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Optimizing sensitivity and specificity of clinical next generation sequencing in constitutional and cancer applications

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 and clinical cancer whole exome/transcriptome (CWES) sequencing since January 2014. We processed and issued reports on over 500 constitutional and about 100 cancer cases. The majority of these samples are from the pediatric population (>80%) both for constitutional and cancer testing. 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 and changed clinical management of pediatric malignancies in about 20 percent of our patients. The fact that a large percentage of cases remain without molecular diagnosis or useful clinical treatment recommendation indicates that we need to improve our assignment of pathogenic effect to mutations in genes not previously linked to disease. We have developed a database we refer to as "SNP-catcher" that integrates patient information with a molecular systems approach to evaluate the significance of mutations. Such systems driven analysis of clinical exome and transcriptome sequencing data is an important step in the accelerated discovery of novel disease causing genes and disease mechanisms and obtaining useful treatment recommendations.

Through my presentation the audience will obtain an understanding of the current state of the art of clinical genomic testing; 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. His interest to pursue a career as a physician scientist led him to Purdue University where he earned his PhD in Biochemistry. He worked under the mentorship of Dr. Howard Zalkin and made important discoveries relating to C1-metabilism in bacteria. Subsequently he completed Anatomic and Molecular Genetic Pathology training and Stanford University as well as postdoctoral training in Michael Cleary's laboratory. He was the first to purify and functionally characterize the Set1 histone methyltransferase complex from *S. cerevisiae* in collaboration with Dr. Roger Kornberg. He co-developed the FAIRE method with Jason Lieb allowing physical fractionation of chromatin based on formaldehyde crosslink ability. Currently he leads a research laboratory investigating the role of transcriptional defects in neurodegenerative diseases, such as AOA2 and ALS4, and is director of the clinical next-generation sequencing facility in the Laboratory of Personalized Genomic Medicine at Columbia University Medical Center in the Department of Pathology and Cell Biology.

pln2104@cumc.columbia.edu

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