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Abstract Topic:- Molecular effects of genetic variation

Abstract Title:- Mission Program on Pediatric Rare Genetic Disorders (PRaGeD)

Presenting author name :- Gowri Rao

Presenting author institute:- Centre for DNA Fingerprinting and Diagnostics

Co-authors name:- Neeraja Chilukoti, Ashwin Dalal

Co-authors institute:-Centre for DNA Fingerprinting and Diagnostics

Aims:-India's large population, practice of consanguinity and biological isolation of some endogamous population groups have resulted in a relatively higher prevalence of rare genetic disorders.NGS-based analysis like Whole exome sequencing and Whole genome sequencing have proven to be crucial in identifying novel genes and novel variants in known genes. Mission program on Pediatric Rare Genetic Disorders (PRaGeD) is a PAN-India initiative involving 16 centres funded by the Department of Biotechnology (DBT), Ministry of Science and technology, Government of India. The aim of the initiative is to diagnose, conduct research and provide genetic counselling for "Pediatric rare genetic disorders".

Methods:- Patients with undiagnosed genetic diseases are recruited through telemedicine with a network of medical colleges (Pediatric Departments), DBT-UMMID centres, and collaborating centres across the country. WES and WGS based analysis of patients and/or parent-offspring trios is performed using bioinformatics pipeline. Database for PRaGeD is developed comprising of variant details identified from the genome analysis. Novel variants identified from the genome analysis are further being characterized for their functional roles using cell lines or model organisms such as mice, drosophila or zebrafish. Awareness programs in the form of workshops, community awareness programs, visit to primary health centres, colleges, schools are being conducted by each centre. Infographics and videos are developed and circulated to create awareness among the people.

Results:- A total of 619 samples from 234 families with unexplained genetic conditions were recruited by collaborating centres and sent to CDFD, Hyderabad for Whole Exome Sequencing (WES) or Whole Genome Sequencing (WGS). WES of 130 cases is completed at CDFD, Hyderabad, 14 cases are solved and data analysis is in progress for other cases. Sequencing is in process or data is awaited for 83 cases. Ten families were recruited for Whole Genome Sequencing (WGS).

Conclusions:- Mission program is a large-scale project and is one of a kind in India with the potential to foster more projects on rare genetic disorders, and create a valuable database of genes and variants for the Indian population. WES/WGS tests through Mission program can be instrumental in aiding the lives of low-income and below-poverty-line individuals with these conditions. Mission program embarks to discover and characterize new genes/variations and achieve genetic diagnoses, with the aim of developing new therapies that can make a significant difference in the lives of children and their families with rare genetic conditions.

Keywords:- Pediatric disorders, Exome, Genome, NGS, Genetic variant, PRaGeD