How genetics transforms our clinical practice

Personalized Medicine can be described as making correct diagnosis and providing the right patient with the right drug at the right dose at the right time. Rapidly evolving advances in genetic technologies, including Next-Generation sequencing (NGS), has opened up a revolutionary era in genetics of human diseases and is transforming medicine into precise, personalized health-care. NGS or “High throughput sequencing” includes different technologies of massively parallel sequencing and microarray analysis that determine the precise order of nucleotides within a DNA molecule in a few hours. Whole exome sequencing technologies have helped us discover the genetic bases of many monogenic diseases and identify novel pathways involved in the pathogenesis and progression of complex disease. Using targeted sequencing, we are now able to rapidly test a large set of candidate genes (for a disease) in a single test, which is a faster and much more efficient substitute for the conventional gene-by-gene approaches. The continuing decrease in costs and run times is expanding NGS into routine clinical practice. Yet the major challenges are not in the technology itself, but in the big amount of data generated by these methods that identify millions of variants among of which one or two may be the cause of a disease; a shift from the identification to the interpretation phase. NGS tools will very soon be adopted by physicians who are consulted to assist in providing a diagnosis for patients with complex clinical manifestations, particularly in cases where genetic contributions are suspected but have proven difficult to identify. Pediatricians are in a unique position to benefit from these advancements in genetics and further promote this approach. NGS can assist them to tailor the treatment of patients to alterations in their genome. These technologies also enable clinicians to re-classify diseases based on genome sequence and subsequently, take a targeted therapeutic approach to optimise treatment outcome. There are, however, challenges that need to be overcome, which include the identification of clinically relevant causative genomic variations across the whole genome and the ability to rapidly and easily analyze and interpret the vast quantity of data produced by genomic techniques.

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. His 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.

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