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Next-generation sequencing in personalized medicine

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The development of next-generation sequencing (NGS) technologies has made sequencing not only rapid and cost-effective, but also highly accurate and reproducible. NGS can be applied to personalized medicine through whole-genome sequencing (WGS) to detect variations across the entire genome, whole-exome sequencing (WES) to identify variants in the coding regions, whole-transcriptome sequencing, and targeted sequencing of specific gene panels and/or well-defined pathways. These advances have increased the utility of NGS in clinical settings, with applications ranging from the identification of rare diseases to the detection of chromosomal abnormalities in maternal-fetal medicine. Deep sequencing and circulating free tumor DNA in the oncology space is also trending toward clinical utility. As NGS becomes widely adopted, there are challenges to overcome, such as consensus on medically relevant genomic variants, burden on patients and clinicians of reporting, intervening, and follow-up of genomic findings, including incidental findings. Regulatory clearance of NGS-based platforms and approval of clinical assays utilizing NGS technologies, are also central to the continued implementation of NGS in personalized medicine.

Biography

Frank S Ong, MD is currently the Associate Director of Medical Affairs at Illumina, Inc. He graduated with honors from the Keck School of Medicine at the University of Southern California. He completed a research fellowship in genetics of blood pressure regulation in the Division of Experimental Pathology at Cedars-Sinai Medical Center and a clinical fellowship in Clinical Molecular Genetics in the Department of Pathology and Laboratory Medicine of UCLA. He has garnered numerous awards for his research in hypertension, including American Heart Association (AHA) Kidney Council New Investigator Award for High Blood Pressure Research and many more He has served as a peer reviewer for *Hypertension, Journal of the American College of Cardiology*, *The Pharmacogenomics Journal*, *Pharmacogenetics and Genomics*, and PLoS ONE. His interest in genetics and personalized medicine has led to active participation in several population genetics consortia, including NHLBI GO Exome Sequencing Project (ESP), CHARGE, and MESA, focusing on the genetics of blood pressure regulation, personal genomics and pharmacogenetics.

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