GEMINI: A SOFTWARE PLATFORM FOR DISEASE DISCOVERY

BIOTECHNOLOGY

Flexible, genomic analysis pipeline used to research rare disease, population genetics and familial disease by exploring human genetic variations.

TECHNOLOGY SUMMARY

GEMINI (GEnome MINing) is a flexible online framework used to explore human genetic variation. GEMINI places genetic variants, sample phenotypes and genotypes, and genome annotations into an integrated database. This provides users easy access to a simple, flexible, and powerful system for exploring genetic variation for disease and population genetics. Users upload a VCF file into a database. Each variant is automatically annotated by comparing it to several genome annotations from other sources such as: ENCODE tracks, UCSC tracks, OMIM, dbSNP, KEGG, and HPRD. GEMINI has quickly become a popular tool for rare disease research with over 500 users worldwide. The platform is used at the University of Washington’s Center for Mendelian Genomics and the USTAR Center for Genetic Discovery.

FEATURES AND BENEFITS

- Facilitates genomic analysis of an individual, family trio (father, mother, child), and large, complex families.
- Interprets genomes of diseases in both research and clinical diagnostic settings.
- Increases accuracy by integrating new and rapidly evolving genome annotations and reference datasets.
- Allows customization of self-contained databases for individual labs or companies.

RECENT PUBLICATIONS


INVENTOR PROFILE

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