Dysplastic Cerebellar Gangliocytoma (Lhermitte-Duclos Disease)

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Introduction

Dysplastic gangliocytoma (Lhermitte-Duclos disease [LDD]) is a hamartomatous lesion of the cerebellum classified as WHO grade 1. While sporadic cases are reported, this entity is typically a component of Cowden (multiple hamartoma neoplasia) syndrome presenting in young adults with a germline PTEN mutation (PTEN hamartoma syndrome). We present a classic case of unilateral Lhermitte-Duclos disease associated with Cowden syndrome.

Case

A 63-year-old male with a known history of papillary renal cell carcinoma and prostatic adenocarcinoma presented with headaches for one year:

-Genetic testing was significant for a **c.49C>T mutation of PTEN.**

-MRI Brain showed a **T2 hyperintense, dysplastic striated (tigroid) appearance of the left cerebellum** at the anteromedial and posterolateral aspects, including the cerebellar vermis and upper left cerebellar tonsil.

-Frozen and permanent H&E-stained sections of the cerebellum showed lesional tissue with increased numbers of **atypical/dysplastic ganglion and ganglioid cells with no high-grade features.**

-The lesional cells involve the upper parts of the internal granular layer and spread to the molecular layer, **expanding the folia.** The lesional cells are accompanied by **abnormal vacuolation** (clear spaces and clefts) of the white matter and neuropil.

Images

Figure 1. Coronal (A) and Axial (B) Brain MRI with the characteristic tigroid appearance of the lesion.

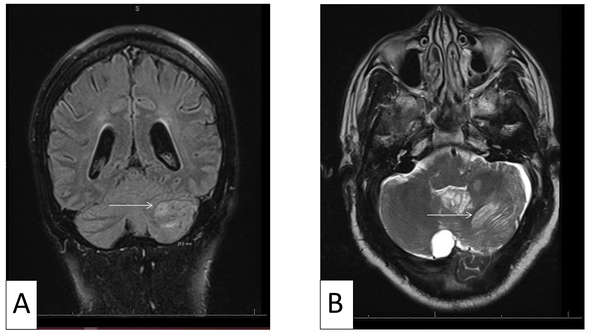

Figure 2. By immunohistochemical staining, PTEN is absent in tumor cell nuclei.

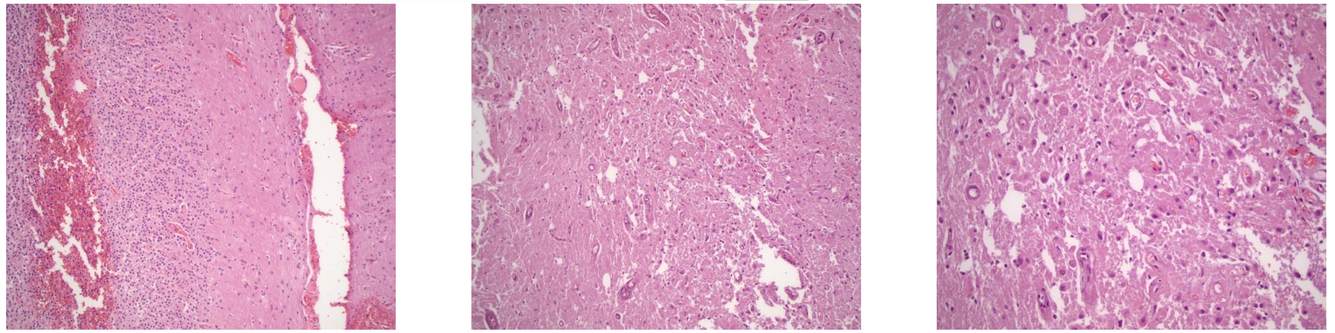

Figure 3. By H&E staining, the normal cerebellum (A, 100x) is shown in contrast to the disorganized layers of the dysplastic gangliocytoma (B, 200x, C, 400x).

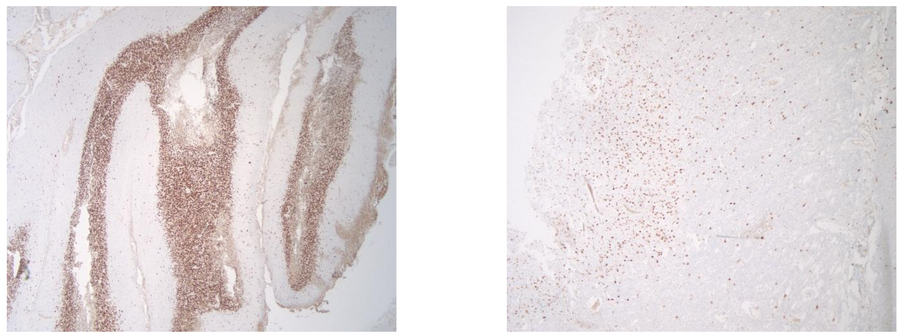

Figure 4. By immunohistochemical staining, NeuN highlights the neurons in normal cerebellum (A, 40x) in contrast to the dysplastic gangliocytoma (B, 100x).

Discussion

-Approximately 40% of patients with Cowden syndrome develop Lhermitte-Duclos disease.

- Lhermitte-Duclos Disease is a rare hamartomatous lesion of the cerebellum with replacement of the normal internal granular layer by small to large ganglion cells.

-On T2 MR, the focal, striped (tigroid) appearance of a unilateral, cerebellar lesion is highly suggestive of LDD.

-Foliar expansion of LDD is best appreciated at low magnification.

-At high magnification, the laminar nature of LDD may not be apparent and the abnormal neurons may be misinterpreted as a gangliocytoma.

-LFB staining allows for identification of myelin within the normally myelin-free molecular layer.

-Clear spaces and clefts are distinctive in white matter, a potential pitfall is confusion with intratumoral microcysts which are common in ganglion cell tumors.

-Prognosis is good with no malignant potential; however, approximately 25% recur and require additional resection if symptomatic.

References

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