Leukoencephalopathy With Calcifications and Cysts Associated With SNORD118 Variants

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A 67-year-old woman presented with gradually progressive gait impairment. Examination revealed left third nerve palsy, ataxia and lower limb spasticity.

Twenty-four years previously, hydrocephalus and multifocal cerebral cysts necessitated ventriculostomy and cyst biopsy (figure 1), followed by foramen magnum decompression 2 years later and, after a further 4 years, cerebellar haemorrhage evacuation.

Leukoencephalopathy with calcification and cysts, a rare, recently described autosomal ribosomopathy(1), was suspected at re-presentation on the basis of neuroimaging (figure 2). The diagnosis was confirmed following re-review of histopathology. Genetic screening detected a likely pathogenic variant in SNORD118 (n.*9C>T) and a variant of unknown significance (n.126C>T).

Progressive, multifocal symptoms are characteristic(2).
Figure 1. Histopathology of brain biopsy.

A. Gliotic brain with Rosenthal fibres (short arrows) and cysts (thin arrows) caused by microhaemorrhage secondary to underlying angiopathy (large arrow). B. Angiopathy with abnormal vessels and luminal thrombosis. C. Dystrophic calcification within the neuropil. This combination is characteristic of leukoencephalopathy with calcifications and cysts.
Figure 2. T2 weighted MRI.

Key radiological features of leukoencephalopathy with calcifications and cysts are demonstrated, with white matter hyperintensity consistent with leukoencephalopathy, calcification (confirmed on CT; dashed arrow) and cyst (solid arrow).
References:


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