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Next generation sequencing: New hope for patients with genetic disorders

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Abstract

Genetic diseases are caused by DNA alterations of the nucleotide composition within the genome that perturb or abolish the function of a gene. Many of genetic diseases are locus-heterogeneous as mutations in any one of many genes can cause similar clinical phenotypes. Moreover, these diseases can be clinically heterogeneous with variable penetrance, severity and age of onset standard approach for identifying the causation in genetic diseases included techniques such as positional cloning and linkage analysis followed by targeted candidate gene sequencing. Many cases remained unsolved (and undiagnosed) due to limitations of the approach.

Next generation sequencing (NGS) has provided us with the ability to investigate the genetic cause of diseases with unknown etiopathology. Application of this state of the art technology has resulted in exciting breakthroughs in genetic diseases. NGS enables us to discover the rare causative variants that cannot be detected by the standard SNP arrays. As the cost of NGS is tumbling, the application of this approach is now very cost-effective, and indeed cheaper than the classical approach. Whole exome sequencing can now be performed in a few days and at a cost of less than $1000. This revolutionary method offers the advantage of directly analyzing the genome for genetic variants thereby combining previous mapping and sequencing efforts. In addition, other genes of interest can be analyzed for variants that might cause the disease or influence its outcome. An important advantage of this method is providing the possibility of identifying mutations in an unknown/ unexpected gene in very small family pedigrees, or even one affected individual, which was not possible with candidate-gene approach. Moreover, this approach can help us with the diagnosis of unknown conditions or heterogeneous known diseases with unusual manifestations. We hope that current increasing progress in investigation of genetic diseases, using NGS, will improve our insight into the pathophysiology of these diseases and will hopefully lead to development of novel, more effective and affordable diagnostic and treatment strategies, and personalized genomic medicine as the ultimate result.

Biography

Alireza Haghighi, MD, DPhil is a clinician scientist. He graduated from University of Oxford and is currently based at the Department of Genetics of Harvard Medical School and Brigham and Women's Hospital. Dr. Haghighi's research focuses on clinical and genetic investigation of inherited diseases, using state of the art technologies such as high throughput genotyping and next generation sequencing. He is also working on translating basic research discoveries into better diagnostics and improved management strategies, and opportunities for developing novel therapeutics to treat disease.

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