

GenoVic system: Build and implementation

Background

A core component of the Melbourne Genomics program 2016 to 2019 was to implement a common system for the management of clinical genomic data. The goal was to design and deliver a technology platform that would underpin the genomic testing process – taking data from the sequencing machine, through analysis, interpretation and to a clinical report – and provide secure data storage.

A rigorous and informative procurement was conducted to select two tools: Agilent's Alissa Interpret for variant interpretation and classification, and DNAnexus for bioinformatic analysis (see project summary, 'GenoVic system: Selecting the best genomic tools').

Melbourne Genomics' GenoVic team then built GenoVic, which commenced clinical operations in early 2019.

About GenoVic

Developed by Melbourne Genomics Health Alliance for implementation by the Alliance members, GenoVic provides a common system to manage, analyse and interpret genomic information.

The system is a collection of software that supports clinical genomic testing, and a cutting-edge, custom-built tool (GOS – Genomic Orchestration Service) that orchestrates the movement of complex data and information to and from external systems as well as within GenoVic. The solution supports the evolving nature of clinical genomics through a flexible, modular design.

GenoVic is now supporting test delivery at five laboratories (Victorian Clinical Genetics Services, The Royal Melbourne Hospital, Monash Health, the Australian Genome Research Facility and one laboratory external to the Alliance). The GenoVic team will continue to onboard laboratories in 2020.

Project description

The objective: to fully support clinical genomic testing by linking together the selected vendor products and integrating these with existing systems in the Melbourne Genomics members.

GenoVic's design is modular and components were delivered incrementally. This approach ensured that the system's individual components were fit-for-purpose and that benefits could be realised as early as possible.

In three releases of work (over the period 2016 to 2018), the GenoVic project team enabled multiple Melbourne Genomics members to deliver clinical genomic tests using a single system, specifically:

- Vendor-supported tools for bioinformatic analysis and variant classification and interpretation
- A purpose-built cloud-based tool, the Genomic Orchestration Service (GOS), which conducts the movement of data and information to and from external systems as well as within GenoVic
- A central cloud-based storage location for the clinical genomic data held in GenoVic

Each of the three releases were scoped to ensure that resulting business changes could be readily absorbed by the Melbourne Genomics members, in line with organisational and technical readiness.

All 10 Melbourne Genomics Health Alliance members were involved: The Royal Melbourne Hospital, The Royal Children's Hospital, The University of Melbourne, WEHI, Murdoch Children's Research Institute,

CSIRO, the Australian Genome Research Facility, Peter MacCallum Cancer Centre, Austin Health and Monash Health.

Activities

Over the period 2016 to 2019, the GenoVic project team:

- Built two shared bioinformatics pipelines, which were tested and deployed on DNAnexus
- Collaborated with the vendor, Agilent, to deliver significant updates to the Alissa Interpret tool in order to better meet Melbourne Genomics laboratories' requirements
- Developed and deployed the Genomic Orchestration Service (GOS) in Amazon Web Services (AWS)
- Developed a GenoVic web portal to enable access to key information flows, clinical reports and to place orders into Alissa for laboratories
- Onboarded four member laboratories to GenoVic, integrating the platform with their respective systems as needed
- Integrated a laboratory information management system (LIMS) with GOS

Outcomes

GenoVic's modular structure enables rapid innovation, and was specifically designed to meet the evolving needs of clinical genomics. Components can readily be 'swapped in' or 'swapped out', as the landscape of genomics changes, and to meet the differing needs of the Melbourne Genomics members.

The development of GOS enabled a shared genomic system across multiple organisations in Victoria, delivering a standardised, integrated and interoperable system that supports better care for patients.

Central to GOS is a common model for cloud-based storage and use of genomic data, leveraging the latest in cloud-based computing technology. GOS operates on demand – activating within milliseconds to perform a task and then deactivating upon completion. This 'pay-for-use' model provides cost-efficiencies in moving, storing and analysing the large amounts of data generated by genomics.

Further, GOS enables more efficient and appropriate storage capabilities for genomic data – the ongoing flow and ultimate size of which is difficult to predict. Thanks to GOS, GenoVic's storage capabilities range from 'highly available' to 'cold' storage (for files that are not expected to be regularly accessed). This flexible capability means there is no need for organisations to finance hardware that may sit idle.

Founded on common industry standards for storing and interacting with data, GOS facilitates effective and efficient integration with members' (and others') existing and developing systems – such as LIMS and potentially, electronic medical records (EMRs).

Lessons learnt

- Cloud infrastructure is a cost-efficient, scalable and secure mechanism for storing and processing genomic data.
- Implementing emerging technologies presents unprecedented challenges that can lengthen project timelines. Particularly in integration and networking assessments, Melbourne Genomics expended additional effort to develop ways to integrate and transmit the type and volume of data required.

Impact

GenoVic supports laboratories to provide an efficient, effective clinical genomic testing and storage service by underpinning the test process from end to end. Flexibility is a key attribute of the system: organisations can choose to implement some or all of GenoVic's functions, as best supports the genomic testing workflow.

One of the vendors (Agilent) has created a new staff position in Victoria.