Clinical exome sequencing of patients from a highly consanguineous population: Novel pathogenic variants impacting neurological function

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Saudi Arabia has a highly consanguineous population with specific geographical regions estimated to have rates exceeding greater than 80%. The downstream effect of such population dynamics is to significantly enrich the frequency at which recessive pathogenic variants occur and consequently their associated Mendelian disorders. This is evident at both a community and a family level. King Fahad Medical City (KFMC) is a tertiary care facility that diagnoses patients with inherited disorders through exome sequence analysis of germline DNA. Many of the biological pathways negatively impacted by these pathogenic changes manifest at a neurological level. These include for example intellectual disability, ataxia, epilepsy and white matter structural changes. The Department of Pathology and Clinical Laboratory Medicine (PCLM) has sequenced the exomes of over 1100 patients. Novel pathogenic variants in genes biologically and clinically linked to specific neurological conditions have been identified. Furthermore the PCLM knowledge base has also implicated novel genes with no known function, to a variety of neurological conditions. Evidence is presented at how genetic analysis of exome sequence data derived from patients orginating within highly consanguineous populations can lead to the identification of novel genes/genetic variants linked to neurological physiology.

Biography

Shahid Mian is a Consultant Clinical Research Scientist, PhD within the Department of Pathology and Clinical Laboratory Medicine (PCLM) at King Fahad Medical City (KFMC), Saudi Arabia. PCLM is a College of American Pathologists (CAP) accredited laboratory. He has responsibility for establishing bioinformatic and variant reporting pipelines for the clinical exome analysis of paediatric patients with suspected inherited disorders. He has reviewed over 500 clinical exome reports produced by third party laboratories for PCLM, independently reported over 100 patient exome results to KFMC physicians and analysed over 1300 exomes bioinformatically.

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