

1. Papillary tumour of the pineal region

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Katherine Poulgrain, Ross Gurgo, Craig Winter, Benjamin Ong, Queenie Lau:
Papillary tumour of the pineal region: *Journal of Clinical Neuroscience* 18 (2011)
1007–1017.

Bujung Hona, Makoto Nakamura, Almuth Brandis, Hartmut Becker, Joachim K.
Krauss: Spinal metastasis of papillary tumor of the pineal region. *Clinical
Neurology and Neurosurgery* 113 (2011) 235–238

2. Primary melanocytoma, sclerotic, of leptomeninges, associated with NF 1

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CHU Ste-Justine, University of Montreal, Montreal, QC

Brat DJ, Giannini C, Scheithauer BW, Burger PV (1999). Primary melanocytic
neoplasms of the central nervous system. *Am J Surg Pathol* 23;(7): 745-754

Liubinas SV, Maartens N, Drummond KJ (2010). Primary melanocytic neoplasms
of the central nervous system. *J Clin Neurosc.* 17(10): 1227-1232, Epub 2010 Jun
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Primary melanocytic tumors of the central nervous system: a neuroradiological
and clinicopathological study of five cases and brief review of the literature. *J
Neurol India* 59(3):413-419

3. Lhermitte-Duclos disease

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Toronto, ON; ²Department of Laboratory Medicine, St. John Regional Hospital, St. John,
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Zhou XP, Marsh DJ, Morrison CD, Chaudhury AR, Maxwell M, Reifenger G,
Eng C. Germline inactivation of PTEN and dysregulation of the phosphoinositol-
3-kinase/Akt pathway cause human Lhermitte-Duclos disease in adults.
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Abel TW, Baker SJ, Fraser MM, Tihan T, Nelson JS, Yachnis AT, Bouffard JP,
Mena H, Burger PC, Eberhart CG. Lhermitte-Duclos disease: a report of 31 cases
with immunohistochemical analysis of the PTEN/AKT/mTOR pathway.

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Fraser MM, Bayazitov IT, Zakharenko SS, Baker SJ. Phosphatase and tensin homolog, deleted on chromosome 10 deficiency in brain causes defects in synaptic structure, transmission and plasticity, and myelination abnormalities. *Neuroscience.* 2008;15:476-88.

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J Neuropathol Exp Neurol. 1988;47:206-16.

4. **Lymphomatosis Cerebri**

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1. **Human Pathology** (2005) 36, 282– 290; Lymphomatosis cerebri as a cause of white matter dementia. Karen E. Rollins, B.K. Kleinschmidt-DeMasters, John R. Corboy, Denise M. Damek, Christopher M. Filley.
2. **Journal of the Neurological Sciences** 293 (2010) 122–124; Lymphomatosis Cerebri – A Rare Cause of Leukoencephalopathy. Lekha Pandit, Yasha Chickabasaviah, Ananthan Raghobaman, Sharik Mustafa, Arvind Vasudevan.
3. **Neuropathology** 2011;31,612-619; An Autopsy Case of Lymphomatosis Cerebri showing Pathological Changes of Intravascular Large B-cell Lymphoma in Visceral Organs. Nozomi Hishikawa, Hisayoshi Niwa, Takashi Hara et al.

5. **Glioblastoma, giant cell**

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Hall JR, Short SC. Management of glioblastoma multiforme in HIV patients: a case series and review of published studies. *Clin Oncol (R Coll Radiol).* 2009;21(8):591-7

Pore N, Gupta AK, Cerniglia GJ, Maity A 2009HIV protease inhibitors decrease VEGF/HIF-1alpha expression and angiogenesis in glioblastoma cells. *Neoplasia.* 2006;8(11):889-95.

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Blumenthal DT, Raizer JJ, Rosenblum MK, Bilsky MH, Hariharan S, Abrey LE. Primary intracranial neoplasms in patients with HIV. *Neurology*. 1999; 12;52(8):1648-51.

6. Polyglucosan body disease (glycogenosis type 4)

F. AlSufiani, M. Shkrum, D. Ramsay
London Health Sciences Centre, London, ON

Taratuto AL, Akman HO, Saccoliti M, Riudavets M, Arakaki N, Mesa L, et al. Branching enzyme deficiency/glycogenosis storage disease type IV presenting as a severe congenital hypotonia: muscle biopsy and autopsy findings, biochemical and molecular genetic studies. *Neuromuscul. Disord*. 2010 Dec.;20(12):783–90.

Nolte KW, Janecke AR, Vorgerd M, Weis J, Schröder JM. Congenital type IV glycogenosis: the spectrum of pleomorphic polyglucosan bodies in muscle, nerve, and spinal cord with two novel mutations in the GBE1 gene. *Acta Neuropathol*. 2008 Nov.;116(5):491–506.

Tay SKH, Akman HO, Chung WK, Pike MG, Muntoni F, Hays AP, et al. Fatal infantile neuromuscular presentation of glycogen storage disease type IV. *Neuromuscul. Disord*. 2004 Apr.;14(4):253–60.

7. Late Infantile Neuronal Ceroid Lipofuscinosis (NCL), with two disease causing mutations in CLN2

L. Hamilton and J. Joseph
University of Calgary, Calgary, Alberta

Jalanko A & T Braulke. Neuronal ceroid lipofuscinoses. *Biochimica et Biophysica Acta*. 2009;1793:697-709.

Kohlschütter A & A Schulz, Towards understanding the neuronal ceroid lipofuscinoses. *Brain & Development*. 2009;31:499-502.

Williams RE et al. Diagnosis of the neuronal ceroid lipofuscinoses: an update. *Biochimica et Biophysica Acta*. 2006;1762:865-872.

**8. Poorly differentiated sarcoma, NOS, with:
a) IHC suggestive of “desmoplastic small round cell tumor”
b) Break apart FISH evidence of a translocation event involving EWS but not WT1.**

C. Dunham, A. Singhal, J. Hukin, G. Hendson

Children's and Women's Health Centre of British Columbia

Luciano N. et al. (2009). Desmoplastic small round cell tumor of the central nervous system: report of 2 cases and review of the literature. *Virchows Arch*; 454: 431-439.

Bouchireb K. et al. (2008). Intracerebral small round cell tumor: an unusual case with EWS-WT1 translocation. *Pediatr Blood and Cancer*; 51: 545-548.

Tison V. et al. (1996). Intracranial desmoplastic small-cell tumor: report of a case. *Am J Surg Pathol*; 20(1): 112-117

9. Primary Rhabdomyosarcoma, Embryonal subtype with diffuse anaplasia

L. Hamilton and J. Chan

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Celli P et al. Primary rhabdomyosarcoma of the brain: observations on a case with clinical and radiological evidence of cure. *J Neurooncol*. 1998;36:259-267.

Taratuto AL et al. Primary rhabdomyosarcoma of the brain and cerebellum. Report of four cases in infants: an immunohistochemical study. *Acta Neuropathol (Berl)*. 1985;66:98-104.

Hawkins C et al. Case of the Month: April 1999 – 44 year old man with a bleeding intracerebral tumor. *Brain Pathology*. 1999;9:741-742.

10. Action myoclonus- renal failure syndrome

J. Ferreira

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Andermann E, Andermann F, Carpenter S, et al: Action myoclonus-renal failure syndrome: a previously unrecognized neurological disorder unmasked by advances in nephrology. *Adv Neurol* 1986, 43:87-103.

C Badhwar A, Berkovic SF, Dowling J, et al: Action myoclonus-renal failure syndrome: characterization of a unique cerebro-renal disorder. *Brain* 2004, 127:2173-2182.

Berkovic SF, Dibbens LM, Oshlack A, et al: Array-based gene discovery with three unrelated subjects shows SCARB2/LIMP-2 deficiency causes myoclonus epilepsy and glomerulosclerosis. *Am J Hum Genet* 2008, 82:673-684.

11. Rhombencephalic PML with JC virus granular neuronopathy

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Koralnik IJ. JCV granule cell neuronopathy: a novel clinical syndrome distinct from PML. *Ann Neurol* 2005; 57: 576-80.

Wüthrich C, Cheng YM, Joseph JT, et al. Frequent infection of cerebellar granule cell neurons by polyomavirus JC in PML. *J Neuropathol Exp Neurol* 2009; 68 (1): 15-25.

Tan CS, Koralnik IJ. PML and other disorders caused by JCV: clinical features and pathogenesis. *Lancet Neurol* 2010; 9: 425-37.

12. Ganglioneurocytoma, WHO grade 2

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Queen Elizabeth II Health Sciences Centre and Dalhousie University, Halifax, Nova Scotia, Canada; and Moncton Hospital, Moncton, New Brunswick

Tsai CY, Tsai TH, Lin CH, Cheng YH, Lieu AS. Unusual exophytic neurocytoma of thoracic spine mimicking meningioma: a case report and review of the literature. *Eur Spine J*. 2011 July; 20(Suppl 2): 239–242.

Gokhan GA, Gurer IE, Akyuz M, Tuncer R. A case of extraventricular neurocytoma of the spinal cord. *Neuropathology* 2008; 28:322–325.

13. Plasma cell myeloma with small lymphocytic morphology, confirmed CCND-1 - IgH t(11;14) translocation and massive amyloid deposition.

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Lehman NL, Horoupian DS, Warnke RA, Sundram UN, Peterson K, Harsh GRt (2002) Dural marginal zone lymphoma with massive amyloid deposition: rare low-grade primary central nervous system B-cell lymphoma. Case report. *Journal of neurosurgery* 96: 368-372 Doi 10.3171/jns.2002.96.2.0368

Tu PH, Giannini C, Judkins A Ret al. (2005) Clinicopathologic and genetic profile of intracranial marginal zone lymphoma: a primary low-grade CNS lymphoma that mimics meningioma. *J Clin Oncol* 23: 5718-5727 Doi 10.1200/JCO.2005.17.624

Abdalla S, May PC, Garimberti E, Naresh KN (2011) Bone marrow trephine biopsy findings in myeloma with small-lymphoid cells and CCND1 translocation. *Am J Hematol* 86: 1038 Doi 10.1002/ajh.22128

Specht K, Haralambieva E, Bink Ket al. (2004) Different mechanisms of cyclin D1 overexpression in multiple myeloma revealed by fluorescence in situ hybridization and quantitative analysis of mRNA levels. *Blood* 104: 1120-1126 Doi 10.1182/blood-2003-11-3837