

AMERICAN ACADEMY OF NEUROLOGY NEUROGENETICS FELLOWSHIP CORE CURRICULUM

1. Introduction

Specialists in Neurogenetic Disorders possess specialized knowledge in the science, clinical evaluation and clinical management of genetic diseases of the nervous system. This encompasses knowledge of the pathophysiology, pathology, diagnosis, and treatment of these disorders at a level that is significantly beyond the training and knowledge expected of a general neurologist.

2. Goals and objectives:

The goal of fellowship training in neurogenetic disorders is to facilitate the development of neurologists with subspecialty expertise in hereditary diseases of the nervous system. These subspecialists will serve to advance the science and understanding of neurogenetic disorders. They will also provide diagnoses, counseling and care for patients with neurogenetic disorders, and will educate other physicians (including internists, family practitioners, neurology residents, general neurologists), other healthcare personnel, and the public about these disorders.

All subspecialists in neurogenetic disorders must acquire expertise in the evaluation and management of patients with hereditary diseases of the nervous system. This includes:

- **Skill in the interview and examination of patients and families with neurogenetic diseases.**
- **Knowledge of the differential diagnoses for the various clinical presentations of neurogenetic disorders,**
- **Knowledge of patterns of mendelian inheritance and non-mendelian inheritance.**
- **Training in the recognition of common dysmorphic syndromes.**
- **Knowledge of the appropriate laboratory investigations for diagnosis of neurogenetics disorders, including the sensitivity and specificity of DNA-based tests.**
- **Expertise in the management of a subset of inherited disorders of the nervous system such as inherited ataxias, Huntington disease, inherited neuropathies, etc.**
- **Expertise in counseling patients and at-risk individuals regarding penetrance, expressivity, prognosis, and recurrence risk.**
- **Knowledge of legal and ethical issues regarding DNA-based and non-DNA-based genetic testing.**

Within the spectrum of neurogenetic disorders, individuals and programs may develop a variety of specialized skills with the above objectives.

3. 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.

4. Content of Subjects to be Taught:

Fellows who are training in neurogenetic disorders must be provided with an advanced and extensive 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 clinical and/or laboratory research methods and the process of writing manuscripts for publication and grant applications.

5. Prerequisites for the Trainee:

The neurogenetic disorders fellow must have completed and accredited residency program in either child neurology or adult neurology.

6. Personnel Needed for the Trainee:

The Director and teaching staff of the neurogenetic fellowship must include at least one full-time faculty member with expertise in neurogenetic disorders for each fellow on the program (i.e., a ratio of full-time neurogenetic faculty to fellows of at least 1:1).

7. 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. All members of the faculty should have an appointment in good standing on the medical staff of an institution participating in the fellowship program.

8. Facilities Need for the Training:

The neurogenetic disorders fellowship must be within a department or division of neurology or human genetics and have facilities adequate for the educational program. Fellows 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 fellowship program.

9. Set-Up for the Training:

The fellowship program must include a clinical experience in which the fellow 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. Participation by any institution providing 2 months or more of training in a program must be approved by the residency review committee for neurology.

The Program Director is responsible for the overall coordination of the program. The Program Director is responsible for the selection of fellows for appointment to the program in accordance with institutional and departmental policies and procedures. The Program Director is also responsible for selection and supervision of the teaching staff and personnel at each institution participating in the program. The Program Director and faculty are responsible for instruction and supervision of the fellows in the program.

10. Methods of Training:

The fellow 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 support services in pathology, radiology, electrodiagnosis and genetic and biochemical laboratory medicine.

Basic science training may include laboratory experience in molecular, genetic or biochemical aspects of neurogenetic diseases or basic mechanisms relevant to those diseases under the supervision of a qualified scientist.

The fellowship 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.

Clinical assignments should include progressively increasing responsibilities for patient care with direct supervision by the appropriate faculty and/or staff. Clinical assignments need not be identical for each resident. Subspecialty experience should accommodate the fellow's individual interest.

The fellow in clinical neurogenetics should take an active role in the teaching and training of neurology residents.

11. Timetable for Training:

The fellowship may be 1 to 3 years duration and must include the equivalent of at least 6 months of full-time patient interaction (including inpatient, outpatient, and clinical laboratory evaluation).

12. Methods of Evaluation of the Trainee:

The Program Director, with participation of members of the teaching staff, shall:

- At least semi-annually evaluate the knowledge, skills, and professional growth of the fellows, using appropriate criteria and procedures.

- Communicate each evaluation to the fellow in a timely manner.
- Provide a written final evaluation for each fellow who completes the program. This evaluation should verify that the fellow has demonstrated sufficient professional ability to practice competently and independently. This final evaluation should be part of the fellow's permanent record retained by the institution.

13. Methods of Evaluation of the Training Program:

The educational effectiveness of the neurogenetic fellowship must be evaluated in a systematic manner. The quality of the curriculum and the extent to which the educational goals have been met must be assessed. Regular evaluations by fellows should be utilized in this process.

14. Mechanisms for Feedback:

As noted above, fellows should complete evaluations of the faculty and curriculum at least every 3 months during the fellowship. In addition, the Program Director or designated neurogenetic faculty member must be available to meet with each fellow on a weekly basis to discuss any concerns that the fellow(s) have regarding the program. The Program Director should also be available on a weekly basis to discuss concerns that the faculty may have with regard to the conduct of the fellowship.

15. Methods of Constantly Upgrading Knowledge:

The faculty and fellows within the program are expected to participate in continuing education in order to expand their knowledge base and remain up-to-date in their understanding of neurogenetic disorders. Activities to accomplish this goal may include:

- Active participation in clinical discussions, rounds and conferences in a manner that promotes the spirit of inquiry and scholarship.
- Participation in journal clubs and research conferences.
- Active participation in regional or national professional and scientific societies, particularly through presentations at the organization's meetings and publication in journals.
- Participation in basic science and/or clinical research.

16. 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.

www.geneclinics.org

www.ncbi.nlm.nih.gov/omim

17. Continuing Medical Education Needed:

At least 30 hours of category 1 of continuing education in neurogenetic disorders must be completed by the neurogenetic specialist every five years in order to maintain qualification.