Lhermitte-Duclos disease

Lhermitte-Duclos disease (LDD), also known as dysplastic cerebellar gangliocytoma, is a rare tumour of cerebellum.

Epidemiology

Lhermitte-Duclos disease typically presents in young adults.

A number of associated conditions have been described, including:

Cowden disease: then termed Lhermitte-Duclos-Cowden syndrome or COLD syndrome.

disorders of cortical formation

megalencephaly

grey matter heterotopia

polymicrogyria

polydactyly

hydromyelia

macroglossia

localised gigantism

leontiasis ossea

Lhermitte-Duclos disease associated with spinal cervical AVF [1].

Pathogenesis

It is probably hamartomatous, although the exact pathogenesis remains unknown.

It is considered a WHO grade I tumour.

Clinical presentation

Symptoms are typically related to raised intracranial pressure, obstructive hydrocephalus and to a lesser degree cerebellar dysfunction.

Pathology

Results from derangement of normal laminar cellular organization of cerebellum. There is thickening of outer molecular cell layer, loss of middle Purkinje cell layer, and infiltration of inner granular cell layer with dysplastic ganglion cells.

Markers

Stains positive for synaptophysin.

Radiographic features

The abnormal tissue involves the cerebellar cortex, and is usually confined to one hemisphere, occasionally extending to the vermis but only rarely extending to the contralateral hemisphere.

CT

may show a non specific hypo attenuating cerebellar mass calcification is sometimes seen

MRI

Widened cerebellar folia with a striated/ tigroid appearance.

T1: hypointense T2: hyperintense with apparently preserved cortical striations DWI: similar to normal cortex may show hyperintensity due to T2 shine through effect T1 C+ enhancement is rare if present usually superficial, possibly due to vascular proliferation 4 MR spectroscopy elevated lactate 1,2 slightly reduced NAA (by about 10%) reduced myo-inositol (by 30-80%) reduced choline (by 20-50%) reduced cho/cr ratio PET/SPECT

FDG-PET: shows increased uptake Tl-201 SPECT: shows increased uptake Treatment and prognosis

The dysplastic mass grows very slowly, and initial treatment revolves around treating hydrocephalus. Surgical resection is often curative, with only a few case reports of recurrence.

Importantly it is crucial to remember association with Cowden syndrome, hence, increase in risk of other neoplasms such breast, endometrial and thyroid cancers. So, recommendation for further imaging or clinical assessment of possible tumours in these locations should be included in the radiologists report.

Case reports

A report of an asymptomatic cervical dural arteriovenous fistula in a patient with Lhermitte-Duclos disease was published in 2006. Almubarak et al. from Riyadh, presented in 2018 the second case of Lhermitte-Duclos disease associated with an asymptomatic spinal cervical AVF in a 17-year-old young woman with literature review of central nervous system vascular lesions in Lhermitte-Duclos disease [2].

[1], [2]
Almubarak AO, Haq AU, Alzahrani I, Shail EA. Lhermitte-Duclos Disease with Cervical Arteriovenous Fistula. J Neurol Surg A Cent Eur Neurosurg. 2018 Dec 5. doi: 10.1055/s-0038-1670636. [Epub ahead

of print] PubMed PMID: 30517962.