

Variable clinical features in Japanese families with autosomal dominant lateral temporal lobe epilepsy (ADLTLE)

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Background and Objective: Autosomal dominant lateral temporal lobe epilepsy (ADLTLE) is one of the hereditary partial epilepsies caused by abnormality of a gene of leucine-rich glioma-inactivated 1 (LGI-1) or epitempin. There were rare reports of its clinical features in Japanese family.¹⁻³ Thus, we tried to clarify the clinical features and pathophysiology of ADLTLE in Japanese families

Methods: In 8 patients out of 15 epileptic members of 3 Japanese families with a diagnosis of ADLTLE treated in Kyoto University Hospital, we investigated clinical features, routine EEG, long-term video/EEG monitoring (1 patient), MRI (1.5T machine in 6 patients, 3T machine in 3 patients), FDG-PET (3 patients) by SPM analysis and MEG (5 patients). In MEG with 306-channel whole-head system (VectorView, Elekta Neuromag), spontaneous epileptiform discharges and auditory evoked fields (AEFs) for pure tone burst (1,000Hz, 50dB SL, pulse width of 10ms, interval of 1,000 ± 70 ms) were recorded. Gene analysis of LGI-1 was done in 3 patients in 2 families.

Results: (a) Mean age at onset was 21 years. Six patients had rare occurrence of generalized tonic-clonic seizures, 6 had partial seizures from the lateral temporal area and 4 had seizures from the mesial temporal area, both of which occurred within the same families. Panic attacks occurred in many patients, all in one family. Three patients had seizures induced by auditory stimuli. Two patients each from the different families had intractable seizures. (b) Epileptiform discharges were recorded only in one patient. Out of 5 patients with MRI examination, one showed a small left superior temporal gyrus, and one had hippocampal atrophy. All the 3 patients with FDG-PET study showed hypometabolism in the unilateral temporal area. MEG showed no spikes in 5 patients investigated, and enhanced AEFs were observed in 3 patients, all of whom had stimulus-sensitive, auditory auras. (c) Two patients in one family showed the same point mutation at exon 8 in LGI-1, and another in the other family had one in the different codon at exon 8 in LGI-1.

Conclusions: In Japanese families ADLTLE, 1) seizures are usually well controlled, but intractable in some patients in different families. 2) In addition to the seizures arising from the lateral temporal area, seizures from the mesial temporal area and panic attacks were also frequently observed, which varied among patients in the same family. These imply modifying factors in manifesting seizure disorders in ADLTLE. 3) Hyperexcitability of the auditory cortices as revealed by MEG can share the common generator mechanism to the stimulus-sensitive, auditory auras.

References

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