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Abstract Topic:- Clinical Genetics

Abstract Title:- ExoGenius: Transforming Rare Disease Care Through Comprehensive Exome Analysis

Presenting author name :- Vinit Gupta

Presenting author institute:- AarogyaAI Innovations Pvt. Ltd.

Co-authors name:- Harshida Gadhavi, Avlokita Tiwari, Praapti Jayaswal, Harsh Sheth

Co-authors institute:-AarogyaAI Innovations Pvt. Ltd., Decipher Dna Pvt. Ltd.

Aims:-To develop an intuitive genomic analysis platform called ExoGenius that enhances accessibility of exome analysis for germline variants associated with rare diseases, eliminating the requirement for coding expertise. This is aimed at empowering clinical geneticists with genomic insights for precision medicine, to address the challenges faced by the over 300 million rare disease patients worldwide. ExoGenius focuses on early detection and precise characterisation of genetic anomalies caused by germline variants. Individually rare, collectively over 6,000 rare diseases affect millions globally, leading to diagnostic delays, high costs, and limited treatment options that create substantial burdens. By making complex genomic analysis easily accessible, ExoGenius helps transform exome analysis to enable precision medicine and targeted therapies for these rare diseases.

Methods:- ExoGenius is a genomic analysis platform designed for clinical geneticists to analyse exome sequencing data through an intuitive drag-and-drop interface, without needing coding expertise.

In the ever-evolving genetics landscape, our product offers comprehensive insights into:

1. Single Nucleotide Variants (SNVs) identifying causal point mutations,

2. Copy Number Variations (CNVs) detecting structural variants associated with disease risk,

3. Trio analysis assessing inheritance patterns for refined diagnoses.

Method:

* After the hospital/diagnostic center collects the sample, performs DNA extraction and sequencing, the raw sequencing data is uploaded to the ExoGenius platform.

* ExoGenius automatically checks the quality of the sequencing data to ensure it is sufficient for analysis.

* The clinical geneticist then inputs the patient's symptoms and conditions into ExoGenius.

* ExoGenius cross-references this information to analyse the sequencing data and identify:The top 50 candidate genes linked to the patient's phenotypes

* Copy number variations in genes associated with rare diseases

* De novo mutations compared to the parents (if trio data available)

* ExoGenius generates user-friendly visual reports on these results, including the data quality, top candidate genes, CNVs, and de novo mutations.

* This entire process takes approximately 6 hours through ExoGenius's intuitive drag-and-drop interface, eliminating the need for coding expertise or complex bioinformatics pipelines.

Results:- ExoGenius generates detailed reports for SNV, CNV, and trio analysis to provide insights into disease-linked variants, validating analysis quality and aiding variant interpretation for genetic diseases. All reports and result files can be downloaded in a single click for further investigation and validation of variants linked to genetic diseases.Results:

SNV Analysis:

1. SNV Analysis Report containing downloadable files:

* multiqc_report.html: Quality control metrics for sequencing data

* sorted_BQSR.bam: Recalibrated alignment file for variant calling

* sorted_BQSR.bam.bai: Index file for efficient data retrieval

* Recalibration plot.pdf: Visualizes improvements in variant calling

* snps.vcf: Identified genetic variants

* exomiser_result.html: Prioritizes candidate disease variants

2. QC Analysis Report (downloadable): Comprehensive QC assessment

3. Recalibration Plot (downloadable): Visualizes base quality improvements

CNV Analysis:

1. CNV Analysis Report with downloadable files

* CNV_Results.zip: Contains CNV plots and QC metrics

* Genome and chromosome figures visualize CNVs

Trio Analysis:

1. De novo Trio Analysis Report (downloadable)

* exomiser_result.html: Prioritises inherited disease variants

* de novo variants.csv: Identifies de novo variants

Conclusions:- -ExoGenius doesn't just offer features; it offers solutions to real-world problems. Consider the plight of rare disease patients, who often suffer from debilitating conditions with limited treatment options. For example, consider individuals afflicted with conditions like autisam, cystic fibrosis, Huntington's disease, and familial breast cancer. ExoGenius's capabilities in pinpointing germline diseases offer newfound hope to these patients. These results are invaluable for clinical geneticists, aiding in informed decision-making for patient care and clinical diagnostics. Additionally, ExoGenius supports research, providing essential genetic insights that drive advancements in understanding and treating rare diseases.

- What sets ExoGenius apart is our focus on simplifying exome analysis to make precision medicine more accessible. Unlike other commonly used bioinformatics software that requires extensive technical expertise, our intuitive interface democratises access without the need for coding knowledge. This is a key advantage over traditional bioinformatics pipelines that require extensive technical knowledge.

- ExoGenius represents more than just a technological advancement; it's a vision for a healthier future. We aim to transform healthcare by detecting diseases earlier, making treatments more effective, and improving the quality of life for individuals affected by rare genetic conditions. It serves as a catalyst for democratising genetic insights, putting the power of genetics in the hands of those advancing rare disease treatments.

Keywords:- ExoGenius, Rare Disease Solutions, Genomic Analysis, Precision Medicine, Germline Variant Detection