CIRCULOGENE

One Tube
One Week
Complete Results

INNOVATIVE BIOINFORMATICS WITH UNMATCHED POWER

THE ONE-AND-DONE CIRCULOGENE BIOINFORMATICS ADVANTAGE

In hereditary cancers, insight into genomic risk factors helps steer improved patient care. Circulogene's cutting-edge bioinformatics applies artificial intelligence, machine-learning and database-driven algorithms to deliver highest quality and most accurate SNV, INDEL and structural variant analysis and classification to clinic.

Aggregated Data From Diverse, First-Class Sources

- Automated, validated and up to date with built-in quality systems
- Data from diverse, first-class data sources aggregated, organized, aligned, annotated and classified
- Constant variant reclassification every 3 weeks

Advanced and Fully Integrated AI Greatly Enhances Confident Calls.

- Machine-educated variant calling of pathogenic, likely pathogenic, VUS variants, including automatic pre-classification
- Approx. 200,000 "global inter-lab" calling data sets, on top of eight major public databases to achieve unparalleled accuracy

Big Data-Sharing With Global Medical Community

- Fast, accurate raw data analysis and data sharing with variant calling and annotation
- Regular upload of our reported variants to share with broader medical community

Comprehensive Intuitive Report

- Variant details
- Clinically relevant and actionable information
- Follows ACMG and AMP guidelines for all mutation types
- Simplified data mining correlates millions of internal and external medical data sources, (both public & private)

Complete End-to-End Solution

- Patient centric
- Data analysis
- Classification
- Interpretation and reporting

Security and Privacy

- HIPAA compliant
- Long-term cloud storage
- Clinical grade security, privacy and compliance

OVER 2 MILLION VARIANT CALL DATA

Unlike most labs that use free, public databases, or rely on their own public-inaccessible databases, Circulogene has built a proprietary aggregate integrating the 8 largest public databases with a global community variant-calling atlas.

HUMAN GENETIC VARIANT DATABASES:

The Genome Aggregation Database (gnomAD)
Variants of 15,496 genomes and 123,136 exomes from seven populations worldwide

ClinVar
Currently holds >160,000 submitted interpretations, representing >130,000 variants, affecting >26,000 genes

COSMIC
>170,000 mutations, >2.9 million experiments, >500,000 tumors

ExAC
60,706 exomes from seven populations

ESP
6,503 exomes from European Americans and African Americans

1000 Genomes Project
Genomic data for 2,504 individuals from five populations

CG69
69 individuals with complete genomes

dbNSFP
83,422,341 nsSNVs and ssSNVs (splicing-site SNVs)

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