GENEYX

Better Data For Better Health

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June 21

Geneyx Analysis: an AI-Based WGS/WES Interpretation Platform

- Rapid and accurate phenotype driven interpretation
- Automated ACMG/ClinGen variant classification
- Comprehensive evidence and custom reporting
- Genetic data storage & NGS pipeline management (incl. DRAGEN)
- Full support for SNP Arrays, Panels, Exome and WGS
- 10x Genomic Scope Interpretation, including regulatory regions
- Customizable workflows and integrations with LIMS/EHR
- Cloud upload and data storage, digital consent management

Automated ACMG/ClinGen Variant Classification

detailed evidence Sample & Case-context classification

ACMG/ClinGen variant classifications

Comprehensive assignment of

and automated scoring including

ACMG classifications are not limited to variant-level annotations but include case-level contextual information incl. inheritance models and associated samples.

Comprehensive assignment evidence

Each classification is accompanied with relevant evidence from various annotations and case-level context.

ACMG interpre	tation (Recess	sive) : PDHA1 Miss	ense (R7	2C)				
	Benign			Pathoge	enic	Computational		
	Strong	Supporting	Supporting	Moderate	Strong	Very Strong	Effect	Missense (R72C)
Population		1		PM2	PSA		Repeat (RMSK)	
Population	BAT [B81 [B82			PM2	284		GERP (NR,RS)	5.07, 5.07
Computed & predictive		BPT BP3 BP4 BP7	PP3	PMA PMS	PS1	PVS1	UNIPROT	B7Z3X5
							PRED (LRT,MT)	D, D
Functional	BS3		PP2	PMT	PS3		Polyphen2 (HDIV,HVAR)	D, D
Segregation	BS4		PPT				SIFT	0.0
			~				SPLICING (ADA, RF)	,
De Novo				PM6	P82		Meta SVM	0.9185
Allelic		BP2		PM3			REVEL	0.95
							Functional	
Other databases		BP6	PP5				# Articles	70
Other data		BP5	PP4				ClinVar	Pathogenic, Likely pathogenic
	L	ikely Pathogenic (PS1+PM2+P	P3+PP5)				InterPro Domain	Dehydrogenase E1





Comprehensive Evidence & Custom Reporting



Comprehensive and flexible reporting enables you to use pre-built and customize templates based on the various tests offered by your lab

Automated evidence collection

Automatically generates full and comprehensive clinical reports leveraging structured data available in the GeneCards Knowledgebase.

Customizable report templates

Full customization enables you to offer your customers fully branded and uniquely designed clinical reports.

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Automated ACMG/ClinGen Variant Classification

Comprehensive and flexible reporting enables you to use pre-built and customize templates based on the various tests offered by your lab

Expand fields your lab covers

Rare disorders, Cardiology, Cancer predisposition, Full health screening and more

Automations and Integrations

Automate your best practices and workflows and integrate with your existir pipelines and information systems (LIMS/EHR)

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Genetic Data Storage & NGS Pipeline Management

Get access to unlimited, silo'd, secure genetic data storage on the Geneyx Analysis platform, and leverage out-of-the-box best practices NGS pipelines for various applications or design your own.

Unlimited Cloud-based Genetic Data Storage

Easily manage and store your genetic data in our HIPAA and GDPR-compliant secure cloud, together with all metadata and relevant clinical data.

Flexible NGS Pipeline Management

Start analyzing your data immediately with best practices-based pipelines for whole genome, whole exome and panels, with support for Copy Number Variations, Structural Variations and Repeats across the genome, fully customizable and designed to fit your application.









Thank you!



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