CHARACTERISTIC FEATURES AND PROGRESSION OF ABNORMALITIES ON MRI FOR CARASIL

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Objectives
Cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL), a hereditary form of cerebral small vessel disease, is characterized by early adult-onset dementia associated with white matter lesions, spondylosis deformans and alopecia. The objective of this study was to clarify the characteristic and sequential brain MRI findings for genetically diagnosed CARASIL.

Methods
Seven patients with CARASIL carrying HTRA1 mutations (representing 6 Japanese families) were included in this study. Eighteen brain MRIs were reviewed and evaluated with a new rating scale based on scoring for abnormal hyperintense lesions and atrophy.

Results
At the last follow-up MRI, all patients had hyperintense lesions on T2-weighted images of the frontal white matter, anterior temporal lobe, external capsules, and thalami. Patients with longer time from the onset of cognitive impairment had higher MRI severity score. The atrophy advanced, followed by white matter lesion progression. During the early stage, hyperintense lesions were observed in the frontal white matter, external capsule, andpons. The MRI findings are not so different from those observed with cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL). During the late stage, the arc-shaped hyperintense lesion from the pons to the middle cerebellar peduncles, which we designated the “arc sign,” became evident. The arc sign was a characteristic finding for CARASIL in the advanced stage.

Conclusion
These characteristic MRI findings for CARASIL are useful for selecting patients for genetic testing. The rating scale correlates well with disease duration and might be useful for assessing disease progression.