Melbourne Genomics
Health Alliance

Global knowledge. Individual care.

GenoVic system: Enhancement via integration

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.

Following a rigorous and informative procurement process, Melbourne Genomics built the GenoVic system, which commenced operations in early 2019[1].

GenoVic is a one-of-a-kind system that supports clinical genomic testing from end-to-end, as well as storing genomic data. 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.

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.

The custom-built Genomic Orchestration Service (GOS) links together tools supporting bioinformatic analysis, variant interpretation and clinical reporting, data storage and management. GenoVic uses industry-standard connectors and protocols (e.g. APIs), which streamline the integration of new applications.

Project description and activities

The objective: to integrate specialised applications to further improve GenoVic's ability to support testing and management of genomic data by Melbourne Genomics member laboratories.

Three specialised applications were integrated into GenoVic during 2019: Shariant, PanelApp and Cromwell.

Shariant

An Australian Genomics initiative, Shariant, is a system for real-time sharing of detailed scientific evidence about clinically interpreted variants, to reach consensus between laboratories and clinical services. The rapid exchange of this key information enables healthcare professionals to more consistently decode the human genome.

GenoVic is now integrated with Shariant. The Royal Melbourne Hospital (RMH) and the Victorian Clinical Genetics Service (VCGS, Murdoch Children's Research Institute) are now making significant contribution nationally, by sharing curated variants into Shariant in a controlled manner.

PanelApp

A Genomics England system, PanelApp, is a publicly available knowledge-base in which lists of genes related to human disorders can be created, stored and queried. PanelApp also includes a crowdsourcing

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[1] See project summaries, 'GenoVic system: Selecting the best genomic tools' and 'GenoVic system: Build and implementation'.

capability that enables genes and gene variants to be added or reviewed by experts worldwide. PanelApp provides the opportunity for standardisation of gene panels by reaching consensus on which genes have sufficient evidence for disease association.

Melbourne Genomics has integrated the Australian Genomics instance of PanelApp into GenoVic. Laboratories are able to use a custom-made portal that enables them to synchronise their gene lists (in GenoVic) to the nationally curated lists within PanelApp. This portal is being actively used by the RMH and VCGS.

Cromwell

Cromwell is an open source bioinformatics tool developed by the Broad Institute of Harvard University and MIT. It is a workflow-execution engine that simplifies the orchestration of computing tasks needed for genomic analysis.

Melbourne Genomics' investigations led to Cromwell being able to run directly on an Amazon Web Services (AWS, cloud-computing) environment[2]. This has given GenoVic users more flexibility in scaling their genomic workflows. For instance, RMH is using Cromwell to automate and enhance its quality control capabilities in GenoVic.

Outcomes

GenoVic has been proven to be a modular system capable of integrating with specialist tools.

These enhancements have provided tangible and immediate means to enable sharing of knowledge across Melbourne Genomics members, nationally and internationally – delivering additional, unexpected benefits to GenoVic users and enabling better patient care.

GenoVic has met member needs and has sufficient flexibility to provide enhanced capability as required.

Lessons learnt

- GenoVic's modular structure enables innovation and sustainability, as it can continue to evolve and meet member requirements as the genomics landscape changes.

- The Genomic Orchestration Service is fundamental to GenoVic's modularity.

- Standard is better than bespoke: the strength of using industry-standard connectors and protocols (e.g. APIs) in GenoVic was shown by the relative ease of these integrations.

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[2] See separate project summary, 'Innovation in clinical bioinformatics'.