Presentation title: **Role of Variant of uncertain significance in genomics era**

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Abstract:

In clinical genetic testing, variants of unclear significance (VUSs) are common that generate dissatisfaction for laboratories, patients, and clinicians alike. The uncertainty surrounding VUSs makes diagnosis and clinical care more difficult. To direct efforts to reduce uncertainty, a thorough evaluation of VUSs across numerous disease genes is required.

According to american college of medical genetics and genomics and the association for molecular pathology, variants are classified as benign, likely benign, pathogenic, likely pathogenic, or variant of uncertain significance for the identification of many rare, novel DNA variants in diverse populations. If the variant is classified as a VUS, it indicates that there was insufficient information available at the time of interpretation to establish whether the variant is associated with disease. A VUS should not be utilized in clinical decision-making, as per the ACMG guidelines. When a patient is found to have a VUS, all clinical judgments should be made based on the patient's medical history, including their family history, rather than the VUS itself.

Regularly review of published literature, new development of any signs or symptoms and parents, affected and unaffected family member genetic testing could aid in the reclassification of VUSs. Together with clinical geneticist and genetic counsellor who plays a vital role in Insilco analysis of the clinical data, variant filtration-analysis, genotypic phenotypic correlation, and other evidence helps to assess if a variant is linked to disease or not. With the help of genetic counseling, they can also help in confirming the diagnosis, risk calculation, provide prognoses, improve treatment plans, and address hazards for family members by reclassifying variations as pathogenic, likely pathogenic, benign or remains VUS.

Keywords- VUS, Genetic Counsellor, Geneticist.

Biography:

She is an expert Genetic Counsellor with more than 3 years of experience. A Certified Genetic Counsellor by DBT-Health care sector skill council of India who has shadowed 2500+ Clinical Cases on the module of genetic counselling. She is Member of ASGC (Arab Society of Genetic Counsellor) since January 2023.

She has also Completed Genomic Variant Analysis and Clinical Interpretation Workshop, Conducted by IGIB-Genomics for Understanding Rare disease India Alliance Network (GUaRDIAN) and PMRF Course on Genome Editing: Introduction to CRISPR- Cas (Module PMRF-ISSS013) conducted by Institute of Smart structures and system (Society reg. by Govt. of Karnataka) and Indian Institute of science, Bengaluru, India. She has also completed her master's in molecular and human Genetics from Jiwaji University. She has completed her bachelor's in biotechnology.

She has reviewed scientific paper, published case reports and presented abstracts and poster in more than 10 scientific journals in both National and International conferences in the area of Human Genetics.