

## Myoclonic epilepsies in Japan

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When we engaged ourselves in genetic study of Dentatorubral Pallidoluysian Atrophy (DRPLA)<sup>1-3</sup>, we found that juvenile cases with DRPLA show progressive myoclonic epilepsy. Progressive myoclonic epilepsy is the syndrome of progressive disease with cerebellar ataxia, myoclonus, epilepsy, and mental deterioration. DRPLA is a common cause for progressive myoclonic epilepsy in Japan. Other causes in Japan are myoclonic epilepsy associated with ragged-red fiber (MERRF), Lafora disease, and Unverricht-Lundborg disease. These progressive myoclonic epilepsy-causing diseases are all hereditary in various forms. Thus it is important to take family history in detail before molecular testing and for diagnosis. During molecular genetical research on such myoclonic epilepsy syndromes, we could find familial adult-onset cases with myoclonic epilepsy in autosomal dominant inheritance showing benign courses. The syndrome, benign adult familial myoclonic epilepsy was recently recognized in Japanese families. We assigned the gene locus to the distal long arm of chromosome 8, 8q23.3-q24.11 by linkage analysis in a large Japanese kindred with a maximum multipoint lod score of 5.42 for the interval between D8S555 and D8S1779.<sup>4</sup> DRPLA and benign adult familial myoclonic epilepsy familial cases have been first reported in Japan and the frequencies of these

two diseases appear to be much higher compared to those in other countries. We have recently identified the gene *CHAC* liable for other symptomatic epilepsy, chorea-acanthocytosis.<sup>5</sup> Chorea-acanthocytosis is also more frequently observed in Japanese population. Hereditary disorders are often dependent on ethnic backgrounds and geographical areas, even among the same ethnic populations.

### REFERENCES

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