

Case Report

Lhermitte Duclos disease: a rare cerebellar lesion with characteristic neuroimaging features

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ABSTRACT

Lhermitte-Duclos disease (dysplastic cerebellar gangliocytoma) is a rare disorder of uncertain pathogenesis characterized by disarrangement of the normal cerebellar laminar cytoarchitecture. A case of histopathologically confirmed Lhermitte-Duclos disease with characteristic imaging findings and successful surgical management is reported and our findings are discussed.

Keywords: Lhermitte-Duclos disease/ dysplastic cerebellar gangliocytoma, Hamartomatous, Cowden syndrome

INTRODUCTION

Lhermitte-Duclos disease (dysplastic cerebellar gangliocytoma) is a rare disorder of uncertain pathogenesis characterized by disarrangement of the normal cerebellar laminar cytoarchitecture.^{1,2} There is considerable debate regarding it being a developmental disorder, hamartoma, or a low-grade neoplasm and hence has been described by a variety of terms, including granule cell hypertrophy, ganglioneuroma, purkinjeoma, hamartoma gangliomatosis, neurocystic blastoma, hamartomoblastoma and granulomolecular hypertrophy of the cerebellum.³⁻⁵

A case of histopathologically confirmed Lhermitte-Duclos disease with characteristic imaging findings and successful surgical management is reported and our findings are discussed.

CASE REPORT

A 26 year old male presented with history of headache, convulsions, altered sensorium and vomiting of one

month duration. Examination of sensory and motor nervous system was normal and there was no history of trauma or other preceding illness.

A contrast enhanced CT scan of the patient was recommended which revealed hydrocephalus secondary to a non-enhancing hypo attenuating right cerebellar mass lesion compressing the fourth ventricle. However no definite imaging diagnosis could be provided. CSF analysis of the patient did not reveal any abnormality and thereafter a ventriculoperitoneal shunt was placed to relieve the hydrocephalus.

Next an MR imaging was done for further characterization of the lesion, which revealed a large non-enhancing area of abnormal signal intensity involving the right cerebellar hemisphere and brachium pontis with prominent widened cerebellar folia having a characteristic striated pattern typical of Lhermitte-Duclos disease/ dysplastic cerebellar gangliocytoma with associated moderate obstructive hydrocephalus (Figure 1 and 2). A small cavernous angioma and a benign developmental venous anomaly were also noted in our patient.

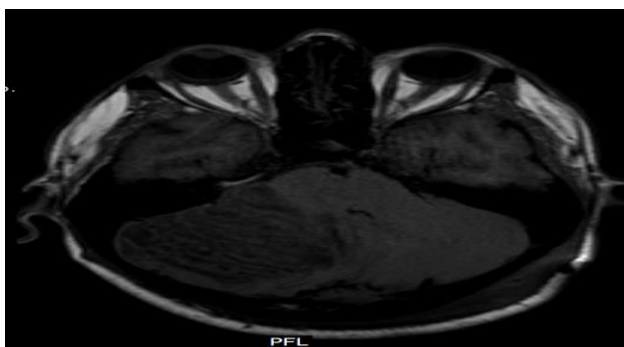


Figure 1: T2W sequences showing prominent widened cerebellar folia with striated pattern of alternating isointense and hyperintense signals.

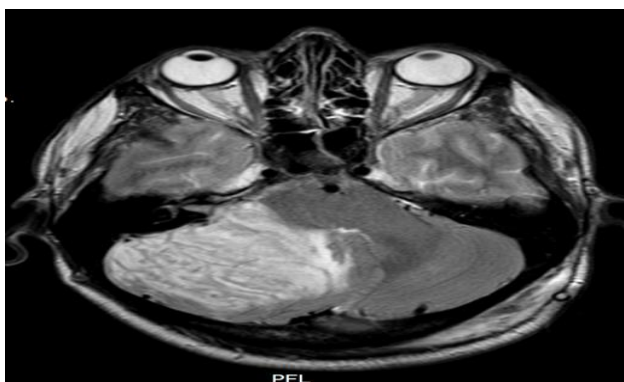


Figure 2: T1W sequences showing prominent widened cerebellar folia with striated pattern of alternating isointense and hypointense signals.

Surgery was then performed with a right paramedian sub occipital craniectomy and debulking of the tumor was done though the entire tumor was not resected due to its close proximity with the brain stem.

Specimen was sent for histopathological evaluation which revealed variable replacement of granule cell layer by a disorganized array of large neurons with attendant expansion and aberrant myelination of the overlying molecular layer which showed coarse spongy change and thinning of folial white matter. Neuronal elements were well differentiated and showed Purkinje cell like forms, with some neurons showing atypical appearance. The axons emanating from these cells were oriented in parallel stacks within the deep molecular zone and in perpendicular stacks more superficially. These features were highly suggestive of dysplastic gangliocytoma of the cerebellum, thereby confirming our imaging diagnosis.

The patient has been under close follow up and is doing well with resolution of his neurological symptoms on his review visit one month after the operative intervention.

DISCUSSION

First described in 1920, by Lhermitte and Duclos as diffuse ganglioneuroma⁶, Lhermitte Dulcos disease is a lesion of variable neoplastic potential, and a characteristic typical striated folial pattern on MR images.

Exact pathogenesis of this disease remains a matter of debate with speculations of this disease having a hamartomatous, neoplastic, or congenital malformative origin.

Association of LDD, especially the adult onset variety with mutations in PTEN (phosphatase and tensin homologue), a tumor suppressor gene located on chromosome ten, has led to the inclusion of this disease in the PTEN hamartoma tumor syndromes (PHTS) and hence favour its hamartomatous origin. It is also considered as a phenotypic variant of Cowden's syndrome (CS), and in 2004, was recognized as a pathognomonic criterion for CS by international cowden consortium criteria.⁷

Cowden syndrome is a prototypic of PHTS and is characterized by a variety of mucocutaneous lesions, macrocephaly, and increased frequency of hamartomas and neoplasia in the breast, thyroid, colon, genitourinary organs, and central nervous system. And ideally a genetic testing for PTEN mutation should have been carried out in our patient, however the financial constraints of the patient limited further evaluation and we resorted to psychosocial assessment and risk counseling of the patient and have kept the patient in close follow up with a plan for annual screening for thyroid, breast and dermatological manifestations along with annual urinalysis.⁷

Macroscopically, dysplastic gangliocytoma consists of a mixture of normal and enlarged cerebellar folia⁸. Histologic examination reveals thickening and hypermyelination of the molecular layer and large pleomorphic cells that replace the Purkinje and granular cell layers.^{2-5,8-10} (Figure 5,6). Demyelination of the central white matter of the folia is also observed.⁸⁻¹⁰ The transition between normal and abnormal cerebellar tissue may be gradual.⁴ Mitoses, necrosis, and neovascularity are not features of dysplastic gangliocytoma.^{1,9,10}

Neuro imaging plays a vital role in the diagnostic process with characteristic appearance of the disease especially on MR imaging. On computed tomographic (CT) images, dysplastic gangliocytoma presents as a hypo to isoattenuating, non-enhancing posterior fossa mass compressing the fourth ventricle. MR imaging is the modality of choice, and reveals a cerebellar mass with a typical striated or tiger-stripped folial pattern that consists of alternating bands on both T1 and T2 weighted images. The bands are hyper- and isointense relative to gray matter on T2 weighted images (Figure 1,2) and iso to hypointense on T1 weighted images¹¹ (Figure 3,4).

Calcification is an uncommon finding, but it has been reported.¹² Most dysplastic ganglio cytomas do not enhance; however, enhancement has been reported and is probably due to the presence of anomalous veins.^{13,14} No diffusion restriction is noted in these lesions. Mass effect is common with compression of the fourth ventricle and occlusive hydrocephalus. MR spectroscopy though not performed in our patient reveals reduced N-acetylaspartate-choline and N-acetylaspartate-creatine ratios compared with those of normal cerebellar tissue.¹⁵ Lactate peak may also be present. MR imaging is highly reliable in diagnosing this condition, obviating biopsy in asymptomatic patients and is also invaluable for preoperative planning and to determine the extent of resection.

Imaging differential diagnosis (esp. on CT) can include cerebellar infarction, cerebellar encephalitis, true dysplasia of the cerebellum, dysplastic cerebellar lesions of tuberous sclerosis, cerebellar astrocytoma, medulloblastoma and cerebellar metastases. However, progressive nature of symptoms in a young male with non-enhancing solid posterior fossa mass and typical tiger striped folial pattern on MR imaging, helps us rule out these possibilities and support a confident pre-operative diagnosis of Lhermitte Dulcos disease in our patient. The case report thus highlights the role of neuroimaging in a conclusive pre-operative diagnosis of this probably hamartomous cerebellar lesion and hence helps in adequately planning the operative approach.

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