

CASE REPORT

INTESTINAL NEURONAL DYSPLASIA, TYPE B - A CASE REPORT

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HOW TO CITE THIS ARTICLE:

H. Lakshmi Vasavi, E. Kiran Kumar. "Intestinal Neuronal Dysplasia, Type B - A Case Report". Journal of Evidence based Medicine and Healthcare; Volume 2, Issue 15, April 13, 2015; Page: 2339-2342.

ABSTRACT: Intestinal neuronal dysplasia (IND) is one of the conditions known to mimic Hirschsprung's disease (HD) both clinically and radiologically, and clinically presents with intestinal obstruction in pediatric age group. Two types are described.¹ IND Type A is characterized by a malformation of the adrenergic nerve supply to the blood vessels and IND Type B, in which the submucosal plexus is involved. Giant ganglia are involved in Type B, and there is abnormal morphology of the nerve cell groups and nerve cell fibres.² In the present case, diagnosis was established mainly by biopsy and microscopic examination of H&E stained sections, which show increased ganglion cells both in number and size, and confirmed by Cathepsin D immunostaining.

KEYWORDS: Intestinal neuronal dysplasia, Ganglion cells, Hirschsprung's disease, Cathepsin D.

CASE REPORT: A 4 year old male child presented clinically with the complaints of chronic constipation and abdominal discomfort. There was a history of previous clinical consultations and symptomatic treatment without causing any relief of symptoms in the last 6 months. Provisional clinical diagnosis of chronic constipation with possibility of intestinal obstructive disease was made and the patient was advised for hospitalization for further investigations and management. On clinical examination, abdominal distension was present. Peristaltic activity was noted on auscultation and X-ray was advised. Plain X-ray revealed air fluid levels in the colon. As HD is possible in this age group, contrast enema was also done, which showed normal caliber rectum with narrow distal segment, a funnel-shaped dilation at the level of the transition zone, and a marked dilation of the proximal colon. HD was diagnosed on the basis of radiological findings. Suction rectal biopsy was advised for confirmation, and suction biopsy specimens were taken at 2cm, 3cm and 5cm, above the dentate line. The optimal size of the biopsy was 3.5mm diameter, so as to include submucosa. The biopsy tissue was formalin-fixed and paraffin-embedded, and stained with H&E for microscopic examination.

Microscopic Findings: Forty serial sections were studied which showed the occurrence in the submucous plexus, of abnormally increased number of giant ganglion cells, appearing as enlarged, multipolar, polygonal-shaped, with amphophilic to eosinophilic vacuolated cytoplasm and oval to round nucleus, with prominent nucleoli.

DISCUSSION: IND is one of the important causes of functional intestinal obstruction in pediatric age group, commonly mistaken for HD.³ In HD there is absence of ganglion cells (aganglionosis), with hypertrophied nerve bundles, whereas in IND, there is abnormally increased nerve fibres with increased number of giant ganglion cells (Hyperganglionosis), with an average of 7-10. IND

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is one of the intestinal dysganglionosis and clinically closely associated with HD, and hence it is a variant of HD.⁴

⁵ other variants of HD are intestinal ganglioneuromatosis, isolated hypoganglionosis, immature ganglia, etc.⁶ There is a role of histochemical and immunohistochemical techniques, which aid in the identification of nerve fibres and ganglion cells. AchE is used as a histochemical stain for identifying hyperplastic nerve fibres, S-100, NSE, Chromogranin, NFP (Nerve filament protein), GFAP and NGFR are used in immunohistochemistry to demonstrate nerve fibres.⁷ Cathepsin – D is a specific antibody used for the identification of both mature and immature ganglion cells, but it does not show immunostaining of nerve fibres.⁸

CONCLUSION: IND should be differentiated from HD, and has to be kept in mind as one of the causes of intestinal obstruction in pediatric age group. This is done on the basis of microscopic findings in H&E stained slides, and with the aid of immunohistochemistry for confirmation, whenever needed.

ACKNOWLEDGMENTS: We acknowledge the colleagues and the technical staff of the department for the cooperation and support.

REFERENCES:

1. Fada B, Prister G, Meier - Ruge W et al. Symptoms, diagnosis and therapy of neuronal intestinal dysplasia masked by Hirschsprung's disease. *J Pediatr surg Int.*1987; 2:76 – 80.
2. Scharli AF. Neuronal intestinal dysplasia. *Pediatric surg Int.*1992; 7: 2-7.
3. Scharli AF, Meier – Ruge W. Localised and disseminated forms of neuronal intestinal dysplasia. *J Pediatric surg.*1981; 16: 164-70.
4. Peng SS, Lee WT, Hsui WM. An unusual cause of abdominal distension in a 4-year old boy with neuronal intestinal dysplasia. *Gastroenterology.*2011 Oct; 141(4):3-4.
5. Friedmacher F, Puri P. Classification and diagnostic criteria of variants of Hirschsprungs disease. *Pediatr Surg Int.* 2013 Sep; 29(9):855-72
6. Geramizadeh B, Akbarzadeh E, Izadi B, Foroutan HR, Heidari T. Immunohistochemical study of enteric nervous system in hirschsprung's disease and intestinal neuronal dysplasia. *Histol Histopathol.*2013 Mar; 28(3): 345-51.
7. Meier-Ruge WA, Bruder E, Kapur RP. Intestinal neuronal dysplasia type B: one giant ganglion is not enough. *Pediatr Dev Pathol.* 2006 Nov-Dec; 9(6):444-52.
8. Skaba R, Frantlova M, Horak J. Intestinal neuronal dysplasia. *Eur J Gastroenterol Hepatol.* 2006 Jul; 18(7):699-701.

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Fig. 1: Intestinal neuronal dysplasia type B, showing number of giant ganglion cells in the sub mucous plexus. (H & E 10X).

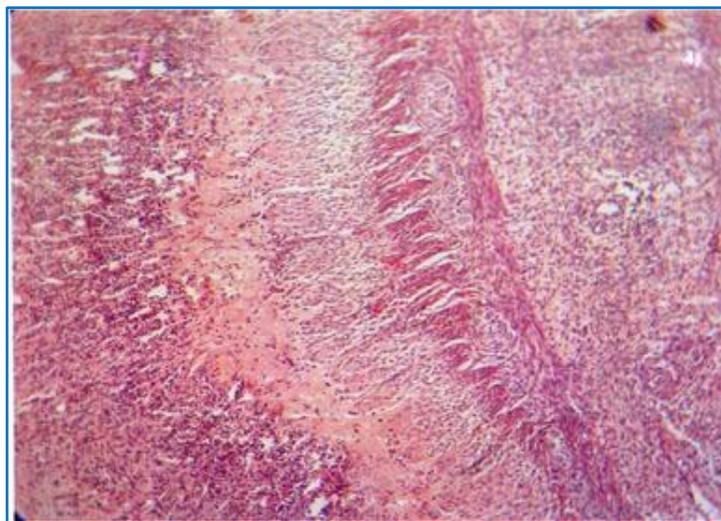


Fig. 1

Fig. 2: Intestinal neuronal dysplasia type B, showing giant ganglion cells in the sub mucous plexus. (H & E 40X).

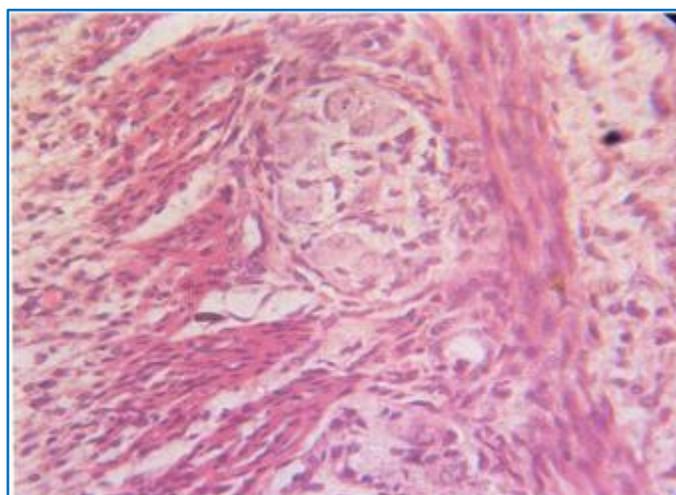


Fig. 2

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Fig. 3: Intense granular cytoplasmic reactivity with collarette around nucleus, in the ganglion cells. (Cathepsin D 40X).

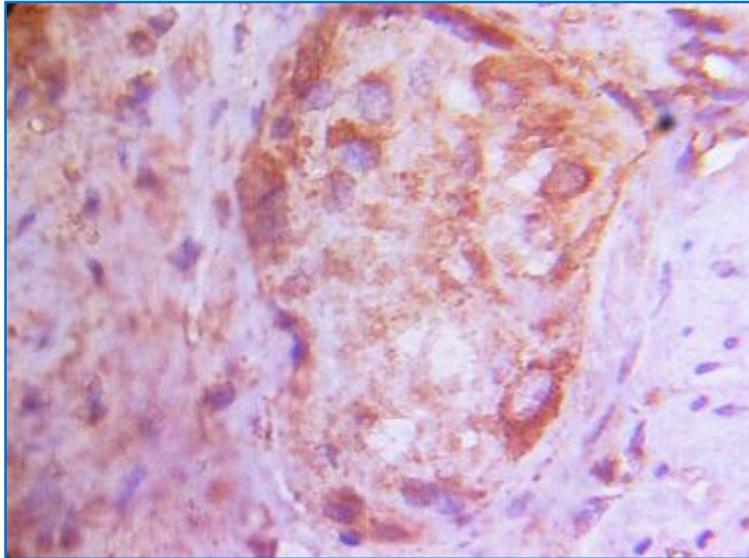


Fig. 3

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Date of Submission: 29/03/2015.
Date of Peer Review: 30/03/2015.
Date of Acceptance: 04/04/2015.
Date of Publishing: 13/04/2015.