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The temporal profiles of changes in nerve excitability indices in familial Amyloid Polyneuropathy

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Familial amyloid polyneuropathy (FAP) is a rare genetic disorder frequently caused by a mutation in transthyretin (TTR) gene. There are two cardinal clinical features of TTR-FAP, autonomic dysfunction and sensorimotor polyneuropathy. To elucidate pathophysiological mechanisms of FAP, we evaluated the changes of the electrophysiological parameters in both patients (TTR, p.A97S mutation) and mutant carriers. The clinical phenotypes, neuropathy severity scores (NDS and ONLS), and the indices from nerve excitability test (NET) were collected. The median NDS and ONLS scores for patients are 54.5 (range, 13-82) and 4.5 (3-9), respectively. The nerve conduction studies showed markedly reduced CMAP and SNAP in patients, but unremarkable in carriers. NET data obtained from ulnar nerves of carriers showed increase of threshold, rheobase, and refractories in recovery cycle (RC). In the patient group, the NET study showed prolonged latency, reduced threshold elevation during hyperpolarizing threshold electrotonus (TE) at 10~40 ms (TEh(10-20ms) and TEh(20-40ms)), and increased refractoriness in the motor axons. There were prolonged latency, increased threshold reduction and S2 accommodation in depolarizing TE, lowed TEh(slope 101-140 ms), delayed time to overshoot after hyperpolarization, increased refractoriness and superexcitability in RC in sensory axons. The regression models demonstrated that the increase of refractoriness and prolonged relatively refractory period are correlated to the disease progression from carrier to patients. A defect in sodium current might be an early pre-symptomatic pathophysiological change considering the marked increase of refractoriness at short-width stimulus. Furthermore, the shallower slope of recovery, delayed time to overshoot after hyperpolarizing TE, and increase of superexcitability suggest a focal disruption of basal lamina and myelin membrane leading to the increase of intermodal capacity.

Biography

Ming-Jen Lee has completed his Neurology resident training at the age of 31 years from National Taiwan University Hospital (NTUH), Taipei, Taiwan. He completed his PhD course from Institute of Neurology, University College London, UK. He is an associate professor in the Department of Neurology, NTUH. He is in charge of the neurogenetic clinic for ten more years and is interested in the disease mechanism of peripheral neuropathy and neurocutaneous disorders. He has published more than 20 papers in reputed journals.

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