A singular case of asymptomatic leukoencephalopathy in a 60-year-old male

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Objective

Adult-onset leukoencephalopathies are considered to be rare. Depending on their aetiologies, they display wide variability as regards the modality of transmission, the clinical and instrumental findings and the progression rate. Here we report a case of leukoencephalopathy with impressive radiological findings in an asymptomatic adult man.

Patient and Methods

A 60-year-old man following an ophthalmological consultation for spontaneous retinal detachment practiced a brain MRI scan which evidenced diffuse white matter abnormalities. Familiarity for neurological disorders was denied and the patient's cardiovascular risk profile was low. He underwent an extensive diagnostic protocol including neurological and neuropsychological evaluation, laboratory assays, lumbar puncture, EEG, motor, visual and auditory potentials. Brain and spine MRI as well as ophthalmological evaluation were performed once again. The diagnostic protocol was completed by means of serum VLCFA assay and genetic testing for *Notch3* mutations.

Results

Neurological examination was completely negative and cognitive functions were preserved. Serum and CSF analyses were normal. Visual evoked potentials revealed a moderate increase in P100 wave latency. Brain MRI evidenced diffuse deep and subcortical white matter hyperintensity in T2-weighted and FLAIR scans, especially in basal frontal, temporal and retro-trigonal regions bilaterally. Capsular fibers and pyramidal tracts up to the brain stem were involved mostly to the right side. There was no contrast enhancement following gadolinium administration. This pattern was considered to be compatible with an atypical presentation of CADASIL; however, after excluding adreno-leukodystrophy thanks to the normal concentration of serum VLCFA, *Notch3* gene analysis was performed and it resulted negative.

Discussion and Conclusions

Asymptomatic leukoencephalopathies, an enigmatic chapter of clinical neurology, may be accidentally found through neuroimaging studies in adults who undergo MRI scans for different reasons. The neuroimaging pattern may suggest their aetiology and guide any genetic analysis. For those with aspects suggestive of white matter damage of vascular origin, *Notch3* and *COL4A1* genes must be examined first. In our case, capsular and temporal involvement was evocative of CADASIL, in spite of the absence of dementia and the other core clinical features. Independently form the genetic diagnosis, which can even be unattainable with the current knowledge, cases like this pose difficult dilemmas for what concerns the prognosis and the risk of recurrence. Therefore a careful clinical and instrumental follow up, even with functional techniques, is needed to clarify if the initial finding has to be considered a preclinical manifestation of an overt disease or just a structural abnormality without pathophysiological correlations.

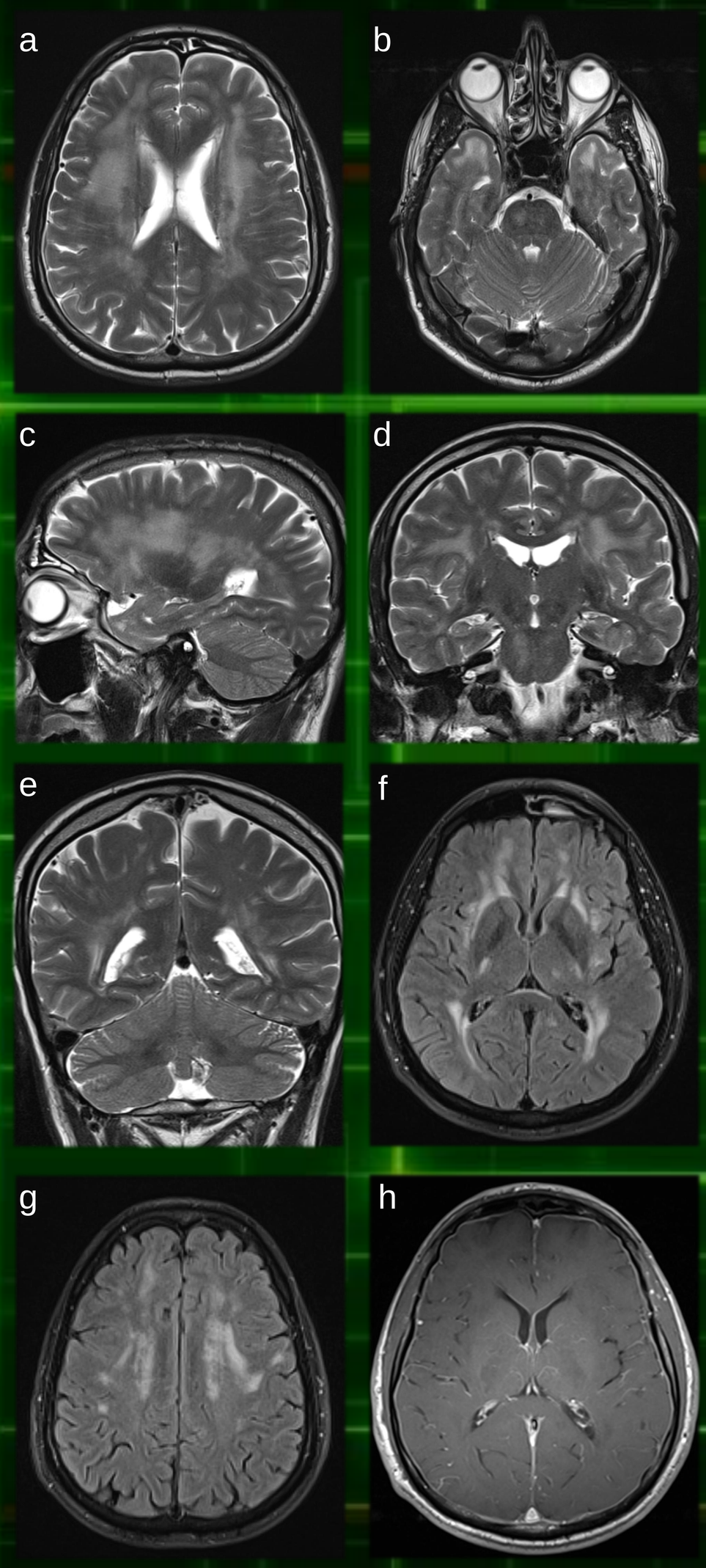

Figure 1.
Cerebral MRI with T2-weighted (a-e), FLAIR (f,g) and T1-weighted post-contrast (h) images. An extensive hyperintensity in subcortical and deep white matter is evident, mainly spreading in basal frontal, temporo-polar and retro-trigonal regions. Capsular fibers and corticospinal tracts are involved up to the brain stem. There is no contrast enhancement following gadolinium administration.

References

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