Educational Programs in Neurogenetics

Advances in genetics, particularly molecular biology, have increased in an exponential fashion in the past two decades. These advances have affected the practice of all branches of medicine, but particularly the field of neurology.

In addition to rare single gene mutations leading to neurological diseases such as Huntington disease or Tay Sachs disease, more common entities such as troke, epilepsy, dementia, multiple sclerosis and autism are usually the result of multifactorial or complex inheritance, with interaction of genetic and acquired factors. These are associated with variations in a number of predisposing susceptibility genes. The latter are now being identified with new techniques such as genome-wide association studies (GWAS) and new generation sequencing.

A number of chromosomal abnormalities have now been identified which are usually associated with mental retardation and malformations of cortical development. More recently, array comparative genomic hybridization (array CGH) has allowed us to find copy number variations (CNV´s) (submicroscopic deletions and duplications) which result in a number of malformation syndromes, as well as mental retardation, autism and dysmorphic features. CNV´s also account for a small proportion of other neurological disorders.

Mitochondrial disorders due to mutations of the mitochondrial genome as well as autosomal mutations affecting mitochondrial function result in many additional neurological disorders.

Due to the complexity and rapid advances in the field, it is important to provide education in neurogenetics to practicing neurologis in order to help them to understand the literature on neurogenetic disorders, and to improve diagnosis, prognosis, counseling, and eventual treatment in a variety of neurological disorders.

In the past, the Neurogenetics Research Group has organized a number of international symposia and workshops dealing with different types of neurological disorders (e.g. ataxias, epilepsies, dementias, etc.) as part of the annual meetings of the Neurogenetics Research Group, at the time of the meetings of the American Academy of Neurology and the American Society of Human Genetics. With the advent of the Special Interest Groups of the AAN, including a group in neurogenetics, we have cooperated with these groups to help organize courses and symposia at the AAN, which attracts an international audience.

We have also organized a number of stand-alone international symposia on various topics in neurogenetics, including tuberous sclerosis, alternating hemiplegia, neuro-acanthocytosis, polymicrogyria and, most recently, progressive myoclonus epilepsies.

Objectives:

We would now like to extend these activities, as follows:

1. Collaborate with the other Research Groups devoted to specific neurological disorders, such as ataxias, ALS, etc to organize independent educational programs and international symposia on specific neurogenetic topics under the aegis of the WFN.
2. Organize neurogenetic teaching activities as satellites or integrated into the educational programs of a number of national and international neurological meetings.
3. Emphasize the teaching of neurogenetics in developing regions. In these regions, specific neurogenetic diseases occurring with high prevalence may be due to high consanguinity rates and population structure. Ideally, the teaching should be done in the region, with participation of local neurologists, but first it may be necessary to support the attendance of individuals from these regions at various national and international meetings where these educational programs are given. These individuals would then collaborate in organizing the courses in the region. We would also collaborate with the Latin American, African and Asian Initiatives to organize these courses.