

Welcome to *Neurology: Genetics*

OPEN

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The powers of human genetics and genetic technologies have transformed the complexities of neurology and neuroscience at the basic, translational, and now also the clinical level. We have left an era of black and white views of causative genetic variation and are entering a period of more than 50 shades of grey, fascinated with DNA variants that increase or decrease risk, epigenetic modification, and an unexpectedly large number of variants of unknown or potentially pathogenic significance. Loss-of-function alleles and even complete human gene knockouts for certain genes appear to be compatible with a normal phenotype.

At the same time, we are witnessing a continued increase in mendelian disorders. In the last 8 years, the number of phenotypes caused by mendelian alleles more than doubled in OMIM, a database of Online Mendelian Inheritance of Man. Next-generation sequencing of the human genome has revealed an abundance of rare, almost private, genetic variants, including copy number variations and a significant amount of de

novo changes, when parents and offspring were analyzed. In light of these developments, the remarkable increase in published papers referencing genetics and the nervous system over the last decades is not surprising (figure).

The mission of *Neurology® Genetics* is the publication of high-quality original contributions that elucidate the role of genetic and epigenetic variation in diseases of the central and peripheral nervous systems. The journal will publish original articles in all areas of neurogenetics, including rare and common genetic variation, genotype-phenotype correlations, outlier phenotypes as a result of mutations in known disease genes, and genetic variations with a putative link to diseases. This will include studies reporting on genetic disease risk and pharmacogenomics. In addition, the journal will publish results of gene-based clinical trials, for example, those targeting disease genes using viral vectors or antisense oligonucleotides. Genetically engineered model systems will not be a primary focus of *Neurology: Genetics*, but preclinical studies using model systems for treatment trials are welcome, including well-powered studies reporting negative results. Each issue will also contain invited review articles, editorial commentaries, and reader correspondence.

Our vision is to become the premiere journal for human neurogenetics written and read by neurologists and geneticists. At the same time we want to provide a forum for discussion by clinicians and scientists of matters as diverse as novel genetic technologies and ethics of genetic testing. *Neurology: Genetics* is a new journal and follows the trend of open access and rapid publication, but it is firmly enveloped into the mantle of the traditional parent journal *Neurology* and a highly reputable publishing company.

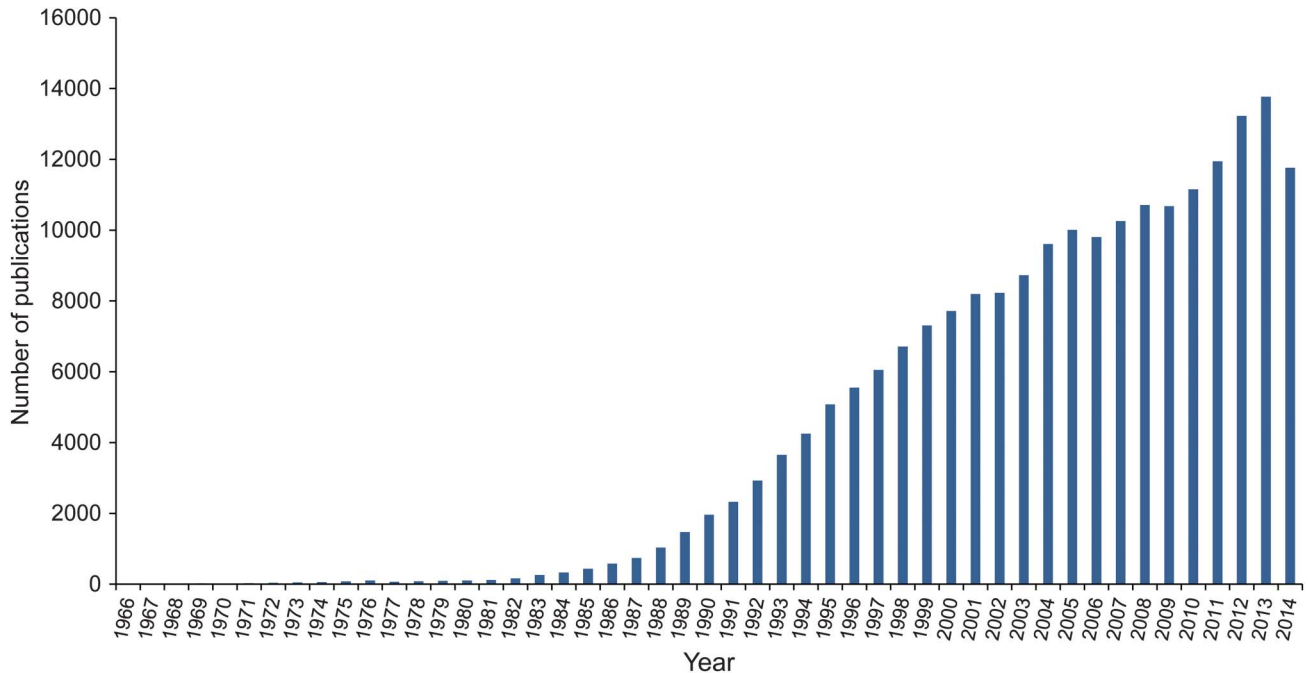
In essence, *Neurology: Genetics* will deal with variations in genotypes and phenotypes and their relationships. Most papers will try to extend a correlation between the two to a causative link. Establishing this causative link between neurologic diseases and genotypes will require diligent analysis by clinicians and scientists as well as a nuanced approach to peer review using common sense and seasoned judgment to

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Figure Increase of publications relating to genetics and neurology



Pubmed was searched for the following advanced search term: (gene[Text Word]) OR mutation[Text Word] AND (brain[Text Word] OR CNS[Text Word] OR nerve[Text Word] OR muscle[Text Word] OR neurology[Text Word]). Data for 2014 are not yet complete. Available at: <http://www.ncbi.nlm.nih.gov/pubmed/>. Accessed March 13, 2015.

determine whether DNA variants are associated with the observed clinical manifestations. One can already predict that this issue will be the topic of many editorials to come.

The group of associate editors is as diverse as genetic research is today in age, geographic distribution, and areas of expertise. Nicholas Johnson is an Assistant Professor of Neurology at the University of Utah focusing on muscle diseases and patient-centered outcomes. Raymond P. Roos is the Marjorie and Robert E. Straus Professor in Neurological Science in the Department of Neurology at the University of Chicago and an expert in motor neuron disease, prion disease, and neurovirology/neuroimmunology. Jeffery Vance is Professor of Genetics at the University of Miami. His expertise is in Parkinson disease and Alzheimer disease as well as inherited neuropathies. Two of our associate editors are located in Europe. Alexandra Dürr is Professor of Neurology at Université Pierre et Marie Curie, Paris. Her fields of study are spastic paraplegia, Huntington disease, and ataxia. Massimo Pandolfo is Professor of Neurology at the Université Libre de Bruxelles with interests in epilepsy, Friedreich ataxia, and stem cells. All of our associate editors are well published in other areas of neurology, neurogenetics, and neuroscience as well.

Every one of our patients has 6 billion base pairs, give or take a few, plus a mitochondrial genome. Their analysis is waiting for us. As Cole Porter wrote for his famous song in 1928, “Let’s do it.”

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DISCLOSURE

Stefan M. Pulst has served on the editorial boards of *Journal of Cerebellum, NeuroMolecular Medicine, Continuum, Experimental Neurology, Neurogenetics*, and *Nature Clinical Practice Neurology* and as Editor-in-Chief of *Current Genomics*. Dr. Pulst conducts research supported by the NIH, Target ALS, and the National Ataxia Foundation. He has consulted for Ataxion Therapeutics, has received research funding from ISIS Pharmaceuticals, has served on a speakers’ bureau for Athena Diagnostics, Inc., and is a stockholder of Progenitor Life Sciences. He has received license fee payments from Cedars-Sinai Medical Center, and has given expert testimony for Hall & Evans, LLC. Dr. Pulst has received publishing royalties from Churchill Livingstone (*The Ataxias*), AAN Press (*Genetics in Neurology and Molecular Genetic Testing in Neurology*, 2nd–5th editions), Academic Press (*Genetics of Movement Disorders*), and Oxford University Press (*Neurogenetics*). Dr. Pulst holds patents for Nucleic acid encoding ataxin-2 binding proteins, Nucleic acid encoding Schwannomin-binding proteins and products related thereto, Transgenic mouse expressing a polynucleotide encoding a human ataxin-2 polypeptide, Methods of detecting spinocerebellar ataxia-2 nucleic acids, Nucleic acid encoding spinocerebellar ataxia-2 and products related thereto, Schwannomin-binding proteins, and Compositions and methods for spinocerebellar ataxia. He receives an honorarium from the AAN as the Editor of *Neurology: Genetics*. Go to Neurology.org/ng for full disclosure forms.

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