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Next generation sequencing in personalized medicine: Advances, translational impact, and future directions

Alireza Haghighi
Harvard Medical School, USA

Personalized medicine is a rapidly evolving health-care, in which individual's molecular/genetic profile is used to customize health management. Next Generation Sequencing (NGS) is an innovative approach that has revolutionized genetic studies. This approach is transforming medicine by providing individual's genetic profile that guides decisions made in regard to the prevention, diagnosis, and treatment of disease; this both maximizes health benefits and minimizes the risk of disease. This diagnostic and prognostic information had previously only been available in multiple analyses. NGS parallelizes the sequencing process, producing thousands or millions of sequences concurrently at high speed and low cost. NGS includes different methods such as whole genome sequencing, whole exome sequencing and targeted sequencing. NGS has led to an explosion of discoveries of causes of genetic diseases, new underlying pathways, and identification of novel mutations that has extensively expanded genotype-phenotype relationships. To benefit from the genomic medicine in improving patient care, clinicians need to be trained to understand the inheritance pattern of diseases, how to confirm the clinical diagnosis with genetic tests and potential novel gene based therapies. There are still many challenges ahead but looking to future, the great power of genomic techniques will allow molecular diagnosis to become a standard investigation in patient diagnosis and therapeutic interventions. The examples of barriers for translating NGS results for routine clinical use include management and interpretation of an enormous amount of information produced by these techniques and the identification of disease etiologies for genetic conditions with substantial genetic heterogeneity, and ethical issues related to this novel method.

Biography

Alireza Haghighi MD, DPhil graduated from University of Oxford and is currently based at the Department of Genetics of Harvard Medical School and Brigham and Women's Hospital. His research focuses on studying inherited disorders, and carrying out disease gene discovery projects and genetic association studies, using state-of-the-art genetic and genomic approaches such as the various microarray-based platforms and next-generation sequencing. He is also working on translating basic research discoveries into better diagnostics and improved management strategies.

haghighimd@yahoo.com