Melbourne Genomics
Health Alliance

Global knowledge. Individual care.

GenoVic system: Selecting the best genomic tools

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. It was envisaged that this system would support genomic testing and, in doing so, support the integration of genomic medicine into routine clinical care.

The ultimate goal was a technology platform that would enable teams to work together across the Alliance to deliver high-quality clinical genomic services to patients.

In 2016, a vendor with expertise in health IT was engaged to work with the Melbourne Genomics members and the project team to develop requirements and propose a future-state solution for a shared clinical system for genomics. The requirements and solution architecture provided a basis for the process to select, procure and implement the core components of a system that would come to be known as 'GenoVic'.

The GenoVic team worked with a number of vendors and key stakeholders within the Melbourne Genomics members to develop, test, enhance and implement an innovative, modular, cloud-based system[1].

About GenoVic

Developed by Melbourne Genomics Health Alliance for implementation by the Alliance members, GenoVic provides a common system to store, manage, analyse and interpret genomic information. GenoVic underpins the genomic testing process – taking data off the sequencing machine, through analysis, interpretation and to a clinical report – and provides secure data storage. 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 comprised of 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 clincial 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 of this project was to procure the components of GenoVic. Procurement was consistent with guidelines of the host organisation (WEHI) and conducted in three stages:

- An expression of interest (EOI) to the open market, in order to identify suitable vendors and products

[1] See project summaries, 'GenoVic system: Build and implementation', 'GenoVic system: Enhancement via integration' and 'Governance of genomic data'.

- An ‘open dialogue’ process to understand how these vendor offerings matched Melbourne Genomics’ requirements and shortlist vendors
- A hands-on pilot to understand which tool would best support the complex and important process of variant interpretation and classification.

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

In August 2016, Melbourne Genomics approached the open market with an expression of interest. The EOI scoped a genomic data repository and diagnostic tools – including a bioinformatic analysis tool with the ability to run pipelines and a variant curation (variant interpretation) tool with the ability to interpret gene variants.

As a result of the EOI, 16 compliant responses were received: 10 vendors offering end-to-end, fully integrated solutions, and six supplying component offerings. An evaluation panel comprising key users from across the Alliance evaluated the EOI responses. As a result, seven vendors were invited to participate in the next phase.

The open dialogue phase (November 2016) saw each shortlisted vendor participate in three interactive, two-way discussion workshops. These workshops provided detailed insights into vendor offerings and proposed delivery approach. The evaluation panel rated each supplier after each workshop.

This process identified one suitable bioinformatic analysis tool (DNAnexus) and narrowed the field to two variant interpretation and classification tools that largely met the Alliance’s requirements. It was agreed that in order to select between these two tools, hands-on use by relevant experts was required.

A hands-on evaluation was designed using structured scenarios, enabling laboratory experts from across the Melbourne Genomics members to use the two tools, then fairly and transparently assess their features. Nineteen expert evaluators worked through workflows for 29 real-world scenarios, scoring the tools for functionality and useability.

Agilent’s Alissa Interpret was selected as the preferred variant interpretation and classification tool. However, there were critical features required by Melbourne Genomics experts that neither curation tool possessed. In order to maximise the implementation, it was agreed with Agilent that these features would be developed and incorporated into the product as an additional ‘statement of work’.

Lessons learnt

- The open (competitive) dialogue is a valuable procurement process to gain a deeper understanding of vendor product offerings than a traditional tender process allows.
- Hands-on experience through the pilot enabled laboratory scientists to feel confident in the variant classification and interpretation tool selected through the process.
- The depth of expert involvement from across the Melbourne Genomics members during procurement proved instrumental in ensuring commitment of the time and resources needed to then successfully implement and configure the selected tools.
- The extra time invested in piloting the shortlisted variant classification and interpretation tools was beneficial in reducing the change management needed for adoption by laboratories.
- Collaborating with vendors resulted in a product that better met members’ needs, offering an overall better outcome for everyone involved.
- Through collaborating with international vendors, it became clear that the clinical genomics work being undertaken by the Melbourne Genomics members is leading-edge, and that there was no ‘out-of-the-box’ information system available to support this work.

Impact

The GenoVic system was built and commenced in clinical operations[2] in early 2019.

DNAnexus and Alissa Interpret were implemented as part of GenoVic, with The Royal Melbourne Hospital, Victorian Clinical Genetics Services and Monash Health using the variant classification and interpretation tool.

Requirements set by Melbourne Genomics drove the successful co-development of Alissa Interpret, offering major enhancements to workflow management, interpretation support and traceability, which are now available internationally as Alissa Interpret v5.2.

[2] 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).