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Lhermitte-Duclos Disease: Diagnosis on MRI, MR Spectroscopy, CT and Positron Emission Tomography

NEERAJ JAIN[1], UDIT CHAUHAN[2], VANDANA GOEL[3], SUNIL KUMAR PURI[4]

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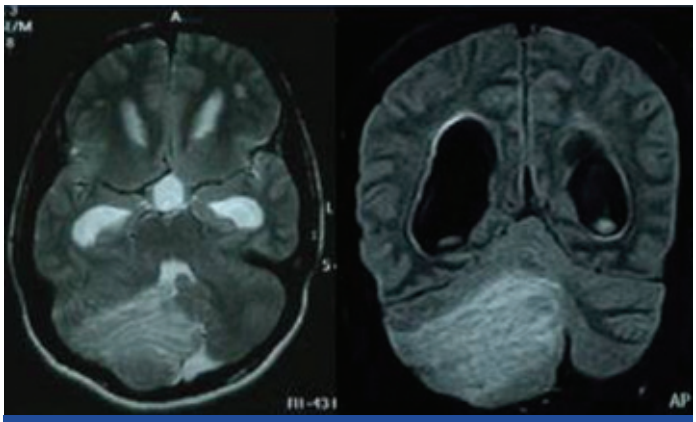
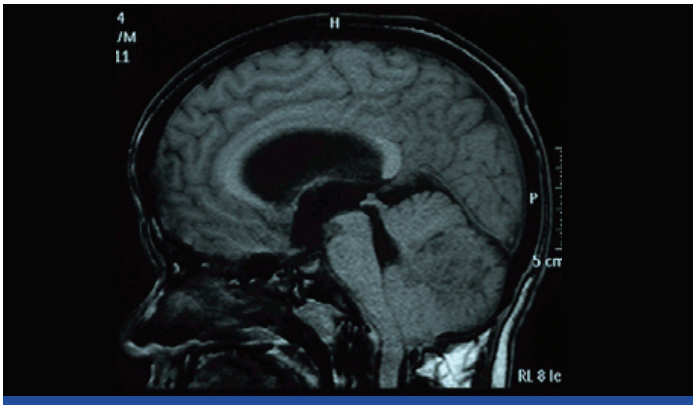
A 30-year-old male patient presented with progressive occipital headache since one year with no associated nausea, vomiting or gait disturbance. Neurological examination revealed negative Rhomberg's sign. Computed tomography revealed non-enhancing hypodense mass in right cerebellar hemisphere compressing fourth ventricle with obstructive hydrocephalus, however it was inconclusive.

Afterwards, MRI was performed which revealed right cerebellar mass with striated hypo and iso intense signal on T1W images, hyper and iso intense signal on T2W images. Mass was compressing fourth ventricle postero-inferiorly with obstructive hydrocephalus. Associated inferior tonsillar herniation into upper cervical canal was also noted [Table/Fig-1,2].

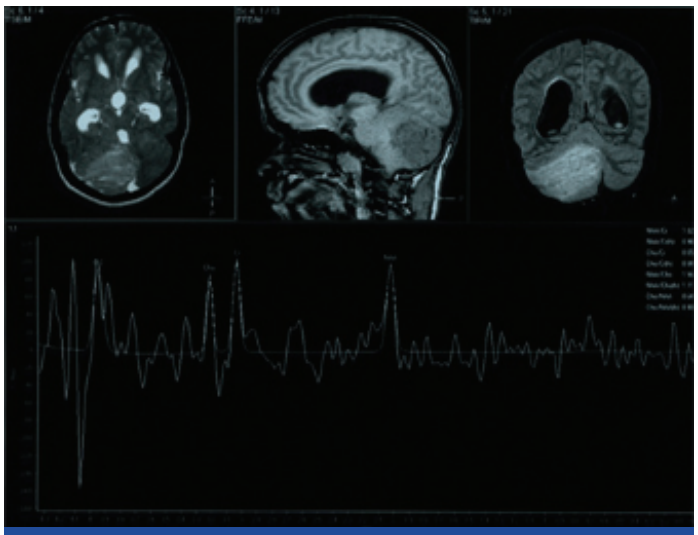
Proton MRS showed reduced NAA, Choline peaks. Cho/Cr and NAA/Cr ratios were within normal limits [Table/Fig-3]. On Flurodeoxyglucose (FDG) Positron emission tomography (PET) and PET-CT examinations the mass lesion showed evidence of hypermetabolism [Table/Fig-4,5].



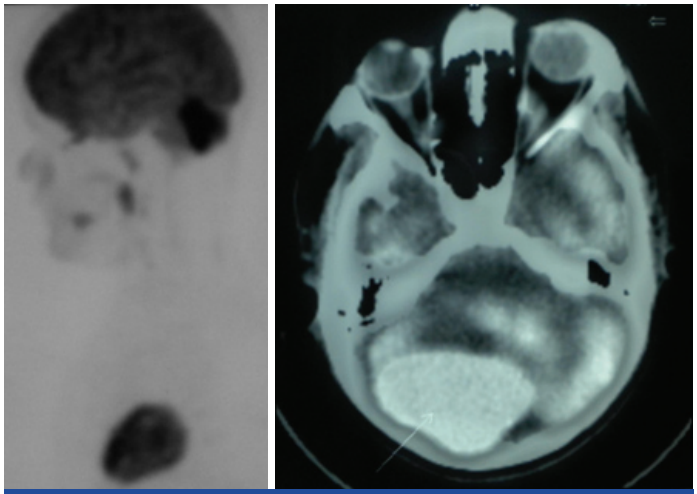
[Table/Fig-1]: T2W axial (A) & FLAIR (B) coronal images reveals right cerebellar mass (arrows) with alternate bands of hyperintense and isointense signal giving "tiger stripes" appearance with mass effect over cisterna magna & 4th ventricle



[Table/Fig-2]: T1W sagittal image reveals alternating bands of relative isointense and hypointense signal within the lesion (arrow) with mild inferior tonsillar herniation



[Table/Fig-3]: MRS image reveals reduced peaks of NAA and choline, decreased Cho/Cr ratio (0.65). NAA/Cr (1.02) & NAA/Cho (0.94) ratios were also reduced compared to normal



[Table/Fig-4]: PET reveals increased FDG uptake in the lesion suggesting hypermetabolism (arrow) **[Table/Fig-5]:** PET, CT reveals increased FDG uptake by the right cerebellar mass (arrow)

Considering diagnosis of Lhermitte-Duclos disease, Ultrasound and CT-scan of chest and abdomen were done, but they did not reveal any abnormality hence ruling out features suggesting Cowden Syndrome.

Patient was operated and avascular mass was resected with free margins and sent for histopathological examination. HPE revealed WHO grade-I dysplastic cerebellar gangliocytoma according to criteria for the diagnosis of Lhermitte-Duclos disease. Postoperative course was uneventful and patient was discharged within 1 week after surgery without any neurological deficit. First described

in 1920 [1] by Lhermitte and Duclos, Lhermitte Duclos disease (LDD) (dysplastic gangliocytoma) is a rare disorder of uncertain pathogenesis characterized by disarrangement of normal cerebellar laminar cytoarchitexture. This entity has been described by variety of terms like granular cell hypertrophy, ganglioneuroma and gangliocytoma dysplasticum [2]. LDD is found to be associated with Cowden syndrome in 40% cases. Cowden syndrome is also known as multiple hamartoma syndromes, a rare autosomal dominant condition characterized by multiple hamartomas and neoplasms of ectodermal, mesodermal and endodermal origin [3]. LDD can also be accompanied by megalencephaly, hydromelia, heterotopia, microgyria, polydactyly, leontiasis ossea and multiple haemangiomas [2]. Our patient did not show any features of this syndrome.

Although LDD usually occurs in third or fourth decades, reported age ranges from newborn to 74 years [3]. Clinically, LDD typically presents with findings of increased intracranial pressure in young adults. Many patients suffer from headache due to increased intracranial pressure caused by mass effect and obstructive hydrocephalus. Nausea, vomiting, visual disturbance and cerebello-vestibular symptoms can also be seen [3].

Classical MRI findings of LDD is described as especially unilateral, non enhancing hemispheric mass with layered appearance consisting of alternating bands of relative iso-, and hypointensity to normal brain on T1W, and relative hyper and iso intensity on T2W images [3]. Wolansky et al., mentioned that “tiger stripes” appearance is so distinctive that that it is unlikely to be confused with another entity. Non-enhancement is not a rule, since LDD lesions with contrast enhancement have been reported rarely [4]. Our case showed right cerebellar lesion with hemispheric expansion and typical striated pattern resembling tiger stripes on both T1- and T2W images with no contrast enhancement or perilesional oedema.

MRS shows some characteristic findings such as decreased NAA with increased lactate peak. Lack of increased Cho/Cr ratio is striking which are in contrast to cerebral tumors where choline levels are raised. Our case showed decrease level of NAA and choline. Cho/Cr ratio was also decreased (0.65). NAA/Cr (1.02) & NAA/Cho (0.94) ratios were also reduced compared to normal.

The disease is difficult to diagnose on CT, which shows poorly defined hypodense lesion with no contrast enhancement but may show calcification and thinning of occipital bone. FDG-PET revealed increased 18-FDG uptake within whole right cerebellar lesion suggesting either increased overall cell metabolism or an isolated up regulation in enzyme activity of hexokinase remains unclear. Histological examination reveals typical findings of thickening and hypermyelination of the molecular layer and large pleomorphic cells replacing purkinje and granular cell layers. Demyelination of central white matter of the folia is also observed.

Conventional MRI shows characteristic and pathognomic features of LDD, however MRS can provide useful additional information regarding benign nature of this lesion. Hypermetabolism on FDG-PET further supports diagnostic accuracy. Thus imaging can achieve accurate preoperative diagnosis of LDD along with delineation of the margins, thereby enabling complete excision.

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PARTICULARS OF CONTRIBUTORS:
1. Senior Resident, Department of Radio Diagnosis, G B Pant institute of Post Graduate Medical Education and Research, New Delhi, India.
2. Senior Resident, Department of Radio Diagnosis, G B Pant institute of Post Graduate Medical Education and Research, New Delhi, India.
3. Assistant Professor, Department of Radio Diagnosis, GG B Pant institute of Post Graduate Medical Education and Research, New Delhi, India.
4. Head and Director Professor, Department of Radio Diagnosis, G B Pant institute of Post Graduate Medical Education and Research, New Delhi, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Neeraj Jain,
Room no – 305, New Resident Doctors Hostel, G B Pant Hospital, New Delhi-110002, India.
E-mail: jain_neeraj2@yahoo.co.in

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