Immunologic and genetic testing in peripheral neuropathies

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Immune-mediated neuropathies and inherited neuropathies comprise major subgroups of peripheral neuropathies. Steps of diagnosis is not different from other neuromuscular diseases, and clinicians should adhere to the basic principles of clinical approach of neuromuscular disease such as history taking including developmental and familial history, general and neurological examination, laboratory tests, electrodiagnosis, and occasionally nerve biopsy. Establishing a correct clinical impression is very important to make a confirmative diagnosis using immunologic or genetic tests. Immunologic tests are important for the diagnosis of various immune-mediated neuropathies in many aspects. The result may confirm the diagnosis, support the proposed mechanism of the disease, or provides an important information of the nature of the disease. Although clinicians do not usually perform the tests by themselves, they should understand the principles, clinical significance, and shortcomings of the various immunological tests. In this lecture, basic principles, interpretation, and clinical significance of important immunologic tests in establishing a diagnosis of immune-mediated neuropathy will discussed. Inherited neuropathies are the most common genetic disease in the field of neurology. However, their diagnosis can be challenging due to marked genotypic and phenotypic variability. For an example, Charcot-Marie-Tooth disease (CMT), which is the most common form of inherited neuropathy, is associated with more than 70 different genes. Recent advances in molecular genetics provides effective guidelines of genetic testing in patients with CMT depending on the inheritance pattern, clinical severity, and degree of slowing in nerve conduction velocities. Basic principles and shortcomings of each genetic tests, suggested algorithms of genetic testing in CMT for clinicians will be discussed in this lecture.