Peripheral Nerve Disorders: Chapter 12. DNA testing in hereditary neuropathies (Handbook of Clinical Neurology)

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The inherited neuropathies are a clinically and genetically heterogeneous group of disorders in which there have been rapid advances in the last two decades. Molecular genetic testing is now an integral part of the evaluation of patients with inherited neuropathies. In this chapter we describe the genes responsible for the primary inherited neuropathies. We briefly discuss the clinical phenotype of each of the known inherited neuropathy subgroups, describe algorithms for molecular genetic testing of affected patients and discuss genetic counseling. The basic principles of careful phenotyping, documenting an accurate family history, and testing the available genes in an appropriate manner should identify the vast majority of individuals with CMT1 and many of those with CMT2. In this chapter we also describe the current methods of genetic testing. As advances are made in molecular genetic technologies and improvements are made in bioinformatics, it is likely that the current time-consuming methods of DNA sequencing will give way to quicker and more efficient high-throughput methods, which are briefly discussed here.

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