Skin Biopsy-based Diagnosis of CADASIL with Atypical MRI Findings

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A 37-year-old man whose father had died after a third stroke at 53 years of age and whose grandfather had died of cerebral infarction in his 40’s underwent brain MRI due to a headache. He had no history of symptomatic stroke; however, the MRI examination showed white matter lesions on FLAIR images (Picture). Although neither the temporopolar white matter nor external capsule, characteristic sites of lesions in cases of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) (1), were clearly involved, the patient’s family history prompted an examination for CADASIL. Consequently, no mutations were found in NOTCH3 exon 3 or 4, where pathogenic mutations are often detected in Japanese patients with CADASIL. In contrast, a skin biopsy revealed typical findings of CADASIL (Supplementary material) (2), and further genetic analyses of the NOTCH3 gene disclosed the p.R332C mutation in exon 6. Although further studies are needed, conducting skin biopsies is worthwhile in patients with symptoms clinically suspicious of CADASIL, even if the MRI findings are not typical.

The authors state that they have no Conflict of Interest (COI).

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References

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