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Geneyx Analysis Technical Datasheet:  
A clinical genetics data management platform

# GENEYX ANALYSIS TECHNICAL DATASHEET: A CLINICAL GENETICS DATA MANAGEMENT PLATFORM

## Overview:

*GeneYx Analysis* is a comprehensive bioinformatic solution for analyzing and interpreting DNA/RNA sequencing data for both NGS and microarrays. This solution offers scalability for short-read and long-read workflows and provides novel biomedical insights for variant-gene and gene-disease associations. With these capabilities, *GeneYx Analysis* has been adopted globally for research, academia, and clinical diagnostic workflows.

## Introduction:

*GeneYx Analysis* empowers hospitals and genetic laboratories with an innovative solution for clinical applications and services spanning germline and somatic workflows. Ingenuity and useability are at the forefront of the application with alignment to both secondary and tertiary workflows. This enables FASTQ and VCF data uploads, among other file formats, for both individual and batch processes. For optimization, *GeneYx Analysis* leverages comprehensive and up-to-date annotation sources for all variant types. This includes single nucleotide variants (SNVs), insertions and deletions (Indels), copy number variations (CNVs), repeat expansions, and structural variations (SVs). Following variant analysis and interpretation, the results can be easily rendered into a customizable clinical report and integrated into a LIMS or EHR system.

## Features:

### Variant Analysis

For variant analysis, the platform offers an intuitive and robust variant browsing interface. Variants are prioritized using customizable filter logic and can be easily sorted using phenotypic prioritization. Annotations are condensed to display the most useful resources but can also be expanded to view the entire genomic information. Hyperlinks are interactive and data can be easily visualized in genome browse for validation purposes. Once a variant has been interpreted, the case can be transferred to other users in your organization and the status can be changed to reflect the current stage in the pipeline.

### Rapid & Comprehensive Annotations

*GeneYx Analysis* optimizes annotation updates for over 50 data sources. These updates occur bi-monthly and include standard ACMG-recommended databases together with all publicly available annotation sources. Licensed annotations can also be integrated on request and all annotation source information can be pulled into a customizable clinical report.



Filters and Tools	RECESSIVE	RECESSIVE COMPOUND HET	DOMINANT HET	RECESSIVE HET	X LINKED	MITOCHONDRIA	OTHER FINDINGS	SECONDARY FINDINGS	CNV/SVs		
Filters	LOCUS	GENE	REF	ALT	AA	HGVS	ZYG	REFSEQ	ACMG	VAR CALLING Q&R	CLINICAL EVIDENCE
Enter Filter Name	7:152158861	KMT2C	A	C		c.11670+2T...	HET	NM_170606.3	LP	Med 38.37 49.33	108.35 1/1 Kleefstra ... AD
GENOMIC AND GENETIC	1:953802	NOC2L	G	A	Q290*	c.868C>T / ...	HET	NM_015658.4	VUS	Med 32.34 51.52	
Depth auto	1:14924716	KAZN	CG	C	G56	c.166delG / ...	HET	NM_001370229.2	VUS	Med 21.30 58.82	
FastTrack auto	1:31814324	SPOCD1	GAG...	G	LGAS...	c.990_1009d...	HET	NM_144569.7	LP	Med 29.25 46.30	
No Mitochondria	1:36093354	ADPRS	C	G	Q354E	c.1060C>G / ...	HET	NM_017825.3	VUS	Med 34.44 56.41	Neurode... AR
noREF auto	1:43338669	MPL	G	A	V114...	c.340G>A / ...	HET	NM_005373.3	BP	Med 40.43 51.81	CONF. LP, ... Myelofib... AD, SO, AR
Always show PAT/LP (ClinVar)	1:44140125	KLF18	GC	G	G502	c.1506delG / ...	HET	NM_001358438.1	VUS	Med 31.22 41.51	
Always show PAT/LP (in house V)	1:46196058	POMGNT1	C	T	R125Q	c.374G>A / ...	HET	NM_017739.4	VUS	Med 34.28 45.16	VUS Muscular... AR
PHENOTYPES	1:70415987	CTH	C	T	T67I	c.200C>T / ...	HET	NM_001902.6	BP	Med 30.34 53.13	CONF. PAT... Cystathio... AR
"Kleefstra syndrome"	1:94042797	ABCA4	G	A	R109...	c.3292C>T / ...	HET	NM_000350.3	LP	Med 42.34 44.74	PAT, LP [Macular ... AD, AR
Weak Association Results	1:111780724	KCNQ3	C	T	R446H	c.1337G>A / ...	HET	NM_001378969.1	VUS	Med 35.31 46.97	Brugada ... AD
DISEASE FREQUENCY (%)	1:220105922	IARS2	T	A	H366...	c.1098T>A / ...	HET	NM_018060.4	VUS	Med 45.38 45.78	Cataracts... AR
0.10%	1:226965300	COQ8A	C	T	R160*	c.478C>T / ...	HET	NM_020247.5	PAT	Med 36.31 46.27	PAT Coenzym... AR
GENE PANELS	1:244856003	HNRNPU	T	C	S690G	c.2068A>G / ...	HET	NM_031844.3	VUS	Med 37.40 51.95	Develop... AD
GENE LIST	2:15474159	NBAS	C	G	A503P	c.1507G>C / ...	HET	NM_015909.4	VUS	High 32.35 52.24	LB Infantile ... AR
VARIANT MAP	2:43875377	ARCG8	G	A	G574R	c.1720G>A / ...	HET	NM_022437.3	VUS	Med 37.32 46.38	PAT, LP [Galblad... AR

Figure 1 : Variant interpretation interface

Annotating a VCF file includes:

1. Variant frequency, clinical significance, and damage effect predictions
2. ClinGen Dosage Sensitivity is available: ClinGen Dosage Sensitivity collects evidence supporting or refuting the haploinsufficiency and triplosensitivity of genes and genomic regions.
3. OMIM for CNV/SV: OMIM phenotype information is now available for CNV/SV analysis. This new field will display OMIM information for all overlapping events observed in the database.
4. ClinVar Origin Annotation: The allele origin, being either germline or somatic, is now displayed when viewing the ClinVar hyperlink for SNVs of interest.
5. Recessive Het Tab: To accommodate for carrier screening workflows, or heterozygous variants with autosomal recessive mode of inheritance, a new Recessive Het tab has been developed that includes the ACMG classification using the autosomal recessive model.
6. Splitting multi-allelic variants and merging identical allelic variants on different transcripts as per ACMG interpretation guidelines.
7. Clinical interpretation of pathogenicity based on ACMG guidelines
8. Annotations based on the user account internal databases, including allele frequency and previous variant and gene annotations.

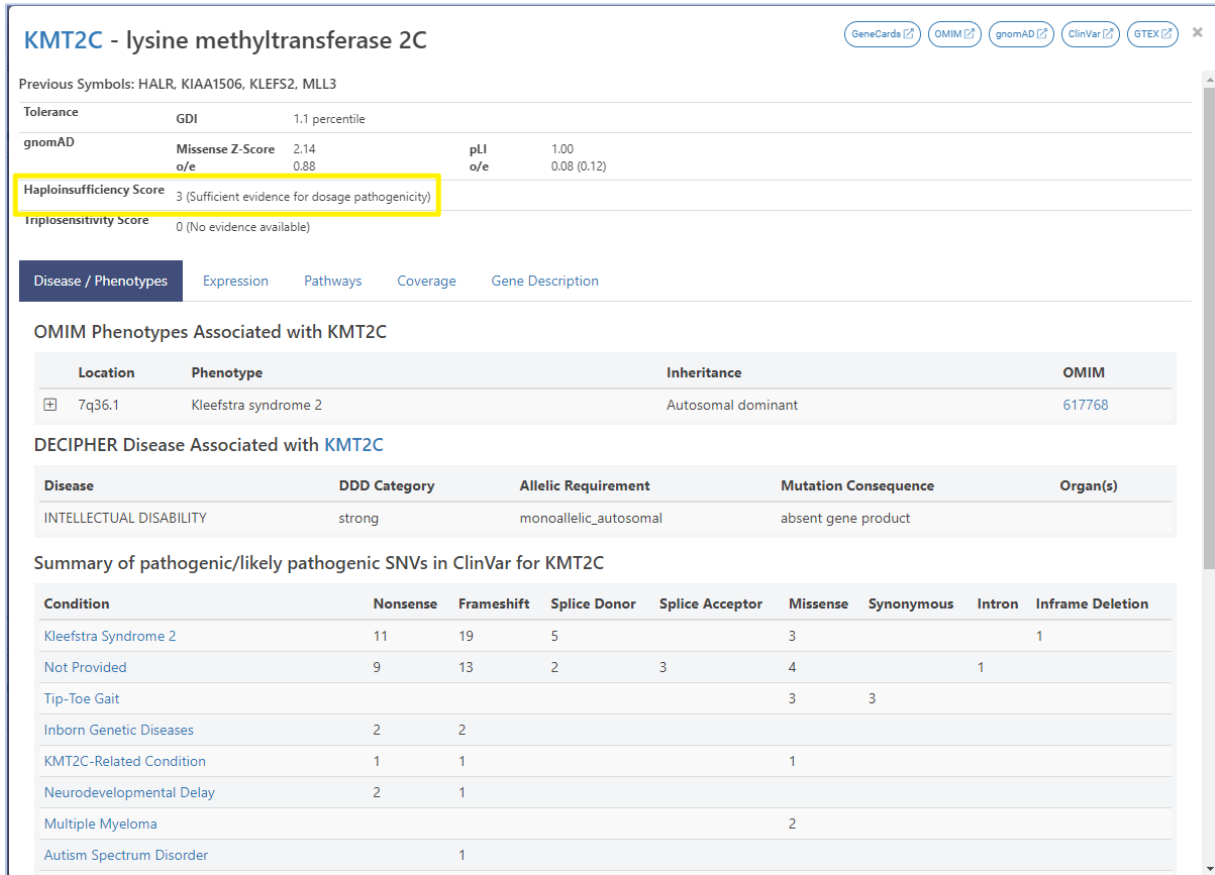


Figure 2 : HI/TP scores are available via ClinGen

CNVs and SVs can be investigated at the gene level and are annotated with clinical data and can be prioritized according to associated phenotype. Compound mutations can also be assessed to view SNVs within the CNV/SV with clinical significance.

### Phenotype-Driven Variant Prioritization

- **Free Text Phenotypes** – Variants can be prioritized based on their association with any free-text biological term or HPO terms, and phenotype specificity is displayed to show gene heterogeneity of the phenotype entered. Multiple phenotypes can be entered using the advanced options and phenotypes can be modified once an analysis has been created.
- **Phenotypic Evidence Score** – Variant scores are created from investigating over 50+ biomedical data sources with the ability to deep-dive into relevant literature to find novel disease associations.
- The GeneYx's phenotyper will highlight association with regulatory regions, as well as enhancers.
- If a phenotype has been observed in a different sample and occurs in the same gene, the phenotypic score will increase based on integration of the users' Clinical Knowledgebase. This is performed on the backend and is proprietary to the users account.

## Association of phenotypes with KMT2C

Matched Phenotypes: "*Kleefstra syndrome*"

Diseases related to KMT2C

- **KLEEFSTRA SYNDROME 2; KLEFS2** <sup>(AD)</sup> [OMIM](#) [Orphanet](#) [Uniprot](#)

**Summaries:**

- A number sign (#) is used with this entry because of evidence that *Kleefstra syndrome*-2 (KLEFS2) is caused by heterozygous mutation in the KMT2C gene (606833) on chromosome 7q36.
- *Kleefstra syndrome*-2 (KLEFS2) is an autosomal dominant neurodevelopmental disorder characterized by delayed psychomotor development, variable intellectual disability, and mild dysmorphic features (summary by Koemans et al., 2017).
- For a discussion of genetic heterogeneity of *Kleefstra syndrome*, see KLEFS1 (610253).
- Kleefstra et al. (2012) reported a girl with *Kleefstra syndrome*-2.
- A form of *Kleefstra syndrome*, an autosomal dominant disease characterized by variable intellectual disability, psychomotor developmental delay, seizures, behavioral abnormalities, and facial dysmorphisms.

**Publications:**

- **Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder.** ([PubmedId 29069077](#))  
 Abstract: *Kleefstra syndrome*, caused by haploinsufficiency of euchromatin histone methyltransferase 1 (EHMT1), is characterized by intellectual disability (ID), autism spectrum disorder (ASD), characteristic facial dysmorphisms, and other variable clinical features.... In addition to EHMT1 mutations, de novo variants were reported in four additional genes (MBD5, SMARCB1, NR1H3, and KMT2C), in single individuals with clinical characteristics overlapping *Kleefstra syndrome*.... Our clinical data delineates the KMT2C phenotypic spectrum and reinforces the phenotypic overlap with *Kleefstra syndrome* and other related ID disorders.

Figure 3: Phenotypic prioritization using key search terms

### ACMG Guidelines

The American College of Medical Genetics and Genomics (ACMG) represents the interests of clinical geneticists, clinical laboratory geneticists, and genetic counselors by providing best practice guidelines. GeneX Analysis adheres to these guidelines by providing:

- *ACMG Automation* – GeneX Analysis automates 18/28 ACMG criteria, reducing workflow complexity and ensuring best practices are followed. Users can define thresholds for application of a subset of criteria.
- *Sample & Case-context classification*- ACMG classifications automatically include case-level information such as inheritance models and relevant findings in associated samples.
- *Ability to modify* – If additional information or expertise is available, users can incorporate their data, modify criteria, make notes, and store changes for all downstream cases.
- Easy access to information relevant to manual curation of variant pathogenicity
- Support for reanalysis with updated evidence and annotations

GeneX Analysis adherence to the ACMG Guidelines has been validated by labs in the United States, Europe, Israel, Hong Kong, and China. Further, it has been approved for use in CLIA-CAP genetic laboratories.

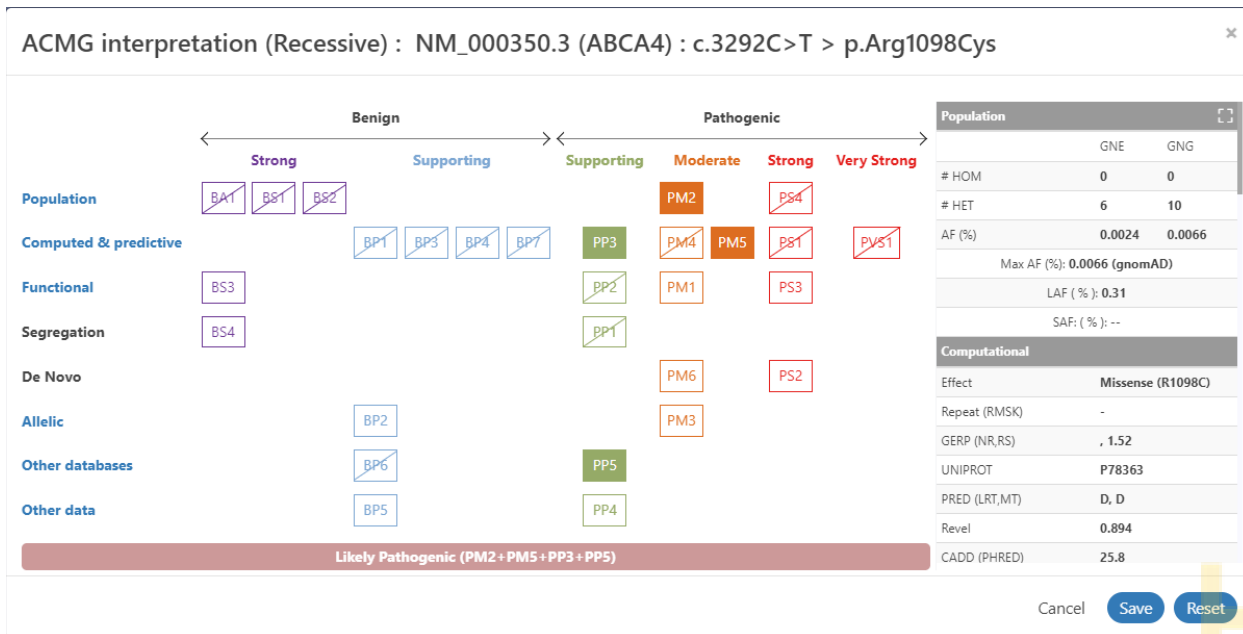


Figure 4: ACMG dialog with ability to modify criteria

### Customer Local Database & Inter-Lab Sharing

Geneyx Analysis automatically calculates an 'in-house' allele frequency annotation for all variants in an account that is derived from all imported samples. This is useful to eliminate common variants or enable variant selection among different ethnic groups. The in-house database also includes annotations and interpretations previously entered by the analysts, which assists in applying accumulated in-house knowledge to new cases. Laboratories may also participate in Inter-Lab sharing, especially if an organization is present in several locations. This permits viewing variant allele frequency within all cases in accounts sharing the data.

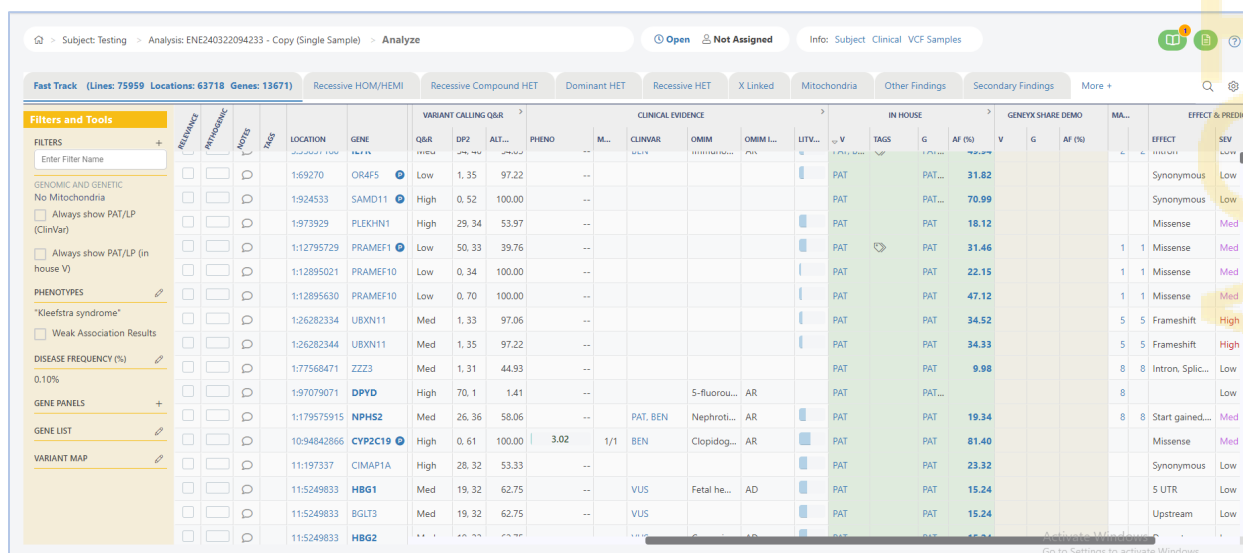


Figure 5: In-house database in green shows allele frequency and previous classifications from other samples



## Whole Genome Sequencing Interpretation

*GeneX Analysis* incorporates a comprehensive genome-wide map of regulatory elements including promoters and enhancers together with their gene associations from various sources. This enables the association of variants in non-coding regions to gene-phenotype relationships and subsequent prioritization.

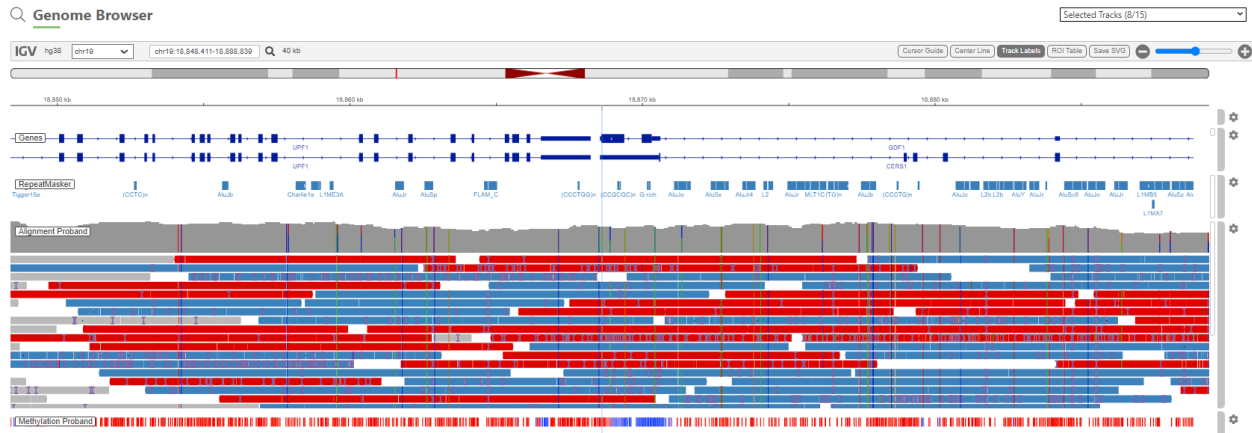


Figure : BAM file visualization using integrated IGV

## Evidence Collection and Reporting

- *Automated evidence collection* – GeneX Analysis supports automatic generation of comprehensive clinical reports leveraging available annotations & phenotypic evidence.
- *Customizable report templates* – Reports are fully customizable to include the organization logo and information that the user deems necessary for a distinctive design.
- *Selected Variants' feature*: Once the relevance and associated information is set, user can review the selections across all the genetic models using this feature.
- *Exporting flexibility* – Reports can be rendered in PDF and JSON format and integrated with LIMs/EHR systems where necessary.

Filters and Tools	Location	Gene	Ref	Alt	AA	HGVS	ZIG	OT	ASS.	ACMG	Variant Calling	QBR	DP2	ALT.	PHENO	M.L.	ClinVar	OMIM	OMIM L.	LITV.	V	G	AF L.	IN HOUSE	MATCHED CNV/SVS	EFFECT	SEV	CAID (PRL)	FREQUEN.	MAX AF (%)
<input checked="" type="checkbox"/>	19:18868625	GDF1	A	G	MDS4T	c.1091T>C / p.MeL...	HET			High	11, 12	52.17	116.53	2/2	PAI, LP	Congenital...	AD, AR		PAL...	PAL...			0.022		Missense	Med	27.80	0.051		
<input checked="" type="checkbox"/>	19:18868907	GDF1	C	A	R27DL	c.809G>T / p.Arg...	HET			High	12, 11	47.83	116.53	2/2		Congenital...	AD, AR		VUS	PAL...			0.022		Missense	Med	24.80	0.0013		
<input checked="" type="checkbox"/>	6:148520341	SASH1	C	T	R314C	c.940C>T / p.Arg...	HET			High	14, 17	54.84	13.82	1/2		Cancer, m...	AR, AD						0.066		Missense	Med	21.60	0.0066		
<input checked="" type="checkbox"/>	1:8361046	RERE	A	G	AGL...	c.2449_2450dup...	HET			Med	12, 10	45.45	12.43	1/2	CONF, VUS...	Neurodeve...	AD						0.087		Inframe indel	Med		0.20		
<input checked="" type="checkbox"/>	17:10647205	MYH3	G	C	S292C	c.875C>G / p.Ser...	HET			High	17, 16	48.48	9.71	1/2	CONF, VUS...	Arthrogryp...	AD, AR						0.022		Missense	Med	29.50	0.099		
<input checked="" type="checkbox"/>	2:178654965	TTN	T	T	TCTT...	E126... c.38068_38069ins...	HET			Med	11, 9	42.86	7.05	1/2		Cardiomyo...	AD, AR						0.022		Inframe indel	Med		...		
<input checked="" type="checkbox"/>	2:178659225	TTN	T	T	TCTT...	E124... c.37315_37316ins...	HET			Med	16, 5	22.73	7.05	1/2		Cardiomyo...	AD, AR						0.022		Inframe indel	Med		...		

Figure 7 : Annotating variants to pull into report and in-house databases

## Storage and Pipeline Management

- *Unlimited Cloud-based Genetic Data Storage* – Easily manage and stores genetic data in a secure private GeneX Azure cloud, (HIPAA and GDPR-compliant).
- *Local or Hybrid Configurations* – On-premise configurations and Hybrid architecture are also available if an organization needs to remain behind local firewalls.



- **Account Management** – Restrictions can be set at a user or group level to ensure assigned, fixed workflows are utilized to streamline processing.
- **Storage Allocation:** Display the storage space allocated or consumed by the data files within the specified timeline. This information allows users to track their storage usage and manage their resources effectively.
- **Audit Trails** – All information from FASTQ to report is documented and can be reviewed at any time.

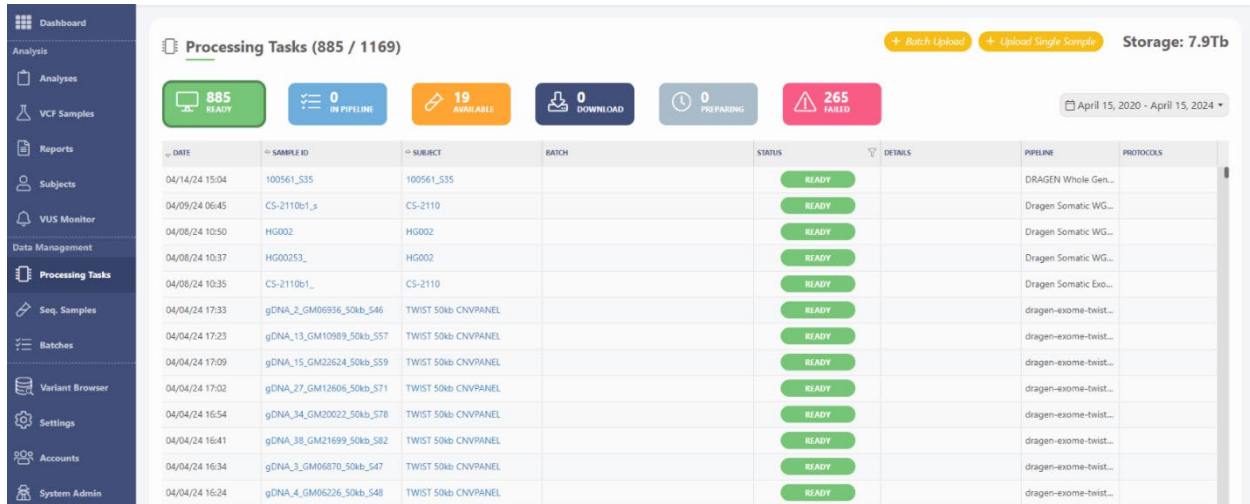


Figure 8 : Complete visibility of data workflows and management features

### Customizable Workflows and Protocols

Genexus Analysis provides customizable protocols which enable laboratories to standardize genetic test and analysis workflows. Protocols can include combinations of simple or complex filters on any set of annotation entities for any gene lists or genomic regions and can be locked as a protocol for defined users. Protocols can also be optimized to automatically render a report on the remaining variants and include any user interpretations for given variants.





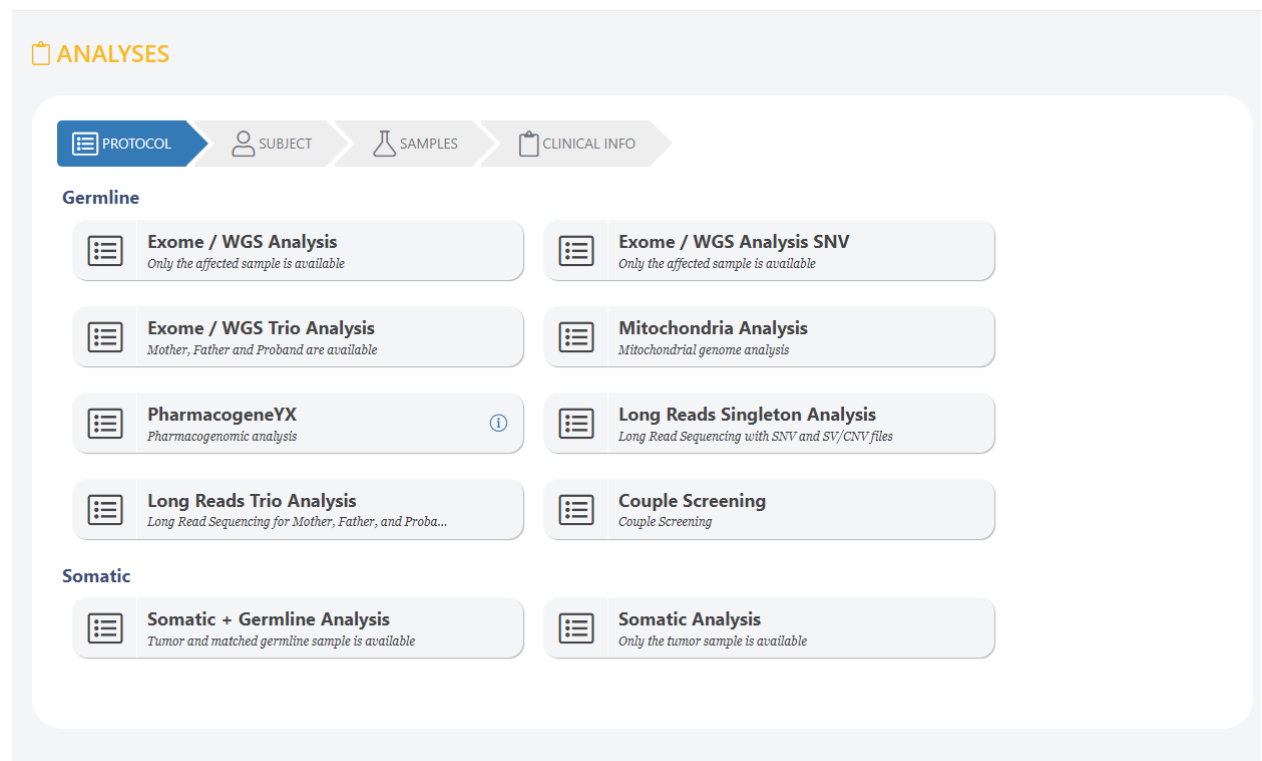


Figure 9: Default and customizable protocols are available

### Automation and APIs

GeneX Analysis API feature allows users to connect with existing LIMS/EHR systems and upload data using command line workflows.

1. Integration with primary and secondary analysis pipelines to allow automated import and annotation of VCF files. This works with single sample and batch workflows.
2. Integration with laboratory information management systems (LIMS) or electronic health record (EHR) systems enables the creation of automated analyses of patient clinical information and the streamlining of reports from GeneX Analysis to the LIMS/EHR.
3. Auto-Reporting for SNVs/CNV/SVs to generate TSV files based on your customized filtering.

Other API features provided by GeneX Analysis can be found here: <https://github.com/geneyx/geneyx.analysis.api/>.



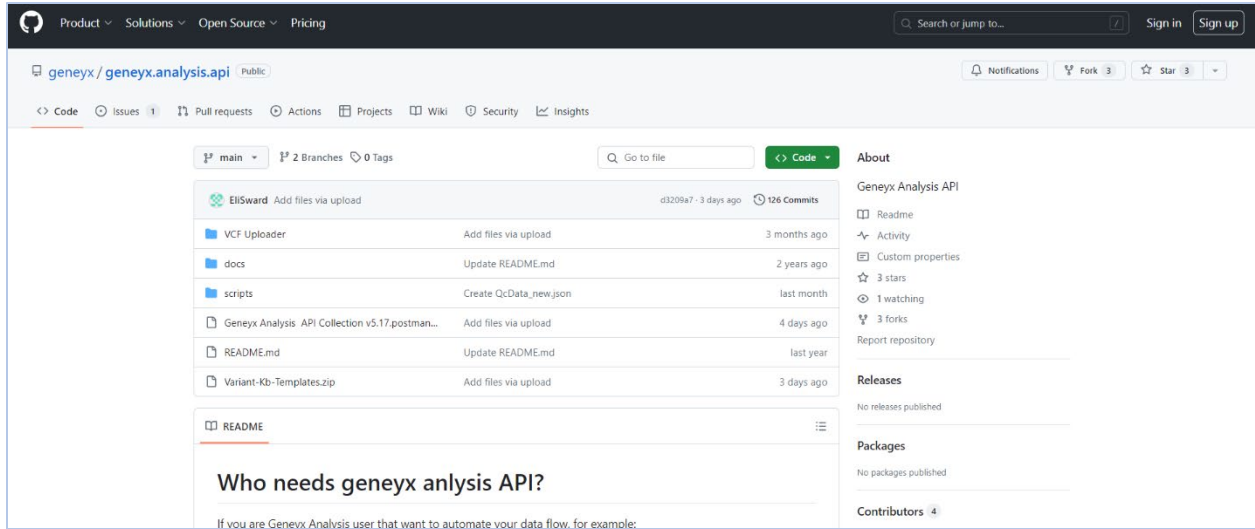


Figure 10: APIs available on our GitHub

### Variant Knowledgebase

Geneyx’s classified variants are organized within a knowledgebase, uniquely tied to each account. Users can now harness the capability to export and import this valuable data. An exemplary use case involves lifting over annotated variants to an alternate genome assembly, leveraging the transformed data as an allele frequency backlog for augmented evidence across different assemblies

Reference	Pathogenicity	Inheritance	Location	Gene	Zygosity	RefSeq	Ref	Alt	OMIM	Interpretation	Recommendations	Phenotypes	Created By	Created	Modified By	Modified
1	SP		7:286468	FAM20C	HET	NM_020223.4	G	GGAC...	259775	This is a variant interpretation	This is a variant level recom...	"noonan syndrome"	Pavel Shata...	04/21/24 02...	Pavel Shata...	04/21/24 02...
1	SP		1:201328373	TNNI2	HET	NM_001276...	G	A	CARDIOMYOPATHY, FAMIL...	Mutations in TNNI2 are ass...	Genetic testing for mutatio...	"Congestive heart failure"	Geneyx Sup...	04/12/24 17...	Geneyx Sup...	04/12/24 17...
1	SP		1:69270	ORAF5	HET	NM_001005...	G	A	NM_001005484.2	Keihanaikukaukahihulihe?e...	Lorem ipsum dolor sit amet...			04/14/24 03...	Pavel Shata...	04/14/24 03...
1	SP		1:69270	ORAF5	HOM	NM_001005...	A	G				(wolman OR ossd OR kuru O...		04/21/24 02...	Pavel Shata...	04/21/24 02...
1	SP		1:69511	ORAF5	HOM	NM_001005...	A	G				(wolman OR ossd OR kuru O...		04/21/24 02...	Pavel Shata...	04/21/24 02...
1	SP		1:7724727	CAMTA1	HET	NM_015215.4	C	G	614756				Pavel Shata...	04/21/24 02...	Pavel Shata...	04/21/24 02...
2	SP		1:859404	LINC02...	HOM	NR_026874.2	C	G						04/14/24 03...	Yoav Eshel	04/15/24 06...
1	SP		1:878314	SAMD11	HET	NM_001385...	G	C					Pavel Shata...	04/21/24 02...	Pavel Shata...	04/21/24 02...
1	SP		1:911595	PERM1			a	g					Pavel Shata...	04/10/24 09...	Pavel Shata...	04/10/24 09...
1	SP		1:911595	PERM1	HOM	NM_001291...	A	G				kuru wolman "Stapes ankyl...		04/14/24 03...	Yoav Eshel	04/15/24 06...
1	SP		1:911595	PLEKHV1	HOM	NM_032129.3	A	G				"Hypertension associated w...		04/14/24 03...	Yoav Eshel	04/15/24 06...
1	SP		1:94471075	ABCA4	HOM	NM_000350.3	A	G	604116			(wolman OR ossd OR kuru O...	Pavel Shata...	04/21/24 02...	Pavel Shata...	04/21/24 02...
1	SP		11:108158921	ATM	HOM		G	G	208900				Pavel Shata...	04/21/24 02...	Pavel Shata...	04/21/24 02...
1	SP		11:8891696	TYR	HET	NM_000372.5	C	A				"noonan syndrome"	Pavel Shata...	04/21/24 02...	Pavel Shata...	04/21/24 02...

Figure 11: Variant Knowledgebase of interpreted variants

### VUS Monitor

Keeping track of variant classification changes can be a daunting task. To simplify this process, we have implemented an auto-notification feature that alerts you whenever variants undergo classification changes according to ClinVar. This proactive notification provides comprehensive details, including a direct link to the analysis, enabling you to perform retrospective analyses with ease and precision.



## VUS Monitor

With the VUS monitor, you can keep track on previously reported variants of uncertain significance(VUS) that have undergone a change in their clinical significance according to ClinVar. Additionally, it provides the ability to reanalyze the sample.

hg19 (c)		hg38 (s)		SUBJECT										CURRENT CLASSIFICATION				UPDATED CLASSIFICATION		STATUS
LOCATION	REF	ALT	GENE	EFFECT	ZYGOSITY	PATIENT ID	SELECTION DATE	SAMPLE ID	ANALYSIS	VERSION	REL.	PATHO...	NOTES	OMIM	CLINVAR	UPDATED CLINV...	NEW ANALYSIS			
19:10273296	GAAA	G	DNMT1	Intron	HET	test for Reports ...	04/07/2024	14082.dragen.wes.hg19.20...	ENE210505203509 - Para...	31	1	VUS	Cereb...			LB	ENE210505203509 - Copy ...	Open		
16:27356203	A	G	IL4R	Missense	HET	YM	09/20/2022	testapi2.vcf	ENE210722121431 - SNV/...	32	2	VUS	(Atopy...	PAT, PROT	BEN			Open		
3:165548529	T	C	BCHE	Intron	HET	39398	06/26/2023	39398.dragen.wes.grch37...	ENE230626153951 - Single...	V5.13	2	VUS		PAT, LP	CONF, PAT, LP, B...			Open		
3:165548529	T	C	BCHE	Intron	HET	100547	07/29/2023	100547.dragen.wes.grch37...	ENE230110144805 - Copy ...	v5.14	1	VUS		PAT, LP	CONF, PAT, LP, B...			Open		
3:165548529	T	C	BCHE	Intron	HET	39398	07/13/2023	39398.seniteon.wes.grch37...	ENE230713142726 - Single...	v5.14	3	VUS		PAT, LP	CONF, PAT, LP, B...			Open		

Figure 12: VUS Monitor to track variants of unknown significance through annotation updates

## Uniparental Disomy

UPD is automatically calculated in *GeneX Analysis* when running trio exomes and genomes by comparing each variant of the proband to the parents. Notifications are provided if significant results are identified.

Identity by Descent			
	Count	Proband	%
HET in both parents	4,780	1,158 (HOM)	24.2%
HOM-ALT in Mother, HOM-REF in Father	1,321	1,250 (HET)	94.6%
HOM-ALT in Father, HOM-REF in Mother	1,409	1,339 (HET)	95.0%

UPD Analysis (Details)		
Chromosome	P Value	Category
1	0.000090	Mat_isoUPD
11	0.00031	Pat_hetero/isoUPD
15	6.6e-59	Mat_hetero/isoUPD

Figure 13: UPD Analysis is calculated for all trio workflows

## Comprehensive and Customizable Reports

A comprehensive and flexible reporting system of modular tools and features that can be combined and customized to create personalized analysis pipelines. These tools cover a wide range of applications, including variant calling, annotation, filtration, pathway analysis, and interpretation. Users can select the relevant tools for each step of the analysis, configuring them to meet their specific requirements. The comprehensive clinical reports can be customized according to the organization's logo and formatting requirements to incorporate static and dynamic information. The reports are also offered in multiple languages, preventing the need to translate generic information.



## Single Sample Analysis Report

### Patient

Name: John Doe  
ID:  
Patient ID: Patient - 24544  
Gender: Male  
Accession: newWGS\_44340.dragen.wgs.hg38.20231211-1548533.vcf.gz  
Case ID: ENE240322094233

### Lab

Referring physician:

### Main Specimen(s)

Specimen Type: Blood

Report Date: 15 Apr 2024

### Clinical Information

Phenotypes: "Kleefstra syndrome"

### Result Summary

 **Result: Positive - Significant Mutation identified**

### SNV Result Details

No	Gene	Transcript	Position	Nucleotide	Amino Acid	GnomAD	dbSNP	Zygosity	ACMG Classification
1	KMT2C	NM_170606.3	7:15215886 1	c.11670+2T>G		--		HET	Likely Pathogenic

1. Variant identified *KMT2C* (c.11670+2T>G)

The Heterozygous A->C Intron, Splice site donor at chr7:152158861 is predicted to result in abnormal protein translation of the

Figure 14: Reporting outputs in Genex are fully customizable

### Account Management and Audit Trails

Account management capabilities enable the group leader to set permissions for individual users to ensure complete control of all processes and steps performed by individual users to streamline workflow and manage data applications. All processes performed in the application are stored and can be reviewed, offering a comprehensive audit trail for laboratories.

Analysis History <span style="float: right;">x</span>			
Modified	By	Type	Details
04/15/24 15:26	Eldar Dedic	Report created	ENE240322094233 - Copy - Single Sample - 240415_1326.pdf
04/15/24 15:22	Eldar Dedic	Report created	ENE240322094233 - Copy - Single Sample - 240415_1321.pdf
04/15/24 14:33	Eldar Dedic	Filter removed	CMH Alternate Read
04/15/24 14:33	Eldar Dedic	Filter removed	CMH Exclusion ClinVar
04/15/24 14:33	Eldar Dedic	Filter removed	CMH GnomAD AC
04/15/24 14:33	Eldar Dedic	Filter removed	CMH GnomAD AF
04/15/24 14:33	Eldar Dedic	Filter removed	CMH GnomAD Hom Hem
04/15/24 14:32	Eldar Dedic	Phenotypes updated	"Kleefstra syndrome"
04/15/24 14:31	Eldar Dedic	Filter removed	CMH Severity

Figure 15: Audit trails output for all actions performed in an analysis

### Long-Read sequencing

GeneX Analysis provides a comprehensive tertiary analysis platform for the efficient processing of Oxford Nanopore Technologies and PacBio Long-Read sequencing data. We've introduced innovative features that facilitate rapid data ingestion and annotation of long-read data, streamlining the entire process of variant interpretation and reporting.

- Phasing analysis: Our specialized tools, including phasing and methylation analysis simplify the bioinformatics process, ensuring accurate variant annotation and classification.
- Comprehensive view of overlapping CNVs and SNVs for compound heterozygous variant detection
- SMART Filtering to quickly reduce background noise associated with long read sequencing data
- Repeat Expansion analysis aimed to identify deleterious repeats and potential gene silencing events
- Ability to import all variant file outputs



Better Data for Better Health





Figure 16: Allele phasing integration where variant is observed in trans

### Data Protection Compliance

- HIPAA (Health Insurance Portability and Accountability Act)
- GDPR (General Data Protection Regulation) compliant.
- ISO 13485( designed for medical equipment and software manufacturers to ensure an internationally recognized standard for quality management.)
- ISO 27001 (**standard for information security management systems (ISMS)**)
- CE IVD Mark
- All private patient information is encrypted.



### Overview Summary

GeneX Analysis is a powerful secondary and tertiary analysis platform for annotation, analysis, and prioritization of coding and non-coding genomic variants using short and long-read sequencing analysis. This platform provides access to an extensive knowledge base of genomic annotations and offers intuitive and flexible configurations. The ease of use and transparency allows quick adaptation and implementation into clinical workflows and innovative features that simplify and accelerate variant interpretation. GeneX Analysis is an end-to-end solution demonstrating a unique paradigm that can increase diagnostic yield and reduce turnaround time for all users.

GeneX Analysis is available for trial at [www.geneyx.com](http://www.geneyx.com)

### Publications:

Beyond the Exome: Utility of Long-Read Whole Genome Sequencing in Exome-Negative Autosomal Recessive Diseases..



Better Data for Better Health