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Next generation sequencing in clinical diagnostic laboratories: Improvement on patient's quality of care

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Abstract

Advances in genomic technology have made sequencing of the entire human genome possible. The next generation sequencing (NGS) technology has been widely used in research laboratories. The ability to sequence a group of target genes by NGS is particularly useful for molecular diagnosis of genetically heterogeneous clinical syndromes while reducing cost and time. However its application to molecular diagnosis in clinical laboratories is still in its infancy due to the lack of commonly recognized guidelines. A group of complex diseases are more challenging to make accurate molecular diagnosis by conventional methods, like mitochondrial disorders, that can be caused by mutations in both nuclear and mitochondrial genomes. To develop a comprehensive NGS based assay for molecular diagnosis of clinically and genetically heterogeneous disorders in a clinical setting requires the implementation of proper quantitative and qualitative controls. The inclusion of control procedures and samples assure the highest quality performance required in a clinical laboratory. Besides quality assurance program, examples will be presented to show the use of pathway-focused gene panels and whole exome sequencing can help to illustrate the positive impact that NGS testing and improve the diagnostic yield while reducing cost and time needed to reach a molecular diagnosis.

Biography

Dr. Victor Zhang has developed and implemented numerous NGS based for molecular diagnosis of heterogeneous disorders. He obtained his Ph.D from Rice University and did postdoctoral studies from Baylor College of Medicine. He completed his clinical fellowship from Emory University School of Medicine. He is currently assistant director of Mitochondrial Diagnostic Laboratory at Baylor College of Medicine, a premier reference testing service center in US. He has published more than 20 papers in reputed journals and been invited to give multiple presentations on these topics nationally.