Better Data for Better Health

The Company

Geneyx was founded with the vision that every patient deserves to be diagnosed and treated with the value of genomics knowledge. As such, our mission has been to develop a clinical genetics data management platform to address critical issues of analyzing and interpreting Next Generation Sequencing (NGS) data in hospitals and genetics labs. Our solution was to introduce a comprehensive yet transparent solution that gives full capabilities to the user while increasing diagnostics yields and turnaround time using best practices and AI.

Geneyx Analysis provides a clinical WGS analysis solutions in a vast number of countries around the globe and is participating in National Genome Projects and academic research institutions.

We strive for a world without undiagnosed and untreated diseases.

By working with physicians and hospital labs worldwide, we continue to make this possible and further support drug discovery and novel therapeutic development. Our comprehensive database includes analysis of highly diverse and data-rich patient cases. New publications describing new Phenotype-Genotype findings are published monthly. Geneyx is teaming with leading strategic partners and collaborates to develop breakthrough solutions.

The Geneyx Solution

Geneyx is a leading tertiary analysis platform for interpreting somatic and germline next-generation sequencing data tailored for high-fidelity long-read sequencing data.

Flexible import capabilities allow for different file formats and batch runs to be processed and analyzed for all sequencing targets, including whole genome, exome, and gene panels. This is coupled with optimized variant calling and annotating to provide quick turnaround times.

Geneyx Analysis stands out for its ability to uncover insights from challenging unexplained cases through advanced processing, visualization, and analysis techniques coupled with updated annotations, phasing, and methylation analysis.

Geneyx combines smart filtering and a novel phenotypic prioritization algorithm that integrates literature and machine learning to present the best candidate variant. This feature is based on identifying publications supporting associations with variant, gene, and disease

or phenotype relationships. PhenoTyper enables improved patient diagnosis and streamlines the clinical laboratory workflow.

Geneyx Analysis can be integrated at any stage of a bioinformatic pipeline. It can be deployed in the cloud, on-premises, or as a hybrid solution, and supports end-to-end workflows, from raw data upload to customizable multilingual clinical reports.

Highlights & Features At A Glance

- Supports all human sequencing targets, genes panels, exomes, and genomes
- Supports Fastq/VCF to clinical report in a single platform
- Includes application for carrier screening and Pharmacogenomic templates and reporting Al-based phenotype prioritizationAnnotation sources with direct embedded links
- Optimized variant annotation and turnaround time
- Phasing analysis
- Visualization of methylation data
- Streamlined workflow through seamless integration SMART filtering for genomes including CNVs, SVs, repeats, fusions, and enhancer analysis
- Customizable filters and workflows for clinical and research settings
- Detailed audit trials and management capabilities
- Free-text or HPO Phenotype-Driven Variant Prioritization
- WGS interpretation with details on regulatory elements
- Automated ACMG/AMP guidelines for variant calling
- Automated Evidence Collection and Reporting
- Customizable and Multilingual Reports
- CE IVD Mark, HIPAA, and GDPR Compliant for Secure Storage and Pipeline Management (ISO 13485, ISO 27001 and ISO 27799)

Raw Data Processing

Geneyx Analysis allows the ability to analyze data from Fastq & VCF, among other file formats, and render a clinical report in one solution. This flexibility enables users to integrate *Geneyx Analysis* at any stage of their bioinformatic pipeline, including secondary & tertiary analyses. In addition, it reduces the need to outsource to other third parties.

- Individual or batch uploads

- API integration supports the incorporation of patient metadata and LIMS communication
- Optimized variant annotation enables a quick turnaround time for analysis

	FASTQ to VCF	VCF Annotation	Total
Panel	< 5 Minutes	< 5 Minutes	~ 15 Minutes
Exome	~ 20M Minutes	~ 5 - 10 Minutes	~ 30 Minutes
Genome	< 2 Hours	< 15 Minutes	<3Hours

Flexible Workflows

Geneyx Analysis can streamline clinical diagnostic workflows while supporting flexibility in clinical research. This platform promotes transparency by allowing full autonomy to the user and gives complete access to customized workflows and protocols. Below are some of the main features that can be automated or customized in an account.

- Incorporate gene lists, gene panels & variant maps
- Automated ACMG/ClinGen Variant Classification with the ability to modify thresholds
- Monthly annotation engine updates via an automated process including OMIM, CADD, Splice-AI, and CIVIC, among +50 other databases
- Annotate CNVs, SVs, repeats & fusions together with Enhancer & Focused SNV Analysis
- CNV/SV visibility within the analysis
- VUS monitor auto-notification
- Geneyx supports the download of BAM, BAI, and QcMetrics files
- Automatic filtering and reporting workflows based on Gene Panel/Exome/Genome and variants of interest
- Ability to make notes on variants while creating the in-house database
- Re-analysis of samples is applicable together with storage of interpretations for downstream case analyses
- Gene Coverage Statistics: Calculate exon and gene coverage statistics at 10X and 20X for the specific genes you define.

- GV Genome Browse Visualization includes creating a new window for IGV, color-coordinated ClinVar variants, the ability to customize the layout, and plotting the variant track

The Geneyx Clinical Applications

The *Geneyx Clinical Applications* are "off-the-shelf" analysis workflows to implement in routine clinical testing.

Carrier Screening

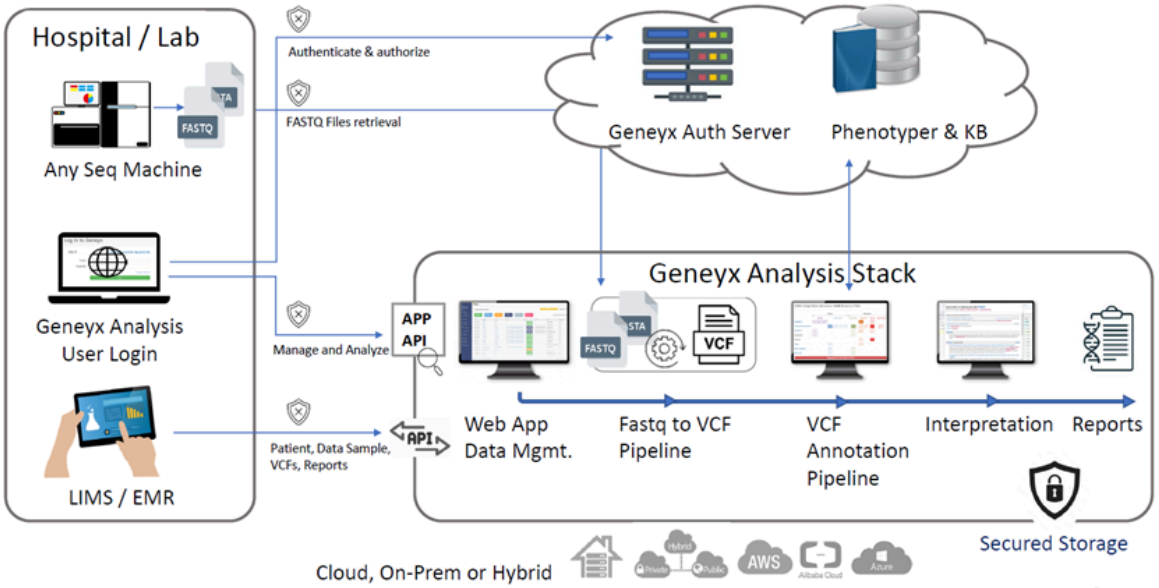
Pre-conception carrier screening has been widely adopted to determine couples' risk of conceiving an affected child with certain autosomal recessive and X-linked disorders. Technology and scientific knowledge have improved carrier screening with greater detection rates of mutations for various populations. The Carrier Screening application analyzes and interprets hundreds of genes to give insights into carrier status for severe and/or debilitating diseases.

Pharmacogenomics

Pharmacogenomics analysis through *Geneyx Analysis* automatically calculates star alleles in all 44 PGx genes with links to relevant information and subsequent reporting on data of known clinical value. The analysis is based on SNP arrays, Exome, and Whole Genome sequencing files.

Integration & IT Architecture

The *Geneyx Analysis* works with hospitals and clinical laboratories to integrate patient data and findings with external data sources by offering comprehensive APIs for LIMs and Electronic Health Record Systems. All data is encrypted and anonymized with deployment on cloud-based (Azure, Ali cloud, Amazon), on-premises behind local firewalls, or a hybrid install.



The Geneyx Analysis end-to-end solution enables Fastq processing to annotate VCF and Clinical Reports within the Geneyx secured ecosystem.

Reporting

The *Geneyx Analysis* supports multilingual and fully customizable reports with the ability to automatically integrate information from more than fifty comprehensive annotation sources.

- Flexible reporting capabilities to accompany specific requirements for organizations
- The report editor enables final modifications to the reports to be signed off with electronic signatures
- Reports are downloadable and can be rendered in multiple formats for incorporation into other formats

Updates Policy

Our development and product teams continually plan and collaborate with our customers and experts to enhance our system and provide our customers with the best possible solution.

The company policy is to update the system monthly and with a significant quarterly release. Don't hesitate to contact our sales team to get our specific release plan.

Publication

For updated publications using *Geneyx Analysis*, please visit https://geneyx.com/category/publications/

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