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Practical approaches for increased specificity and sensitivity of clinical next generation sequencing testing

Peter Lajos 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 cases mostly trios. 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 successful clinical operation. Population allele frequency, data from parents and precise, yet concise phenotypic description are the corner-stone 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. 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.

pln2104@cumc.columbia.edu

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