

From Sequencing to Insight: An In-House, Unified Platform for Sample Tracking, QC Assessment & Genomic Data Exploration in Research and Clinical Settings

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Background

Sidra Medicine's Integrated Genomics Services (IGS) supports a wide range of research projects, national genomic initiatives, and clinical diagnostic workflows.

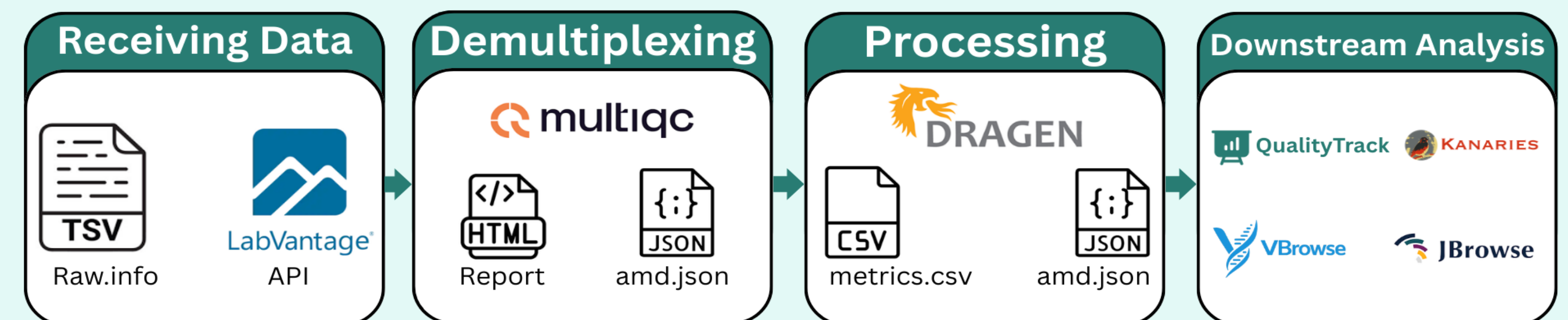
These activities generate large volumes of genomic data and complex metadata that must be tracked, quality-controlled, and interpreted efficiently. Moreover, such data is being produced across the different IGS cores, making it more complicated to track.

Ensuring transparency, reproducibility, and compliance with accreditation requirements necessitates a unified digital infrastructure capable of supporting both upstream quality management to help the Cores, and downstream genomic data exploration to support researchers and clinicians.

Aim

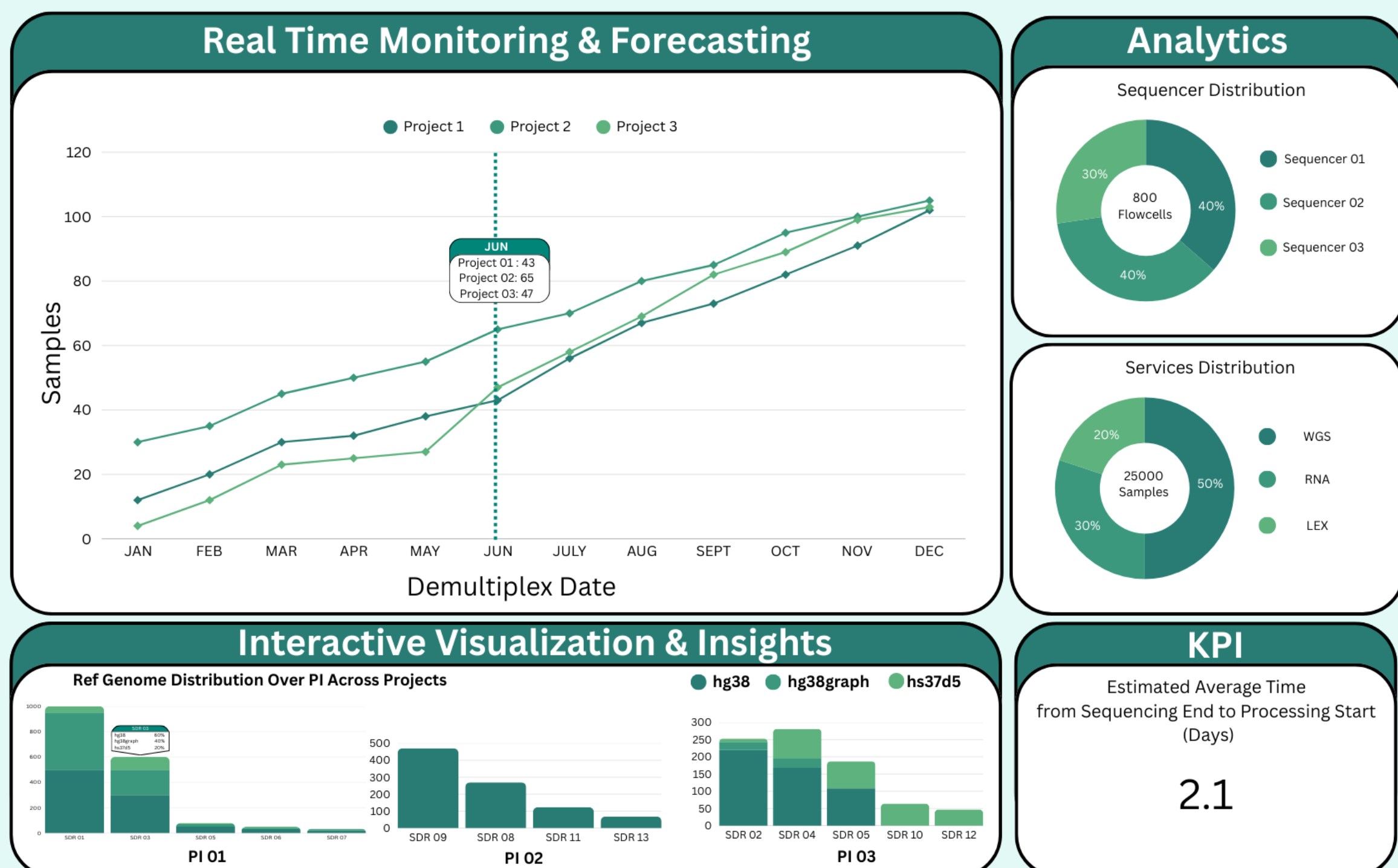
To develop and deploy an in-house, unified, web-based platform that:

- Provides real-time sequencing quality intelligence (QualityTrack)
- Harmonizes genomic and sample metadata across multiple cores
- Enables interactive genomic data visualization (JBrowse)
- Supports scalable, cohort-level variant interrogation (Vbrowse)
- Serves both research and clinical genomics workflows
- Offers role-based access and intuitive interfaces tailored to the users



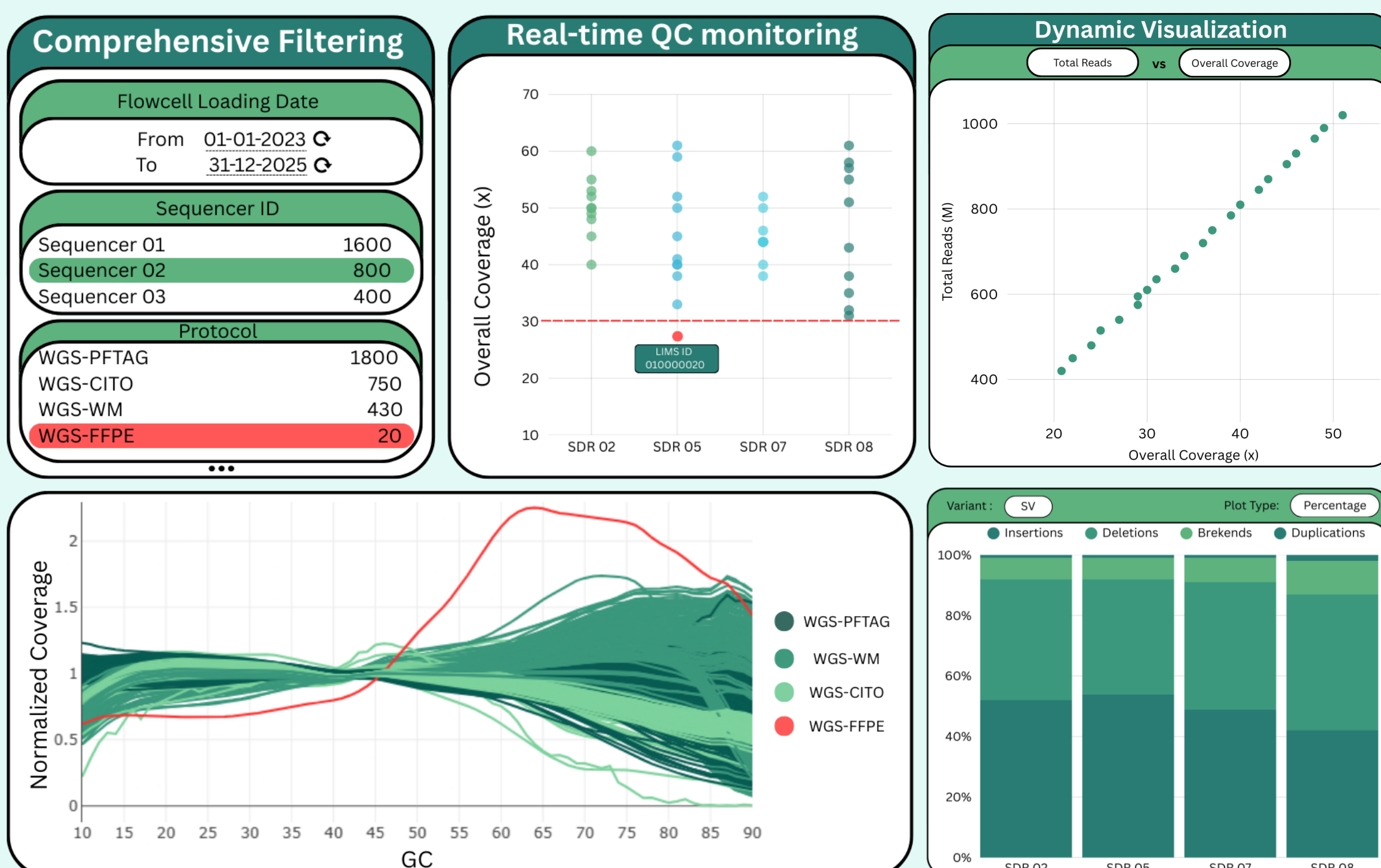
Dashboard

Overview of Samples' Metadata & Instrument Performance Across Multiple Platforms



Quality Control Metrics

Extensive Assessment of DRAGEN Mapping QC and Variant Calling QC



Authentication & Authorization

Unified Access Portal Integrating Institutional Credentials with REDCap

Sign In

Enter your username and password to sign in

Your Username: @smrc.sidra.org

Password:

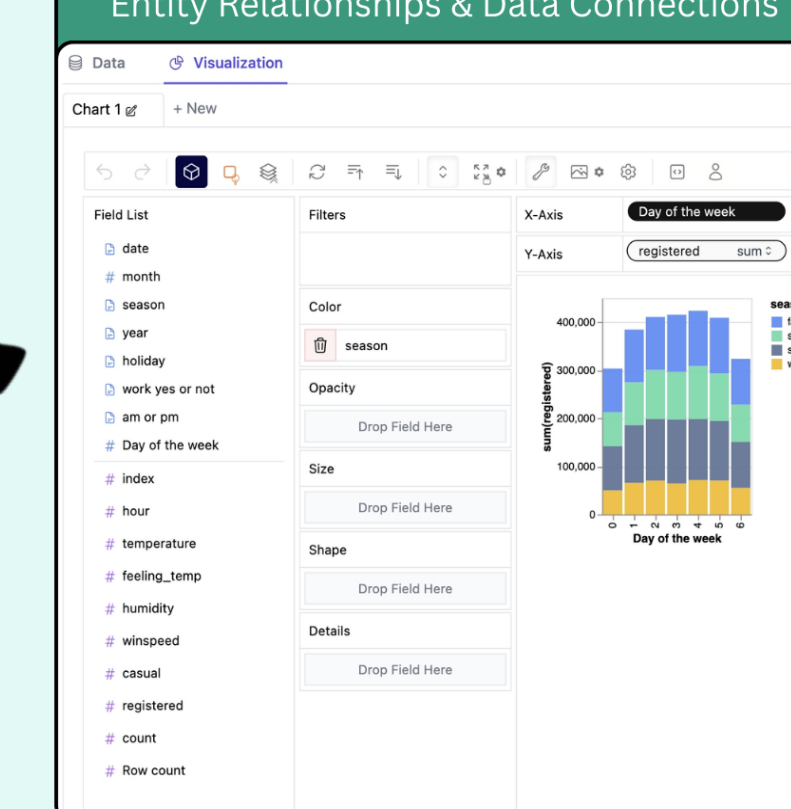
SIGN IN

Tabular View with Tool Integration

Sample	Flowcell	Service	Ref. Genome
000000010	HLHWNXXXX	WGS-PFTAG	hg38
000000020	ZJHJFCXXX	mRNA	hg38graph
000000030	HWYJLJXXX	LEX	hg38

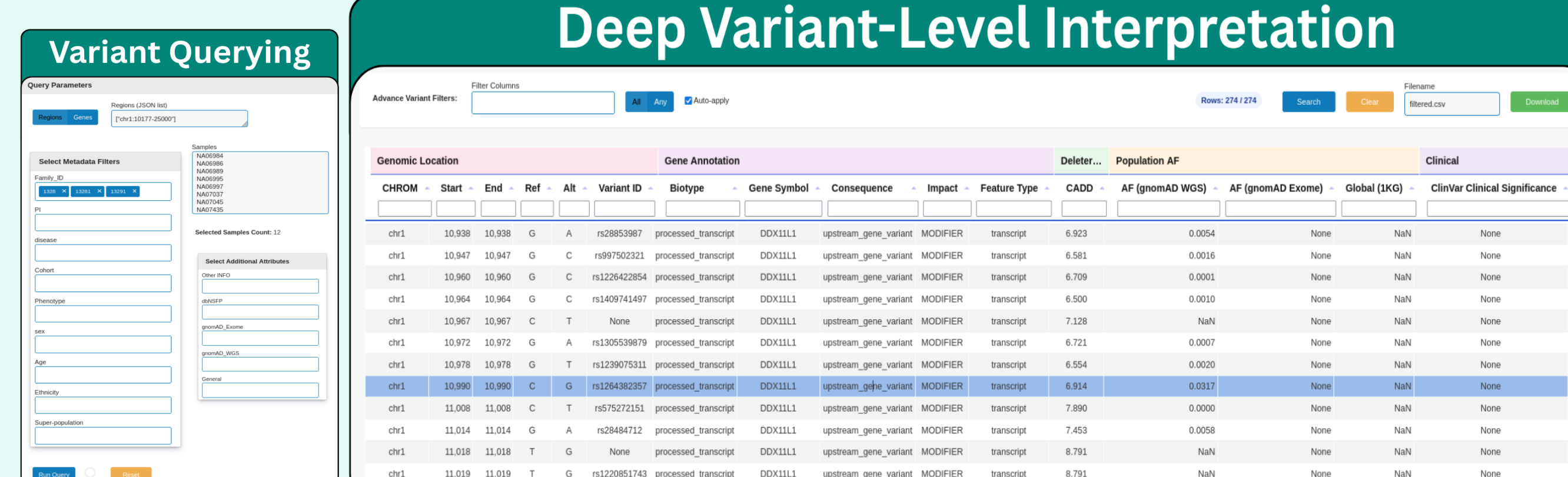
Graphic Walker

Interactive Visual Explorer Tool for Navigating Entity Relationships & Data Connections



VBrowse

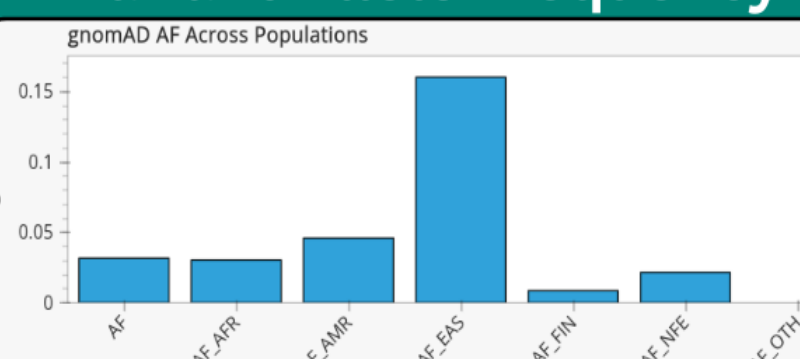
End-to-End Variant Exploration & Interpretation in One Interface



Variant Summary Table

Field	Value
ID	rs1264382357
Ref / Alt	C/G
Genomic Coordinates	chr1:10990-10990
gnomAD (variant)	1-10990-C-G
Franklin (variant)	1-10990-C-G

Variant Allele Frequency



Variant Consequence Info

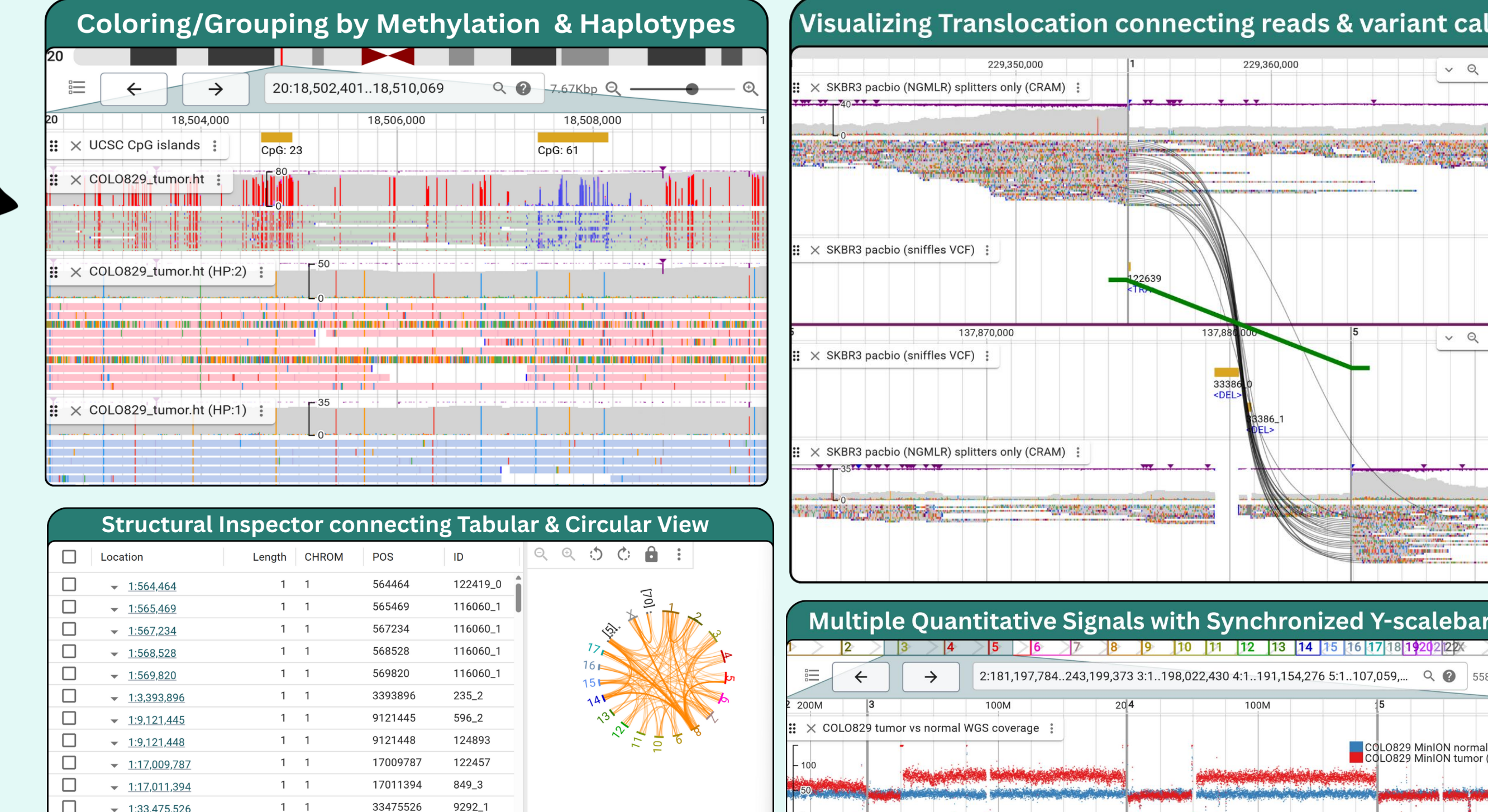
Field	Value
upstream_gene_variant	
MODIFIER	
Gene Symbol	Genecard (COX11), Ensembl (COX11)
Gene ID	N/A
Feature Type	transcript
Feature ID	N/A

ClinVar Annotation Table

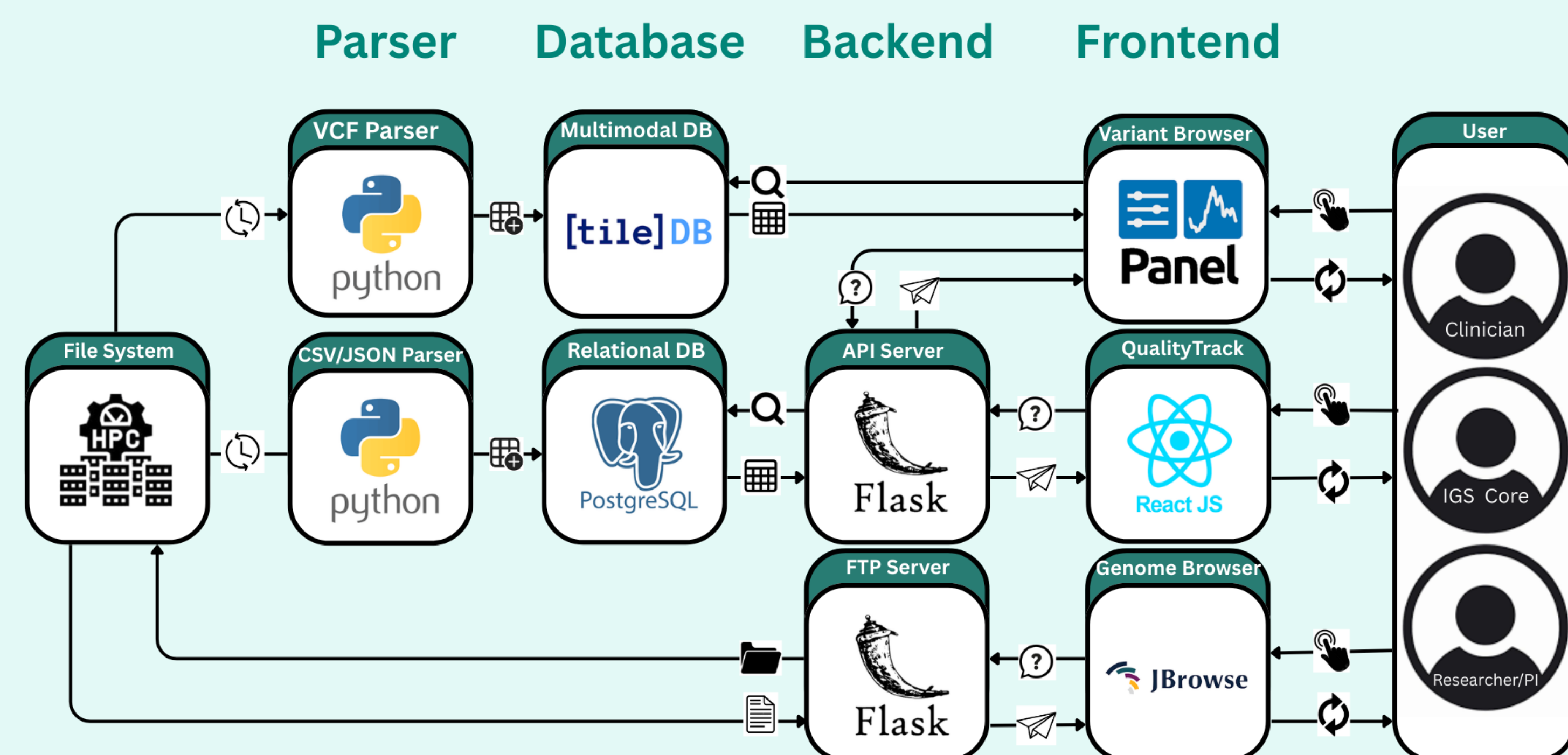
Field	Value
Disease Names (CLNDI)	Familial Hypercholesterolemia
Review Status	Reviewed by expert panel
Clinical Significance	Pathogenic
Variation ID	223658

Jbrowse

Open-source Genome Browser with Dynamic Client-side Interface



Methods



Conclusion

- Faster turnaround times through streamlined data access
- Improved reproducibility & end-to-end traceability for compliance
- FAIR-aligned data stewardship supporting interoperability of datasets
- Enhanced communication across research, clinical, and IGS core teams
- AI-ready datasets for future precision diagnostics & advanced analytics

Acknowledgement

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