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Abstract Topic:- Clinical Genetics

Abstract Title:- Reanalysis of Exome Sequencing: Fresh Insights into Unsolved Rare Diseases

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Aims:-With the rapid evolution of bioinformatics and genomics, this study seeks to revisit exome sequencing data of patients with undiagnosed rare diseases. The goal is to potentially uncover causative variants that might have been missed or misinterpreted during initial assessments due to the limited genomic knowledge of the time.

Methods:- Between 2020 and 2023, exome datasets of 45 probands with previously unresolved exome outcomes were re-evaluated. Initial findings ranged from insignificant variants to those of uncertain significance across various genes. An updated in-house exome analysis pipeline was utilized for this re-examination, and variants' clinical relevance was assessed using contemporary databases, tools, and prediction algorithms.

Results:- The reassessment yielded 8 new pathogenic or likely pathogenic variants distributed across 8 genes. As a direct outcome, these findings enabled definitive diagnoses for 8 patients, paving the way for enhanced medical management, tailored therapeutic strategies, and insightful genetic counseling.

Conclusions:- The dynamic progression of genomics underscores the importance of consistent data re-evaluation. Such periodic re-analyses are crucial to ensure comprehensive and up-to-date diagnostics for rare genetic disorders.

Keywords:- Genetic Diagnosis, Undiagnosed diseases, Exome Re-analysis, Causative Variant Identification, In-house Exome Analysis Pipeline