Lhermitte – Duclos disease: A case report

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ABSTRACT

Introduction: Lhermitte – Duclos disease (LDD) is a rare disorder characterized by a slowly enlarging mass lesion in the cerebellum. **Case report:** A case of histopathologically confirmed Lhermitte-Duclos disease is reported and our findings are discussed. Lhermitte-Duclos disease (Dysplastic gangliocytoma of the cerebellum) is a rare space occupying lesion of the posterior fossa with some typical neuroradiological features which can be better diagnosed in the magnetic resonance imaging (MRI) era.

Key Wards: Cerebellum, Dysplastic gangliocytoma, Lhermitte – Duclos disease

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INTRODUCTION

Dysplastic gangliocytoma of the cerebellum was first described by Lhermitte and Duclos in 1920 and is a rare hamartomatous lesion of the posterior fossa.[1] Dysplastic gangliocytoma is a rare disorder, characterized by a slowly progressive unilateral tumour mass of the cerebellar cortex. The male:female ratio is 1:1 and the average age is about 34 years. MRI is the diagnostic method of choice and also important for follow-up examinations. [2]

CASE REPORT

A twenty-five year old male patient presented with complains of headache, vomiting and giddiness for one and half year. Frequency of complaints was increased since last six months. MRI Brain revealed an enlarged left cerebellum with alternate bands of hypo and hyper intensities (tiger-striped appearance) on T2-weighted images. The lesion was characterized by regional enlargement and thickening of the cerebellar folia. Diffuse minimal enhancing lesion with mild mass effect

over brain stem and fourth ventricle was noted. Mild cerebellar tonsillar herniation, compression over lateral aspect of medulla, pons and mild midline shift on right side was present. Compression over inferior forth ventricle and mild obstructive dilatation of ventricular system was noted [figure – 1(a-d)]. Possibility of dysplastic cerebellar gangliocytoma was given.

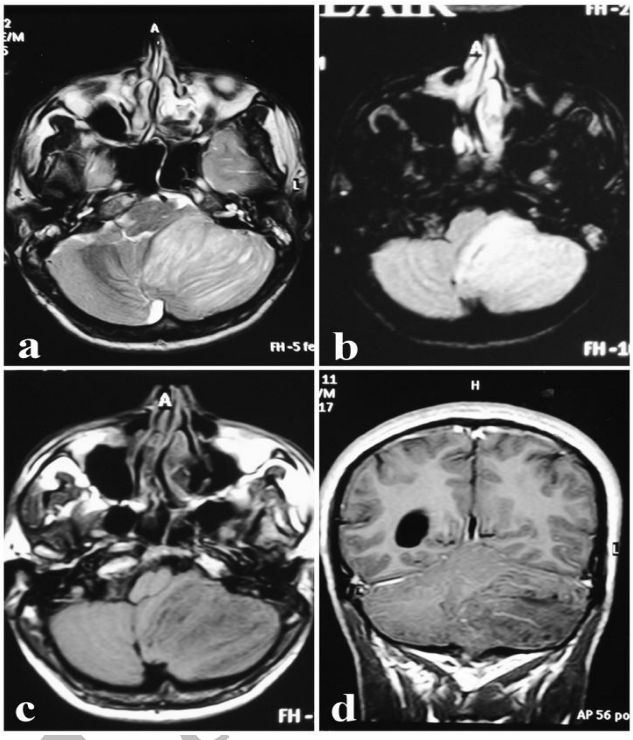

Figure 1 MRI Brain. a & b- hyperintense lesion on T2 – weighted and FLAIR images (tiger – striped appearance), c & d- hypointense lesion on T1 – weighted images.

Biopsy was performed and sent for histopathological examination. Histologically the section showed distortion of the normal cerebellar architecture. The principal abnormality was massive replacement and expansion of the internal granular layer (IGL) by large pleomorphic cells with eosinophilic cytoplasm, vesicular nuclei and prominent nucleoli, suggesting neuronal differentiation. Dysplastic ganglion cells were evident. The cells were haphazardly oriented with regard to one another, lacking the polarity and apical dendritic specialization of purkinje cells. Artefactual vacuolization was seen that may be responsible for

prominent stripes in MRI. No mitoses were found [figure – 2(a-f)].

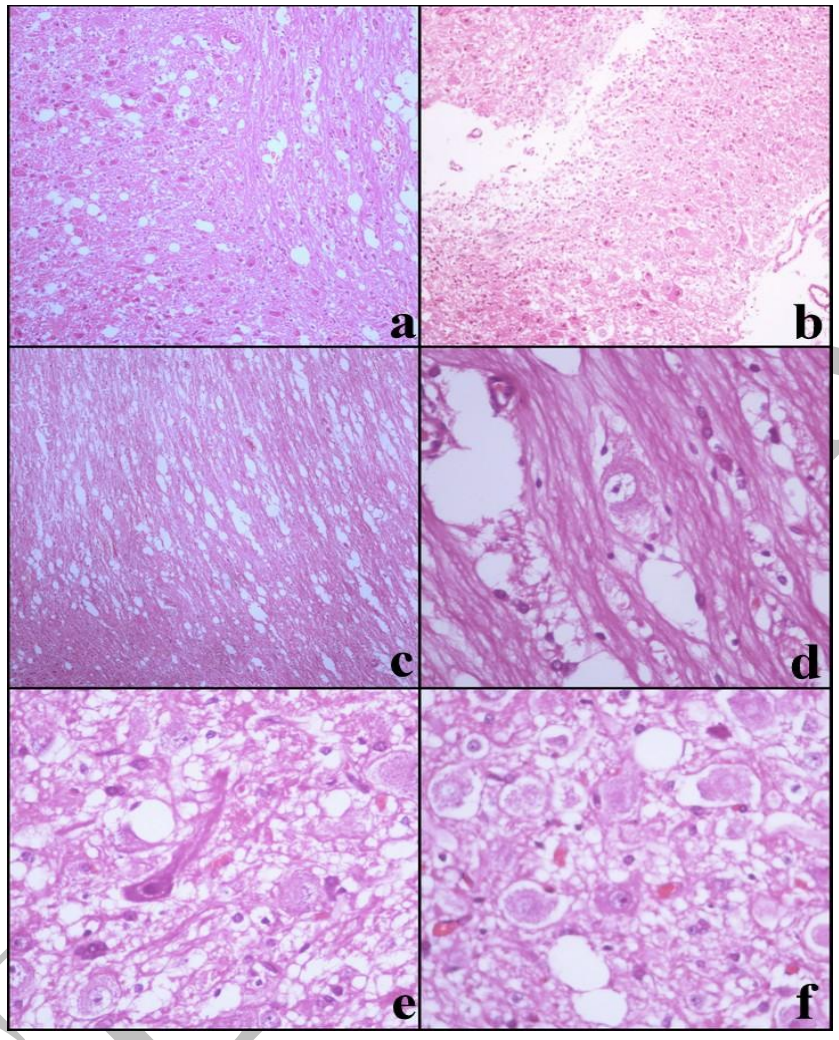

Figure 2 Lhermitte – Duclos disease. a- distortion of normal cerebellar architecture(H & E,10x), b- expansion of IGL (H & E,10x), c & d- artefactual vacuolization (H & E,10x & 40x), e & f- dysplastic ganglionic cells (H & E,40x).

Final diagnosis was given as Dysplastic cerebellar gangliocytoma WHO grade I (Lhermitte-Duclos disease)

Post-operative course was uneventful. Follow up reveals that the patient is well without any neurological complains.

DISCUSSION

Lhermitte-Duclos disease is a rare hamartomatous lesion involving the cerebellum causing progressive mass effect in the posterior fossa. [3]

It can occur sporadically, but also in a familial form. Ambler et al. described the first familial association of the tumor in a mother and son. [4] The male:female ratio is 1:1. The age ranges from newborn to the sixth decade. Most cases of Lhermite-Duclos disease occur in adults, usually in the third and fourth decades. Our patient was 25 years old. Clinical symptoms and signs are usually headache, nausea, vomiting, gait ataxia and papilloedema. In the present case the patient reported no personal or family history. The lesion was found to be hypointense on T1- and hyperintense on T2-weighted and FLAIR magnetic resonance images. [5]

Published papers have reported that Lhermitte-Duclos disease is a hamartomatous lesion rather than a neoplasm. This is supported by genetic findings, which suggest that these lesions are a developmental abnormality rather than a neoplasm. [2] A high PTEN-mutation frequency in Lhermitte-Duclos disease has been reported in several recent studies. [3]

Treatment is usually surgical removal of the lesion and the surgical procedure should be as radical as possible especially in young patients.[6] But the problem of surgical removal of these tumors is missing the borderline between tumor and healthy cerebellum tissue so that incomplete removal of the tumor is not rare.

The prognosis is favorable and the mortality is low. The efficacy of radiation therapy is unknown and not indicated in the initial treatment. [2]

REFERENCES

1. Lhermitte J, Duclos P (1920) Sur un ganglioneuroma diffus du cortex du cervelet. Bull Assoc Franc Cancer 9: 99–107
2. Buhl R, Barth H, Hugo HH, Straube T, Mehdorn HM (2003) Dysplastic gangliocytoma of the cerebellum: rare differential diagnosis in space occupying lesions of the posterior fossa. Acta Neurochir 145(6):509–512
3. Chiofalo MG, Cappabianca P, Basso De Caro MLD, Pezzullo L. Lhermitte – Duclos Disease. J Neurooncol (2007); 82: 183-185
4. Ambler M, Pogacar S, Sidman R (1969) Lhermitte-Duclos disease (granule cell hypertrophy of the cerebellum). Pathological

analysis of the first familial cases. J Neuropathol Exp Neurol 28: 622–647

5. Nowak DA, Trost HA, Porr A, Stolzle A, Lumenta CB (2001) Lhermitte-Duclos disease (dysplastic gangliocytoma of the cerebellum). Clin Neurol Neurosurg 103: 105–110
6. Milbouw G, Born JD, Martin D, Collignon J, Hans P, Reznik M, Bonnal J (1988) Clinical and radiological aspects of dysplastic gangliocytoma (Lhermitte-Duclos disease): A report of two cases with review of the literature. Neurosurgery 22: 124–128