

International Conference and Expo on **Audiology and Hearing Devices**

August 17-18, 2015 Birmingham, UK

Clinical next-generation sequencing for constitutional hearing disorders

Peter L Nagy

Columbia University, USA

Our Laboratory of Personalized Genomic Medicine (LPGM) at Columbia University Medical Center started to offer clinical whole exome sequencing (WES) in January 2013. We processed and issued reports on over 500 constitutional cases including approximately 70 cases involving hearing loss. Next-generation sequencing in the clinical practice allows for a critical review of the literature describing the pathogenicity of specific mutations or the disease relatedness of specific genes and also provides an important discovery tool for new disease genes and disease causing mutations. Because of the large volume and complex nature of the data obtained from large panels and whole exome sequencing testing, the management of the data in a transparent, yet powerful analytical framework is the key to a successful clinical operation. We have provided diagnosis for constitutional patients in about one third of the cases we analyzed. The full potential for discovery of new disease associated genes and disease causing mutations can only be realized if there is a tight collaborative effort between the clinicians performing the interpretation and structural biologists and analytical chemists and cell biologists who can help predict and verify the effects of variants identified.

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: