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Bioinformatic approaches to assess the genetic basis of Neurological Disease

Next generation sequencing is facilitating the rapid and cost effective surveilance of human genomes in order to identify variants of pathological consequence. Inherited neurological disorders represent one key area that have strong potential to benefit from such a co-ordinated genetic interrogation. Diagnostic confirmation of the presence of deleterious DNA changes could lead to effective personalised management protocols for the patient and also aid in informed decision making with respect to family planning for parents of affected children. New potential therapeutic targets may be elicited if the gene or its associated physiological pathways could be modulated by pharamacological intervention, thus amelorating the deleterious effect of the variant change.

Bioinformatics is a broad term for a set of compuational tools that facilitate variant identification and classification (e.g. benign or pathogenic). In combination with adequate clinical phenotyping data, the genetic locus/loci responsible for the disorder can be identified. This lecture provides an introduction to some of the computational approaches that can be adopted by clinical exome sequencing teams and how this information can be exploited to assist in immediate and long-term clinical management protocols.

Speaker Biography

Shahid Mian is a Consultant in Clinical Research at King Fahad Medical City (KFMC), Saudi Arabia. He is responsible for both the development and implementation of bioinformatic pipelines that are applied to genomic sequence analyses. Dr Mian has computationally processed over 800 patient exomes and is a member of the clinical reporting team within the CAP (College of American Pathologists) accredited Pathology and Clinical Laboratory Medicine at KFMC. This team is responsible for supporting physicians in the management of patients with inherited genetic disorders including those with neurological impacting disorders.

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