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Three unusual cases of benign pediatric CNS tumors including Lhermitte-Duclos disease

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Background: Lhermitte-Duclos Disease (LDD) is a rare disorder characterized by a slowly growing tumor in the cerebellum, the nature of which is uncertain. Three cases of tumours in the cerebellum were initially described pathologically as typical lesions for LDD. Due to the unusually frequent presentation of this disease, a pathological revision was conducted, concluding that just one case represented LDD. In addition to this, a literature search was performed to enlighten this rare disease, its relation to Cowden Disease and the discussion of its true classification.

Methods: Literature was searched for on PubMed. Medical records and pathologic evaluations of 3 patients from the Neurosurgical Department at Rigshospitalet were reviewed.

Results: Less than 300 cases of LDD have been reported, often accompanied by Cowden Disease. Looking at the histopathology, LDD presents as a benign lesion composed of dysplastic granule cells expanding by hypertrophy and thus compromising the normal architecture of the cerebellar cortex. The association with Cowden Disease suggests an ethology based on a loss-of-function mutation in the PTEN gene (10q23.3). PTEN normally acts in the PI3K-pathway, regulating cell size, migration, survival and proliferation. It seems, though, that in neuronal cells this mutation does not cause excessive proliferation and the tumour can therefore not be classified as a neoplasia.

Case reports: Three children (age 3 months, 2 and 14 years) all operated for a cerebellar tumour. Biopsies from all patients were reviewed in the pathological department. In the youngest patient, the histopathology was characteristic for an LDD lesion, but analysis showed no PTEN-mutation. The second case most likely represented a pilocytic astrocytoma and the third patient probably represented a classical gangliocytoma.

Conclusion: LDD should be classified as a malformation caused by aberrant migration and dysplasia of granule cells, perhaps caused by disturbances in the PI3K-pathway. In childhood there is no direct association with Cowden Disease.