

Leukoencephalopathy with calcifications and cysts: a case study with long-term follow-up

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Chief complaints

- A 34-year-old right-handed man with normal early development had:
 - Slight weakness in his right leg;
 - Headaches
 - Seizures
 - Memory difficulties.

History of illness

- No family history of epilepsy or other genetically determined disorders;
- Age 14: new-onset severe headache, followed by vomiting and a bilateral tonic-clonic seizure;
- Age 19: Focal to bilateral tonic-clonic seizures recurred;
- Age 27: Persistent headache; MRI revealed large bilateral parietal cysts that were subsequently neurosurgically drained;
- During the next two years: mood fluctuations, memory problems, episodes of confusion, ataxia, and writing difficulties developed.

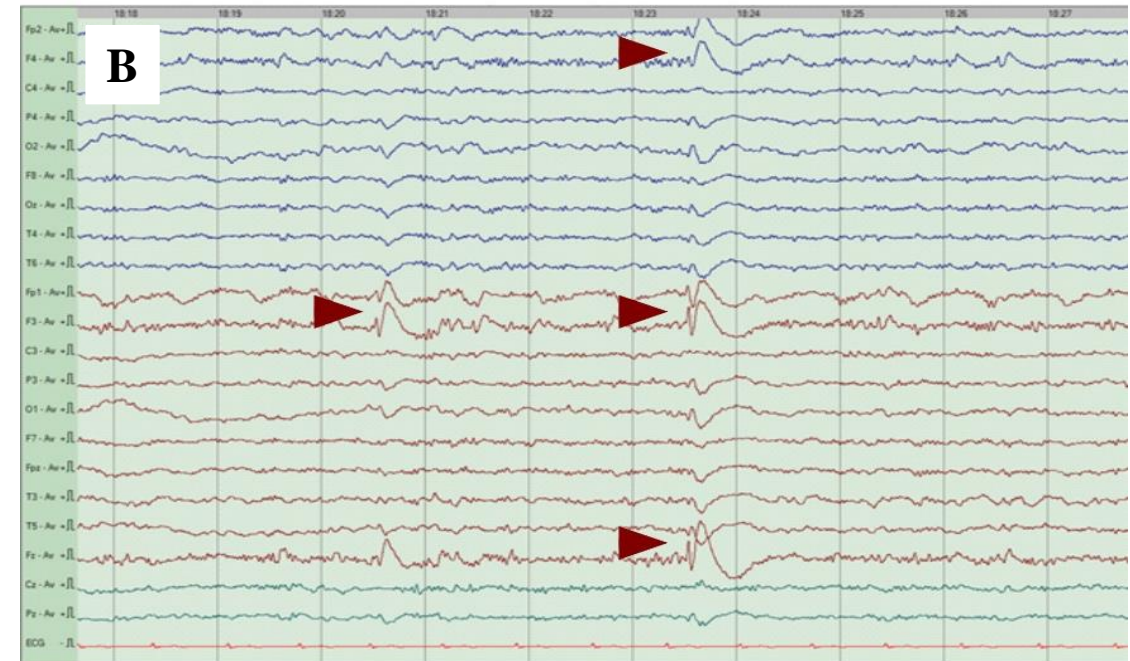
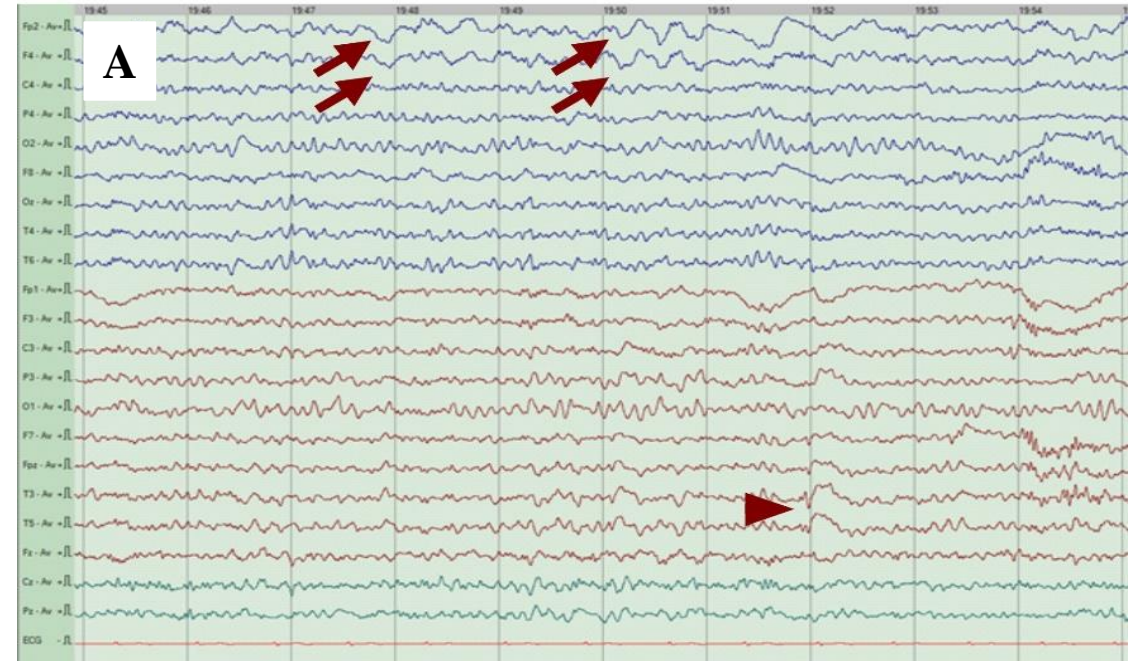
Examinations

- Neurological examination:
 - Receptive aphasia, right leg weakness, bilateral positive Babinski reflexes, ataxic gait, discoordination, Gerstman syndrome; diffuse muscle hypertonus;
- Neuroophthalmological investigations with fundoscopy, visual acuity, and visual field - no abnormalities;
- Serological test results for Echinococcus, Taenia solium, HIV 1 and 2, and Treponema pallidum were negative;

Follow-up EEG

A - Focal slowing in the right frontal and fronto-central regions (arrows) and spike-wave complexes in the left temporal region (arrowheads) (common average montage; LFF: 0.3 sec, HFF: 30 Hz, Sen.: 150 μ V/cm) (29 y/o);

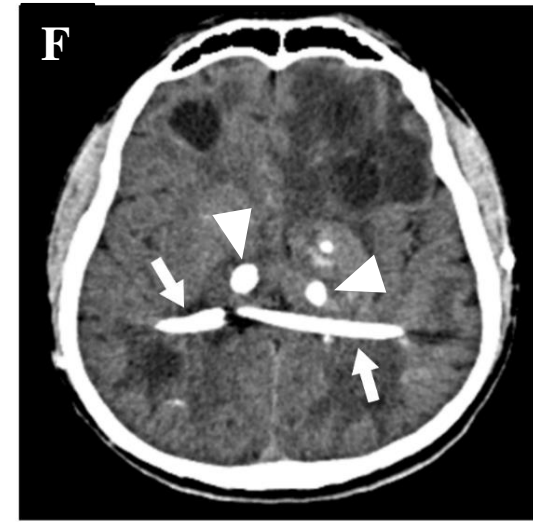
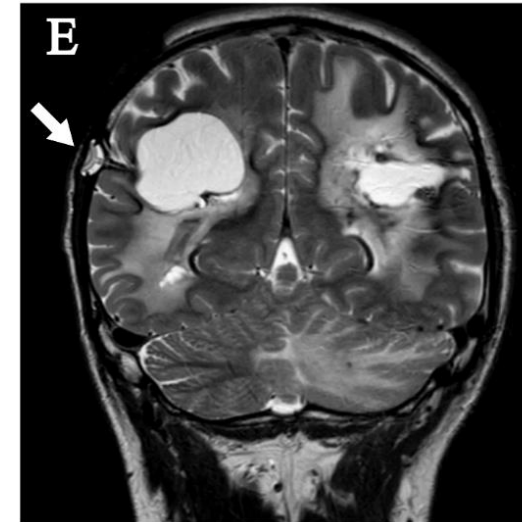
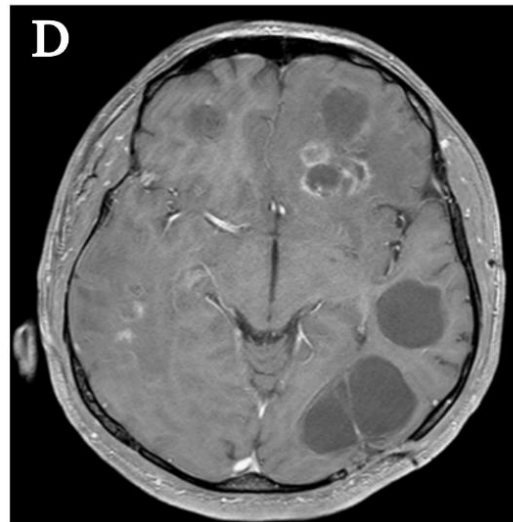
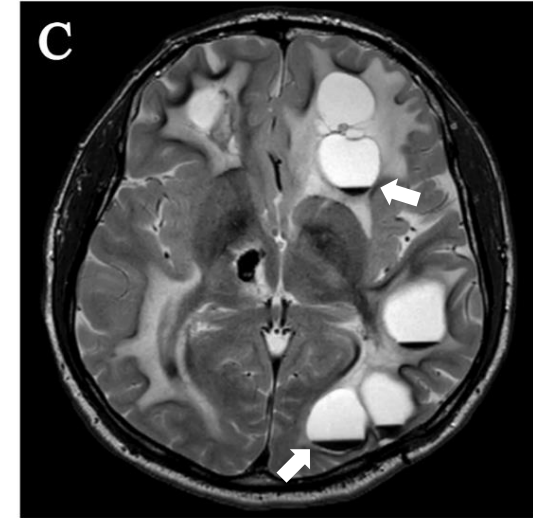
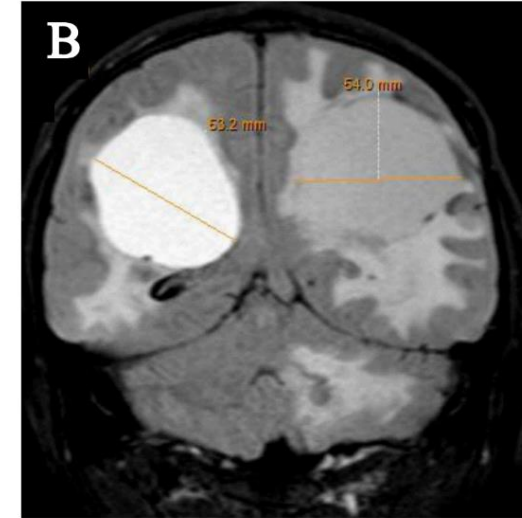
B - Spike-wave complexes (arrowheads) in the left frontal, fronto-central, and sagittal-fronto-central region with the occasional emergence of the same patterns in the right hemisphere (common average montage; LFF: 0.1 sec, HFF: 30 Hz, Sen: 150 μ V/cm) (30 y/o);



MRI brain

The classic neuroradiological triad of LCC: edematous leukoencephalopathy, cerebral calcification, and parenchymal cysts);

A - Axial T2- weighted and **B** -coronal T2-weighted fluid attenuated inversion recovery image - bilateral fronto-parietal cysts with dimensions (30 y/o); **C** - Axial T2-weighted image with a fluid-fluid level in the cysts (arrows); **D** – post-contrast T1W image with various degrees of cyst wall enhancement; **E** - coronal T2-weighted image shows shunted parietal cysts (arrow - shunt over the right parietal cyst); **F** - CT image with cysts, bilateral basal calcifications (arrowheads) and cysto-ventriculoperitoneal shunts (arrows) (images **C** to **F**, 34 y/o);



Treatment and final diagnosis

- Various anti-seizure medication (ASM) therapies have been used with intermittent effectiveness;
 - Last ASM therapy - carbamazepine-1200 mg/day and phenobarbital 100 mg/day, with seizures occurring once every 3-4 months;
- Ventriculo-cysto-peritoneal shunting was performed several times with initial remarkable improvement, however, the neurological symptoms gradually reappeared;
- **Final diagnosis** - leukoencephalopathy with cerebral calcifications and cysts (Labrune syndrome).

References for further reading

- Labrune P, Lacroix C, Goutières F, de Laveaucoupet J, Chevalier P, Zerah M, et al. Extensive brain calcifications, leukodystrophy, and formation of parenchymal cysts: a new progressive disorder due to diffuse cerebral microangiopathy. *Neurology* 1996;46:1297–301.
- Jenkinson EM, Rodero MP, Kasher PR, Uggenti C, Oojageer A, Goosey LC, et al. Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. *Nat Genet* 2016;48:1185–92. <https://doi.org/10.1038/ng.3661>.
- Kobets A, Oriko D, Groves M, Robinson S, Cohen A. Surgical considerations in Labrune syndrome. *Childs Nerv Syst* 2021;37:1765–70. <https://doi.org/10.1007/s00381-020-04861-7>.