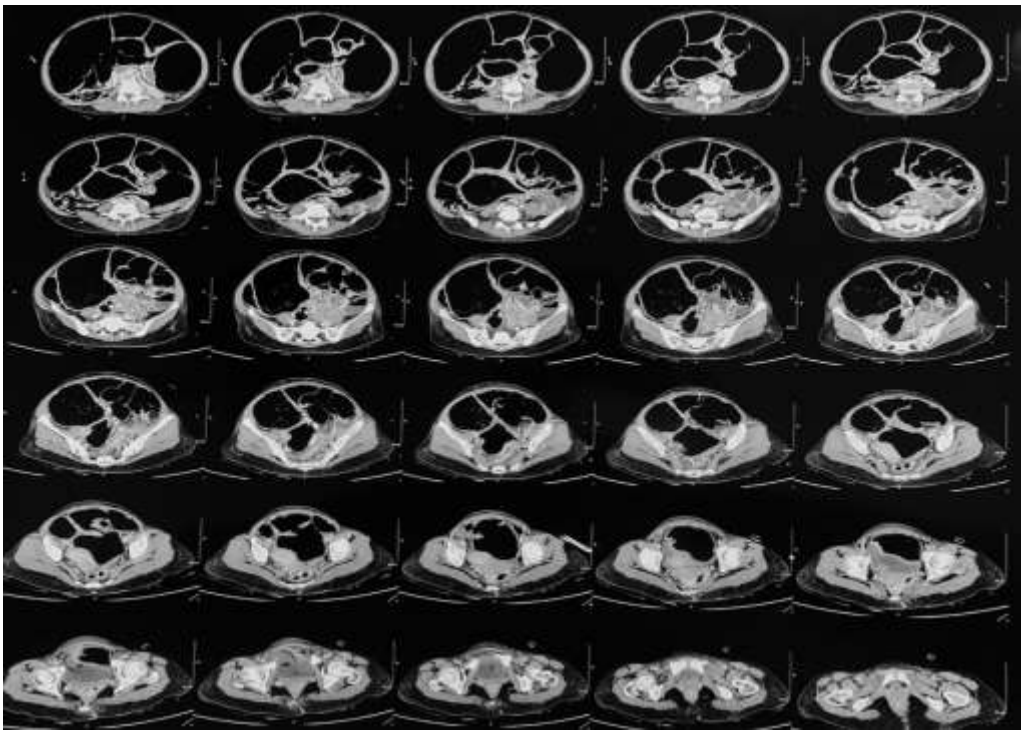


Fig. 2 a. & b. Abdominal Computed Tomography scan showing massive bowel dilatation with no mechanical obstruction

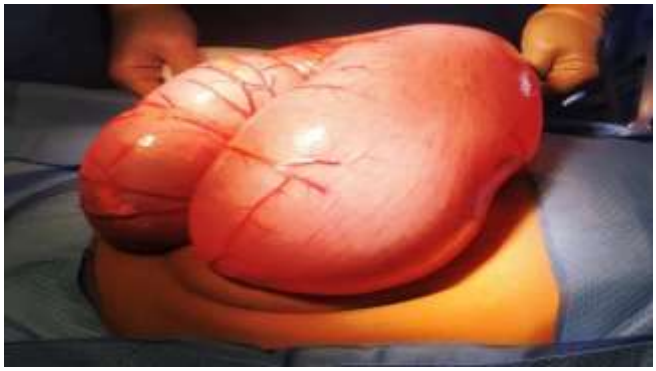


Fig. 3a. & b. Intraoperative picture showing massively dilated large bowel and a decision was undertaken to do a total colectomy with Hartmann's procedure along with an ileostomy in the RIF

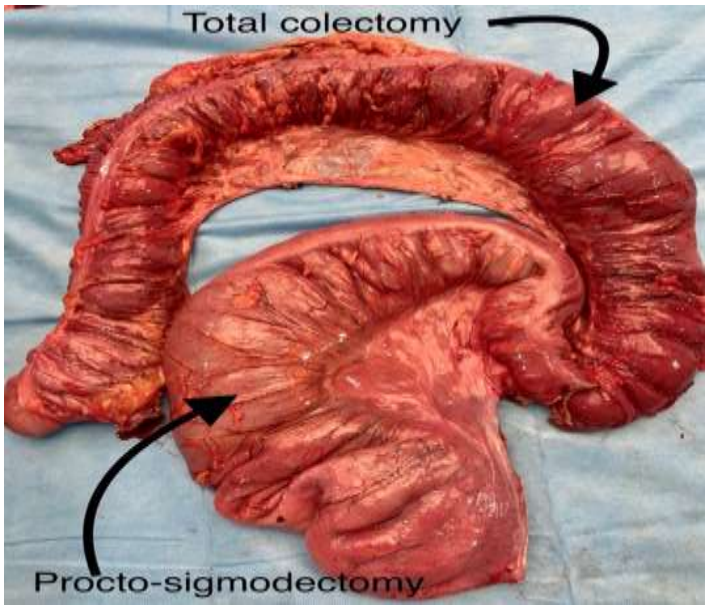


Fig. 4. Postoperative Specimen of Total Colectomy

Gross examination of the specimen showed thick walled and fibrosed large bowel. Mucosa was flattened, ulcerated, congested and erythematous. The lumen was markedly dilated, and no specific lesion or polyp was seen. Microscopic examination and staining revealed completely ganglionated plexuses with very subtle alterations in morphology, size and number of ganglia. A few crowded miniatured ganglia were seen along with giant ganglia. Minimal desmosis in some areas and serosal congestion was present. No ganglionitis or mucosal pathology was noted and no melanosis was present.

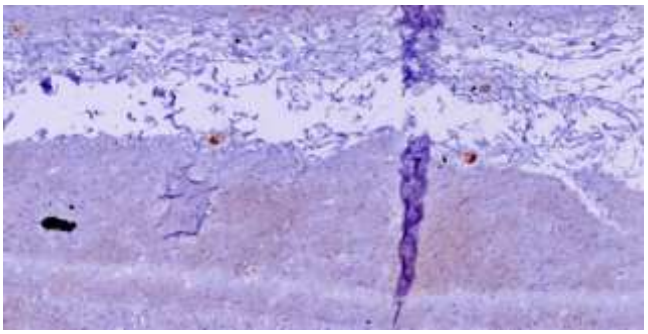


Fig. 5. Calretinin stain

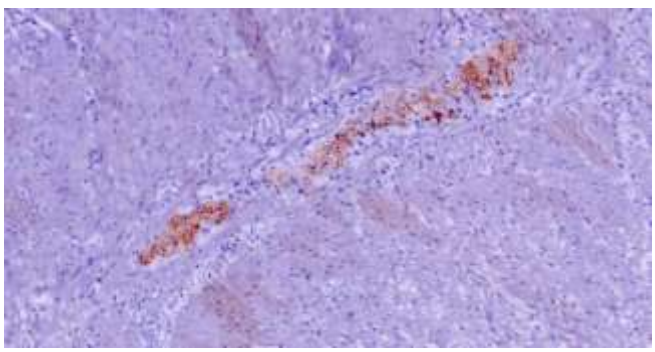


Fig. 6. Synaptophysin stain A diagnosis of Intestinal Neuronal Dysplasia, which is a very rare disorder in adults, was made

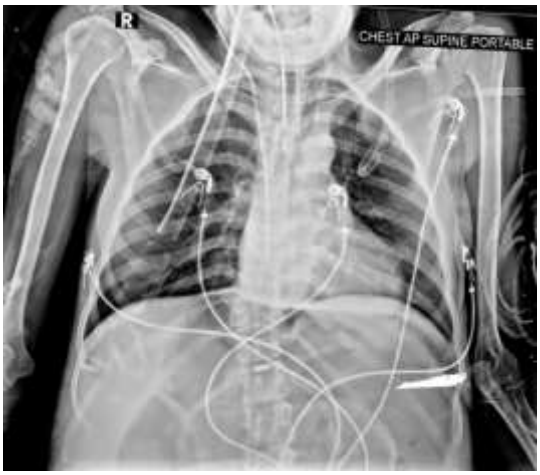


Fig. 7. Post operative chest X-Ray

The patients condition improved post-operatively and the post-operative Chest X-Ray showed good expansion of lung.

The patient was discharged on Post-operative day 10, on full diet and a functioning ilestomy.

3. DISCUSSION

In this case report, we presented the clinical and histopathological features of a patient with Intestinal Neuronal Dysplasia (IND), a rare and complex gastrointestinal disorder that affects the enteric nervous system. Our patient presented with a history of chronic constipation, abdominal pain, and vomiting, which are typical symptoms of IND. The histopathological analysis of the surgical specimen revealed the presence of abnormal ganglion cells and nerve fibers in the submucosal and myenteric plexuses of the intestine, confirming the diagnosis of IND. This finding is consistent with previous reports in the literature, which have described similar abnormalities in the enteric nervous system of patients with IND.

The management of IND is challenging, as there is no consensus on the optimal treatment strategy. In our case, we opted for a surgical approach, which has been shown to provide symptomatic relief in some patients with severe or refractory symptoms. However, the long-term outcomes of surgical intervention for IND remain unclear, and further studies are needed to evaluate the efficacy and safety of this approach.

Another important aspect of IND is its potential association with other neurocristopathies, such as Hirschsprung's disease and neurofibromatosis type 1. Therefore, a comprehensive evaluation of the patient, including a thorough medical history and physical examination, is crucial to identify any potential underlying genetic or syndromic conditions that may be associated with IND.

Our case report highlights the diagnostic and therapeutic challenges of IND and underscores the need for further research to better understand the pathophysiology and optimal management of this rare disorder. Clinicians should maintain a high index of suspicion for IND in patients presenting with chronic constipation and other gastrointestinal symptoms, particularly in the absence of a clear etiology.

4. CONCLUSION

In conclusion, our case report highlights the importance of a multidisciplinary approach to the diagnosis and management of IND, involving gastroenterologists, pathologists, and surgeons. By increasing awareness of this rare but important disorder, we hope to improve the outcomes of patients affected by IND and to stimulate further research in this field.

CONSENT

As per international standards or university standards, patient(s) written consent has been collected and preserved by the author(s).

ETHICAL APPROVAL

As per international standard or university standards written ethical approval has been collected and preserved by the author(s).

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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