Lhermitte-Duclos disease

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A 26-year-old male presented with history of headache of six months duration. The review of history was unremarkable. General examination and neurological examination was within normal limits. Magnetic resonance imaging (MRI) was taken which revealed a T2 hyperintensity in the right cerebellar hemisphere with preserved cerebellar architecture and alternating hypo and hyperintense stripes (tiger-striped) [Figure 1]. Contrast study was done which showed no significant enhancement [Figure 3]. The lesion was producing mild brainstem compression. This MRI picture with Tiger-striped appearance is classical of Lhermitte-Duclos disease (LDD). The lesions which were not obeying vascular boundaries with preserved normal cerebellar architecture ruled out infarcts and other tumors as the cause. Patient refused for a surgery so the plan was to follow up the patient.

Lhermitte-Duclos disease was first described in 1920 in the literature under the names of Purkinjeoma, granular cell hypertrophy of the cerebellum, hamartoma of the cerebellum, dysplastic gangliocytoma, ganglioneuroma, and gangliomatosis of the cerebellum. Patients tend to be young adults and may present with signs of cerebellar dysfunction or increased intracranial pressure secondary to obstructive hydrocephalus. The cerebellar signs are minimal or absent in up to one half of those with the lesion. LDD presents on the MRI as a nonenhancing unilateral lesion in the cerebellum with mass effect on the surrounding structures. The lesion is hypointense on the T1 weighted images and hyperintense on the T2-weighted images with alternating parallel hyperintense and isointense stripes which are characteristic of the disease [Figure 2]. These bands correspond to the inner molecular and granular layer of the cerebellum. Loss of central white matter within the folia also contributes to
Tonsillar herniation and hydrocephalus are quite common and are caused by the mass effect of the lesion to the adjacent cerebellar parenchyma. T1-images are usually unremarkable, because no contrast enhancement is detected as there is no significant disturbance in the blood-brain barrier.[1]

Diffusion properties in LDD can be very variable depending on the contributions from the inner layer and the thick outer molecular layer with large dysplastic neurons.[2,3] MRS in LDD showed decrease in the N-Acetyl Aspartate (NAA)/Creatine (Cr) and NAA/Choline (Cho) ratios with near normal values of Cho/Cr, as well as an obvious lactate peak and lack of lipids suggestive of a benign hamartoma.[2,3] SPECT, FDG-PET and dynamic susceptibility-weighted MR perfusion images has been shown to demonstrate increased perfusion in LDD. 11C-methionine positron emission tomography visualizes the lesion of LDD as a high uptake area.[4]

The typical clinical course of LDD consists of insidious expansion of a posterior fossa mass. Decompressive surgery for symptomatic patients has been successful both in relieving symptoms and in improving longterm survival from 2.5 to 11 years.[5] The outcome in patients who were not operated on was uniformly poor. During surgical exploration, no tumor mass can be found. This macroscopic appearance is confirmed by histopathological findings demonstrating a transitional area between normal and pathological cerebellar tissue. Thus, the absence of tumor limits in the depth of the cerebellar hemisphere constitutes the major technical problem during surgery.[5]

Macroscopically there is widening of the cerebellar folia which efface the sulci. The histopathological findings consists of widening of the molecular layer with abnormal myelination that is occupied by abnormal ganglion cells, absence of the Purkinje cell layer and hypertrophy of the granular cell layer, with atrophy of the cerebellar white matter.[1] LDD is associated with Cowden’s syndrome (CS), a rare autosomal dominant familial cancer syndrome with multiple manifestations including trichilemmomas, diverse hamartomas, intestinal polyposis, palmoplantar keratoses, oral papillomatosis, and an increased predisposition to breast cancer and thyroid tumors.[1] Both conditions are linked to mutations in the PTEN gene, which controls cell apoptosis, migration and differentiation.

References

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