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Bioinformatic approaches to assess the genetic basis of Neurological Disease

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Introduction:

Neurological disorders include a good spectrum of diseases within the central systema nervosum (CNS). Up till now, many neurological disorders are classified, with symptoms varying from cognitive dysfunction to manic behavior or depression. thanks to the complex nature of this group of diseases, it's difficult to spot the mechanisms using conventional methodologies, where only small pathways around specific target genes are investigated. the arrival of systems biology approaches has made it possible to review these complex problems from the whole-genome perspective. Within the recent years, genomic technologies are increasingly applied to the investigation of neurological disorders.

Alzheimer's disease (AD) may be a major sort of neurodegenerative diseases. AD starts from amnesia and cognitive deficit within the early stage and gradually evolves into severe dementia within the late stage. The pathological hallmarks of AD include extracellular deposit of amyloid plaques and intra-neuronal neurofibrillary tangles (NFT). Although the disease-causing mutations are identified for the familial early-onset AD (FEOAD), the genetic landscape has been perplexing for the late-onset AD (LOAD) that constitutes ~95% of all AD patients [4]. The prevailing hypothesis for the disease mechanism of AD has been based on the studies of FEOAD, which advocates the central role of amyloid-β (Aβ) within the chain of events resulting in neuronal death and cognitive and behavioral symptoms. However, Aβ-based interventions haven't been successful within the clinical trials thus far. Thanks to the shortage of effective treatment for curing or slowing down AD, it becomes imperative to look for novel risk genes and drug targets, also as biomarkers for early diagnosis.

Objective:

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 profit from such a co-ordinated genetic interrogation. Diagnostic confirmation of the presence of deleterious DNA changes could lead on to effective personalised management protocols for the patient and also aid in informed deciding with reference to family planning for parents of affected children. New potential therapeutic targets could also be elicited if the gene or its associated physiological pathways might be modulated by pharamacological intervention, thus amelorating the deleterious effect of the variant change. Bioinformatics may be a broad term for a group 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.

Neurological disorders comprise a spread of complex diseases within the central systema nervosum , which may be roughly classified as neurodegenerative diseases and psychiatric disorders. The basic and translational research of neurological disorders has been hindered by the problem in accessing the pathological center (i.e., the brain) in live patients. The fast headway of sequencing and exhibit advances has made it conceivable to explore the illness instrument and biomarkers from a frameworks point of view. In this audit, ongoing advances inside the revelation of novel hazard qualities, treatment targets and fringe biomarkers utilizing genomic advances will be examined. Our major focus are going to be on two of the foremost heavily investigated neurological

disorders, namely Alzheimer's disease and autism spectrum disorder.

Autism spectrum disorder is a neurodevelopmental disorder characterized by social and communication deficit as well as stereotyped and repetitive behaviors [6]. According to a recent survey, 1 in 68 US children has ASD. In contrast to AD, the disease onset for ASD starts from 3 years aged to infancy . The gender ratio is approximately 4:1 disfavoring boys. Like other psychiatric disorders, there are not any clear pathological hallmarks for ASD [1]. It is believed that brain wiring is altered in ASD children, although the precise interplay between gene and environment has not been clarified. In terms of the genetic factors, some sorts of ASD could also be caused by rare mutations, while others could also be thanks to the mixture of common variations [7]. The genetic alterations in ASD are also more complex than those in AD, which include copy number variation, insertion, deletion and single nucleotide polymorphism (SNP). In addition to the genetic and environmental factors, prenatal and perinatal factors may also contribute to the development of ASD.

Genomic investigations of neurological issue include the examination of the genome, transcriptome and epigenome. There are two sorts of innovations accessible for genomic examines, including sequencing and different exhibit stages. For the examination of genomic variety, the examples for the most part originate from fringe blood, in spite of the fact that salivation has likewise been utilized.For the investigation of transcriptome, brain tissue is that the most studied since it's more relevant to the disease mechanism. The peripheral blood and spinal fluid (CSF) have also been investigated, mostly for the invention of novel biomarkers. These three tissues have also been utilized within the investigation of epigenomic alteration. additionally, skin fibroblast has been increasingly utilized in induced pluripotent somatic cell

Conclusion:

Genomic investigations of neurological issue include the examination of the genome, transcriptome and epigenome. There are two sorts of innovations accessible for genomic examines, including sequencing and different exhibit stages. For the examination of genomic variety, the examples for the most part originate from fringe blood, in spite of the fact that salivation has likewise been utilized.For the investigation of transcriptome, brain tissue is that the most studied since it's more relevant to the disease mechanism. The peripheral blood and spinal fluid (CSF) have also been investigated, mostly for the invention of novel biomarkers. These three tissues have also been utilized within the investigation of epigenomic alteration. Additionally, skin fibroblast has been increasingly utilized in induced pluripotent somatic cell

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.