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ISSN 0722-5091

DOI 10.5414/NP301276
e-pub: February 12, 2020

Teaching case 2-2020: Lhermitte-Duclos disease in a female patient with Cowden syndrome

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Key words

Cowden syndrome –
Lhermitte-Duclos
disease – PTEN

We present a rare case of adult Lhermitte-Duclos disease (aLDD) in the setting of Cowden syndrome in a 37-year old female patient. Clinical history together with neuroimaging and neuropathological findings are presented.

The patient was admitted to Neurosurgery because of blurred vision and severe headaches associated with nausea and vomiting, persisting for 2 weeks. She also reported tremor and paresthesias in the left upper extremity. A CT scan revealed a $6 \times 5 \times 3$ cm space-occupying posterior fossa lesion on the left side with distortion of the 4th ventricle and signs of ipsilateral tonsillar herniation. Ophthalmologic examination revealed a marked bilateral papillary edema. A subsequently performed MRI scan confirmed a demarcated, striated lesion with alternating T1-isointense and hypointense bands (Figure 1A, B). Contrast enhancement was completely absent. The aqueduct was not occluded; however, the temporal horns of the lateral ventricles were slightly enlarged. The lesion was surgically resected.

Neuropathological evaluation revealed small portions of adjacent normally configured cerebellar tissue and a marginally pleomorphic and slightly hypercellular tumor. The tumor replaced the cerebellar cortex in a band-like fashion and contained multiple gangliocytes with distinct nucleoli (Figure 1C, H & E staining). The underlying white matter was characterized by loosening of the tissue and very occasional scattered dysplastic gangliocytes. No mitotic figures were encountered in the tumor, and immunohistochemical staining for MIB1 marked

only few and small nuclei (not shown). The band-like tumor was strongly positive for synaptophysin (Figure 1D) and contained multiple ectatic blood vessels, which could be clearly depicted by CD34 immunohistochemistry (Figure 1E). CD34 staining additionally revealed upregulated expression in glial cells within the tumour in a focal and bushy-like fashion, while the gangliocytes remained negative. Finally, immunohistochemistry for PTEN, which was absent in the enlarged neurons but preserved in the blood vessels (Figure 1F), corroborated the neuropathological diagnosis of a dysplastic cerebellar gangliocytoma and thus aLDD.

aLDD is diagnostic for Cowden syndrome [1]. Reassessment of the patient's clinical history revealed a follicular thyroid carcinoma and a ductal breast carcinoma in situ. Moreover, review of the relevant literature confirmed the MRI study as highly typical for aLDD [2].

This case underscores the importance of clinical, imaging, and neuropathological synopsis in order to adequately diagnose and counsel the patient as well as monitor carcinoma recurrences or other manifestations in this patient, such as endometrial carcinoma.

Funding

No funding received for this study.

Conflict of interest

The authors declare no conflicts of interest.

Received
February 3, 2020;
accepted
February 3, 2020

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Figure 1. Post-contrast axial (A) and coronal (B) T1-weighted image of the space-occupying lesion. C: H & E stained section of the band-like superficial hamartomatous lesion with multiple gangliocytes showing distinct nucleoli (inset); D: Synaptophysin IHC shows strong reactivity of the tumorous neuropil matrix; E: CD34 IHC reveals reactive glial upregulation in a bushy-like fashion. In addition, multiple ectatic blood vessels are highlighted; F: PTEN IHC with preserved endothelial staining and negativity of gangliocytes.

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