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Clinical Next-generation sequencing for constitutional disorders and cancer management

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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 cases, mostly trios. The majority of these samples are from the pediatric population (>80%). The most common clinical scenario is a child with developmental or intellectual delay with or without dysmorphic features with clinically unaffected parents. In almost every case, patients have had extensive genetic workup prior to WES including biochemical, mitochondrial, single-gene testing, targeted gene panel, traditional karyotyping and microarray chromosome analysis. Of the cases analyzed to date we have identified pathogenic or probable pathogenic mutations responsible for the patients' condition in about 30 percent of the cases. Continued improvement and automation of the analytical pipeline allowed for identification of novel connections between genes and disease in several constitutional genetic disorders. Through my presentation the audience will obtain an understanding of the current state of the art of clinical genomic testing; they will become familiar with the major factors that determine the precision and sensitivity of pathogenic mutation detection; have a thorough understanding of the importance of proper implementation of structural and functional basic science data sources into the clinical analysis pipeline. I will outline the contribution of clinical data collection to discoveries in basic science and review the obstacles to and opportunities for more efficient collaboration between clinical medical centers and the pharmaceutical industry.

Biography

Peter L Nagy received his MD degree from the University of Pecs, Hungary in 1989. He 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.

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