

**American Academy of Neurology
Neurogenetics Section
Residency Core Curriculum**

10/01/01

Introductory Comments

The impending completion of the Human Genome Project, and the vast amount of information that is likely to be gained from its completion, has already revolutionized the practice of medicine. There is hardly a specialty or sub-specialty of medicine that has not been impacted by the revolution in Genetics and Genetic Information in its everyday practice. This statement is certainly no truer than in the field of Neurology, in which journals are inundated with articles discussing genetics, neurologists are contacted by an ever growing list of laboratories offering genetic tests for particular inherited neurological disorders, and an increasing number of patients and their family members who are requiring and discussing information related to inheritance and genetic etiology of a spectrum of neurological disorders. The members of the Neurogenetics section of the Academy believe that it is a particularly important and pertinent time to be including formal training in Neurogenetics into the approved neurology residency program. The guidelines presented below are items that we believe are sufficient for minimal, approved standards. It is the hope of the section that a resident would receive more training than the minimal guidelines, due to the perceived impact that the Genome Project is likely to have on medical practice over the next ten-twenty years.

Clinical requirements

- Basic skills in the interview and examination of patients and families with neurogenetic diseases (i.e., how to take a meaningful family history).
- Knowledge of the differential diagnoses for the various clinical presentations of neurogenetic disorders (neuromuscular, movement disorders, cognitive disorders, behavioral disorders).
- Knowledge of patterns of mendelian inheritance and non-mendelian inheritance and basic molecular genetics concepts, including types of mutations.
- Knowledge of the differences between tests that determine disease-causing mutations, and those that detect/determine “at-risk” alleles/genotypes.
- Basic training in the recognition of common dysmorphic syndromes with neurological outcomes, such as Down’s, Turner’s, XLMR syndromes.
- Knowledge of the appropriate laboratory investigations for diagnosis of neurogenetics disorders, including the sensitivity and specificity of DNA-based tests, as well as their interpretation.
- Knowledge on how to access pertinent and up-to-date information relating to the diagnosis of neurogenetic disorders and the tests for these disorders.
- Knowledge in the *diagnosis and* management of a subset of important inherited disorders of the nervous system such as inherited ataxias, Huntington disease, inherited neuropathies, muscular dystrophies, phakomatoses).

- Basic training in genetic counseling of at-risk individuals; ability to acknowledge which cases need referral to an appropriate geneticist for further discussion.
- Appreciation of the ethical issues involved in genetic testing and counseling.

1. Definitions:

Neurogenetic disorders are a subsection of neurology that includes diseases of the central and peripheral nervous systems and muscle that are caused or primarily influenced by genetic or inherited factors.

2. Content of Subjects to be Taught:

Residents who are training in neurogenetic disorders must be provided with a basic and advanced background in the molecular biology, neurophysiology, neuroanatomy, neuropharmacology, and neuropathology of these conditions. They also must be exposed to the clinical presentation, laboratory investigation, management and genetic counseling of a wide-range of neurogenetic disorders. They will be trained in basic clinical and/or laboratory research methods. It is well appreciated that no such formal training programs exist, and that a recommended curriculum needs to be developed by the Section on Neurogenetics. In addition, the Section believes that as part of the Neurology residency, that a total of one-month in a neurogenetics outpatient clinical experience be required.

3. Personnel Needed for the Trainee:

The Director and teaching staff of the neurogenetic residency component must include at least one full-time faculty member with expertise in neurogenetic disorders

4. Qualifications of the Trainers:

The Program Director must have completed at least 3 years of formal academic training and experience in the pathophysiology, evaluation and management of neurogenetic disorders.

5. Facilities Need for the Training:

This residency component must be within a department or division of neurology or human genetics and have facilities adequate for the educational program. *Residents* must have ready access to a major medical library. There must be access to an on-site collection of appropriate texts and journals at each institution participating in the *residency* program.

6. Set-Up for the Training:

The residency must include a clinical experience in which the *resident* develops expertise in the evaluation and treatment of patients with a variety of neurogenetic disorders. The program design and/or structure must be approved by the sponsoring institution's residency review committee for neurology or medical genetics as part of the regular review process. The residency review committee for neurology must approve participation by any institution providing one month or more of training in a program.

7. Methods of Training:

The resident must have instruction and practical experience sufficient to develop diagnostic and therapeutic skills necessary to provide care for patients with neurogenetic disorders. The clinical experience must include opportunities to observe, evaluate, and manage patients with a wide variety of hereditary diseases of the nervous system. Clinical experience must include inpatient consultation,

outpatient care, and knowledge of the appropriate use of support services in pathology, radiology, electrodiagnosis and genetic and biochemical laboratory medicine. The residency program will conduct formal lectures and teaching conferences. These conferences must include discussions of the molecular biology, neuropathology, neurophysiology, and the clinical diagnosis and management of neurogenetic disorders.

8. List of References/Resources:

Baker, D., et al: A Guide to Genetic Counseling. Wylie-Liss, 1998.

Baraitser, M: The Genetics of Neurological Disorders. Oxford University Press (2), 1990.

Jorde, L, Carey J, Bamshad, M, White R: Medical Genetics. Mosby (2), 1999.

Kandel, E, Schwartz, J, Jessell, T: Principles of Neural Science. Elsevier (3), 1991.

McKusick, V, Francomano, C, Antonarakis, S, Pearson, P: Mendelian Inheritance In Man: A Catalog of Human Genes and Genetic Disorders. The Johns Hopkins University Press (12), 1999.

Pulst, S: Neurogenetics. Oxford University Press. 2000

Rosenberg, R, Prusiner, S, DiMauro, S, Barchi, R: The Molecular and Genetic Basis of Neurological Disease. Butterworth-Heinemann (2), 1997.

Vogel, F, Motulsky, AG: Human Genetics: Problems and Approaches. Springer (3), 1997.

Websites:

www.geneclinics.org

www.ncbi.nlm.nih.gov/omim