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A framework for translating advances in molecular genetic technologies into clinical laboratory practice

Malgorzata Jaremko and Alexander Bisignano
Phosphorus Diagnostics, USA

Advances of the molecular diagnostic testing platforms, including development and implementation of NGS based genetic testing contribute to the improvement of disease prediction, diagnosis, and treatment. However, the future of genomic medicine relies on the capability of molecular genetics laboratories to develop and validate evidence-based and cost-effective laboratory tests. These laboratories face many challenges including establishing clinical utility, validating analytical performance of laboratory developed tests, and managing costs of platform development and subsequent consumables. Along with the molecular and instrumentation challenges, laboratories are faced with a myriad of software options (e.g., Galaxy, Amazon, GATK, BaseSpace, and Clarity) when establishing a reliable bioinformatics pipeline and LIS system. Finally, there is a lack of consensus and consistency in the quality standards across the industry (e.g., read depth, variant curation, and clinical validation structure). In this study, we present a framework for the consistent development of accurate, high-quality, NGS diagnostic tests. Our process is broken into stages from gene selection through clinical validation and implementation. Based on the experience in our own CLIA-laboratory, we present lessons learned in the development of NGS targeted panels for sequencing and CNV analysis for various indications including infertility, hereditary cancers, arrhythmias, cardiomyopathies and lipidemias.

Biography

Malgorzata Jaremko has completed her PhD in Pharmacogenomics from Wroclaw Medical University and Postdoctoral Clinical Fellowship from Mount Sinai School of Medicine, NY. She is board certified by American Board of Medical Genetics and Genomics in Clinical Molecular and Clinical Biochemical Genetics; and she is Fellow of American College of Medical Genetics and Genomics, as well as National Academy of Biochemistry. She has extensive experience in directing clinical molecular laboratories, and currently serves as the Senior Director, Clinical Laboratory & Molecular Diagnostics, and CLIA-Director of Phosphorus Diagnostics genetic testing laboratory.

m.jaremko@gmail.com

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