

Lhermitte-Duclos disease

Identity

Other names Dysplastic gangliocytoma of the cerebellum

Inheritance Sporadic, or autosomal dominant if associated with [Cowden disease](#)

Clinics

Phenotype and clinics Lhermitte-Duclos disease consists in the occurrence of a slowly enlarging mass within the cerebellar cortex corresponding histologically to a cerebellar hamartoma.

Clinical manifestations are commonly a long standing history of vague defined neurological symptoms related to raised intracranial pressure and cerebellar signs affecting usually a young adult.

Diagnosis is based on cerebral imaging methods, mainly NMR-imaging.

Therapy consists of decompression of the posterior fossa by total surgical removal of the tumour mass.

Histopathological finding confirm the diagnosis of dysplastic gangliocytoma of the cerebellum in front of a hamartoma lesion with widening of the molecular layer occupied by abnormal ganglion cells, absence of Purkinje cell layer and hypertrophy of granular layer.

Related syndromes: Association with other lesions such as macrocephaly, polydactyly, multiple hemangioma, goiter, intestinal polyps was often observed. One case of familial cluster was reported. In fact, many cases of dysplastic cerebellar gangliocytoma are related to Cowden disease and such patients show various finding of this autosomal dominant condition with variable expression.

Neoplastic risk In cases of Lhermitte-Duclos related to a Cowden disease, malignant tumours characterizing this affection (mainly [breast carcinoma](#) and thyroid carcinoma) can occur.

Genes involved and Proteins

Gene Name [PTEN \(or MMAC1 or TEP1\) in cases related to Cowden disease.](#)

Location 10q23

Protein

Expression 403 amino-acids, phosphatase with tumor suppressive effects, negative regulator of the PI3K/Akt signal cell pathway by dephosphorylating PIP3

External links

[OMIM](#) [158350](#)

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