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Integration of next-generation sequencing data with analytical chemistry, structural biology and cell biology

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 a key to a successful clinical operation. Population allele frequency, data from parents and precise, yet concise phenotypic description are the cornerstone for successful clinical evaluation of the pathogenicity of variants identified. 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. My presentation will focus on the need to foster and strengthen this multidirectional information flow. I will review the resources that are already available and propose ways to improve them through integrating new data types or design of more user-friendly interfaces.

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-metabolism 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 methyl transferase 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 crosslinkability. 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.

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