

International Conference and Expo on Audiology and Hearing Devices

August 17-18, 2015 Birmingham, UK



Peter L Nagy

Columbia University, USA

The role of clinical genomic testing in diagnosis and discovery of pathogenic mutations

Next-generation sequencing in clinical practice allows for a critical review of the literature to evaluate disease relatedness of specific genes and pathogenicity of individual mutations, while providing an important discovery tool for new disease genes and disease-causing mutations. Data obtained from large panels, whole exome or whole genome sequencing, performed for constitutional or cancer cases, need to be managed in a transparent, yet powerful analytical framework. Assessment of reported pathogenic potential of a variant or disease association of a gene requires careful consideration of population allele frequency, variant data from parents, and precise, yet concise phenotypic description of the entire family and other individuals or families that have the same variant. The full potential for discovery can only be realized if there is data sharing between clinicians performing the interpretation worldwide and structural biologists, analytical chemists and cell biologists interested and knowledgeable of the structure and function of the genes involved.

Biography

Peter L Nagy has received his MD degree from the University of Pecs, Hungary in 1989. He has obtained his PhD at Purdue University in Biochemistry under the mentorship of Dr. Howard Zalkin and his Anatomic and Molecular Genetic Pathology training at Stanford University working on the MLL gene with Michael Cleary and Roger Kornberg. His research is on neurodegenerative disorders like ALS and young adult onset ataxias (AOA2). He built and oversees the clinical next-generation sequencing facility in the Laboratory of Personalized Genomic Medicine at Columbia University Medical Center.

pln2104@cumc.columbia.edu

Notes: