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.

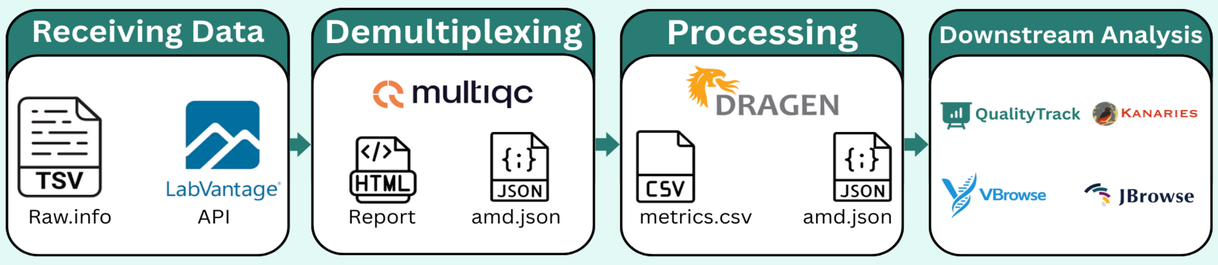
These activities generate large volumes of genomic data and complex metadata that must be tracked, quality-controlled, and interpreted efficiently. Moreover, such data is being produced across the different IGS cores, making it more complicated to track.

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

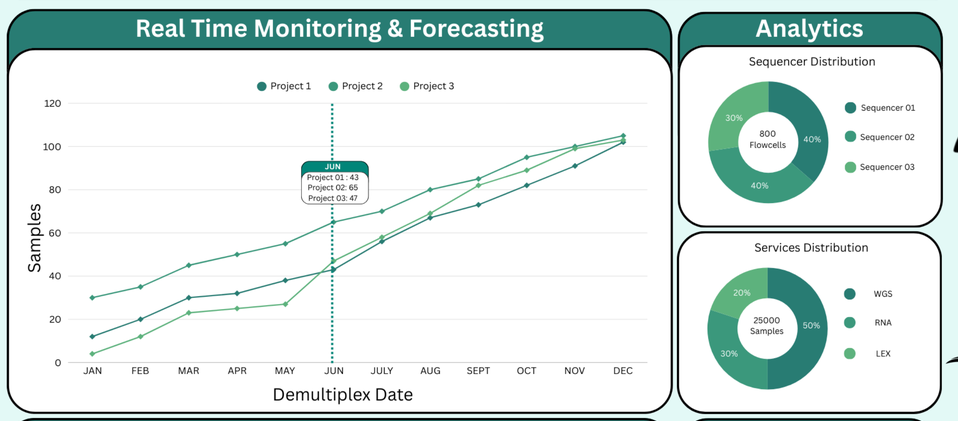
To develop and deploy an in-house, unified, web-based platform that:

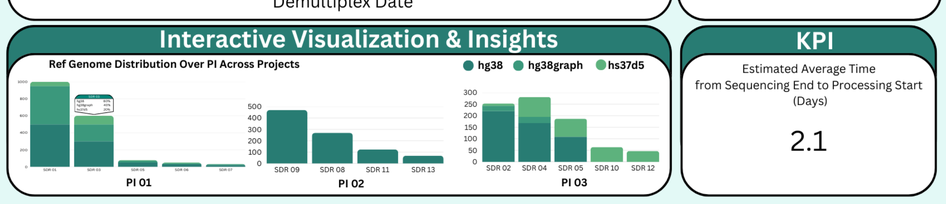
- Provides real-time sequencing quality intelligence (QualityTrack)
- Harmonizes genomic and sample metadata across multiple cores
- Enables interactive genomic data visualization (JBrowse)
- Supports scalable, cohort-level variant interrogation (Vbrowse)
- Serves both research and clinical genomics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

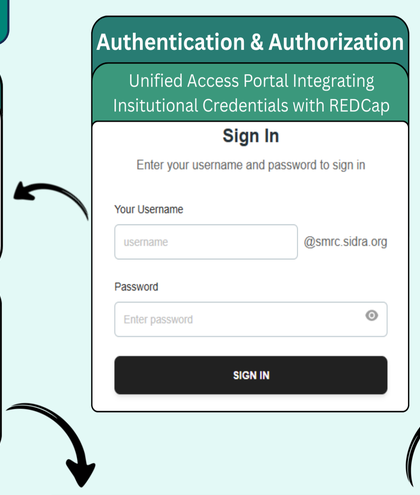


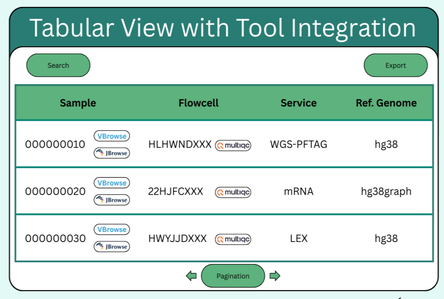
Dashboard

Overview of Samples' Metadata & Instrument Performance Across Multiple Platforms



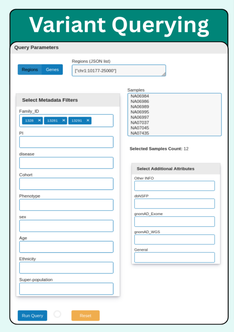


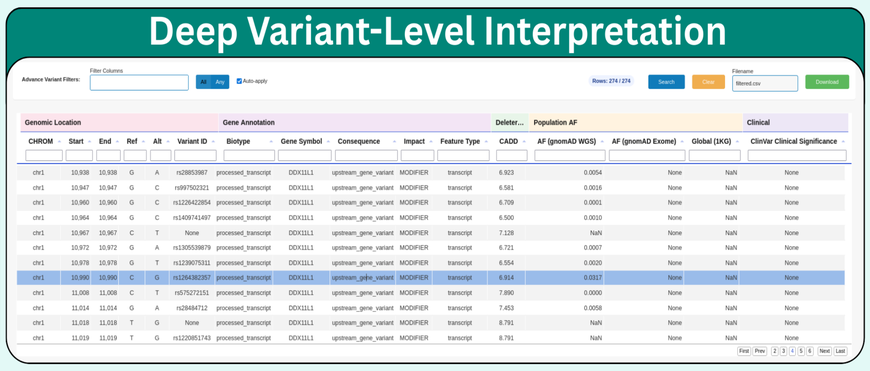


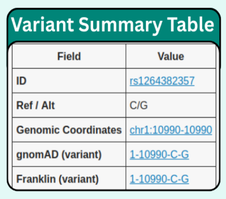


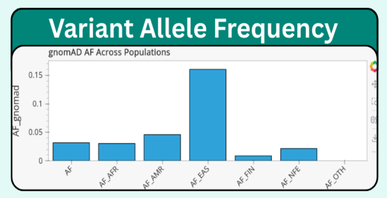
VBrowse

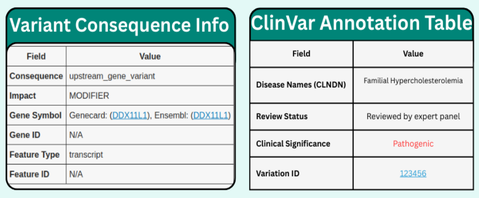
End-to-End Variant Exploration & Interpretation in One Interface





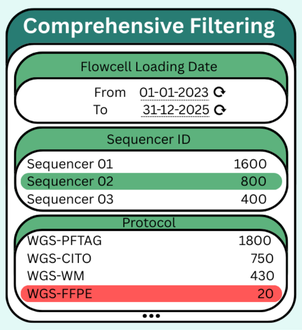


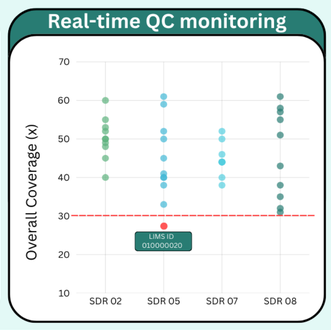


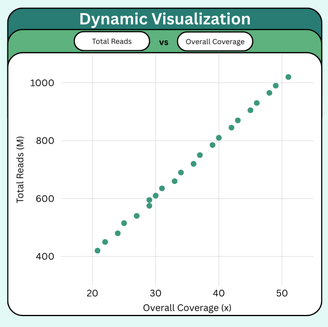


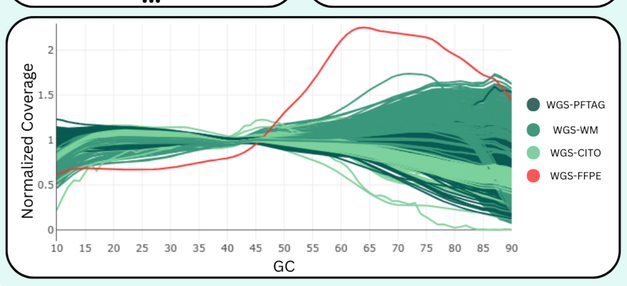
Quality Control Metrics

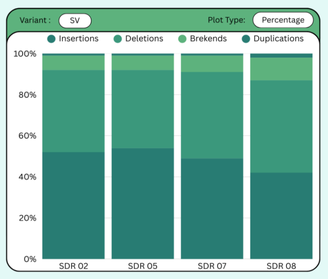
Extensive Assessment of DRAGEN Mapping QC and Variant Calling QC

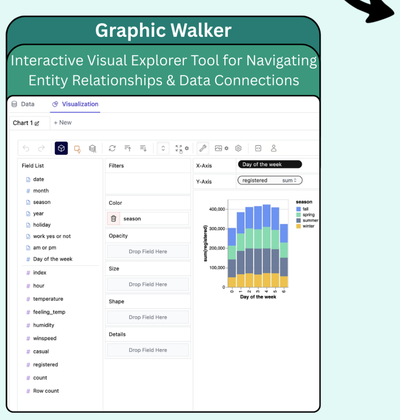






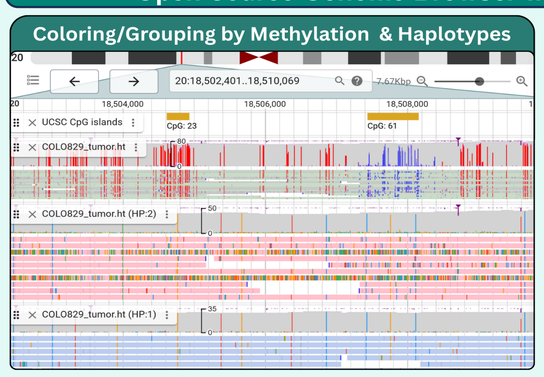


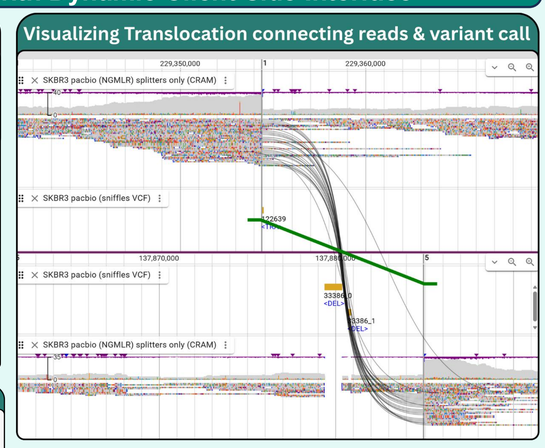


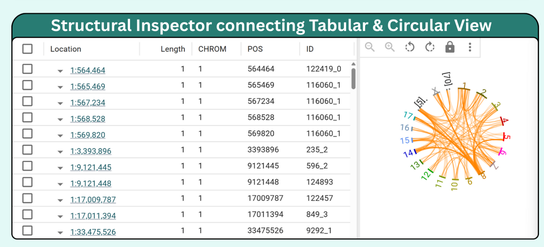


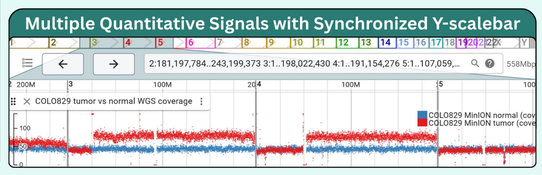
Jbrowse

Open-source Genome Browser with Dynamic Client-side Interface

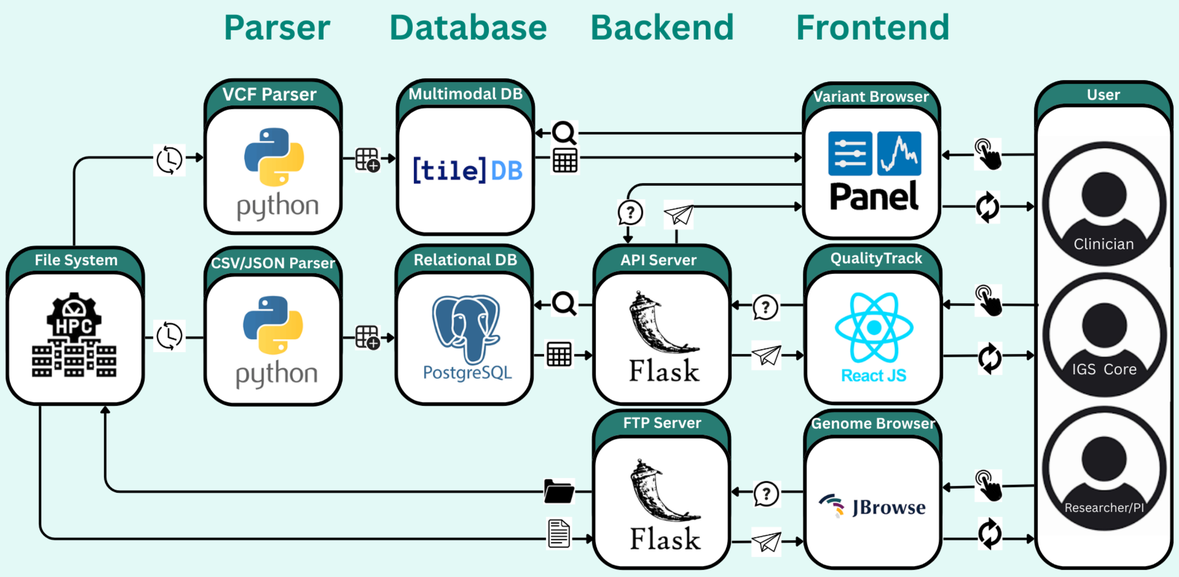








Methods



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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