Melbourne Genomics
Health Alliance

Global knowledge. Individual care.

Prototype systems for genomic data

Background

The delivery of high-quality genomic medicine depends on ethical, secure access to genomic information for clinicians and patients.

Melbourne Genomics Health Alliance has broken new ground in demonstrating that multiple organisations can adopt and share approaches at each point in the patient care pathway – including infrastructure, software, policies, procedures and agreements. Prototype data and technology systems were developed to underpin initial delivery of genomic testing across multiple clinical laboratories.

These prototype systems for genomic analysis and interpretation were used by four laboratories to support clinical testing for almost all the genetic disease Clinical Flagships[1] in the Melbourne Genomics 2016 to 2019 program.

Publication

"Cpipe: a shared variant detection pipeline designed for diagnostic settings", Sadedin, S.P., Dashnow, H., James, P.A., Bahlo, M., Bauer, D.C., Lonie, A., Lunke, S., Macciocca, I., Ross, J.P., Siemering, K.R., Stark, Z., White, S.M., Melbourne Genomics Health Alliance, Taylor, G., Gaff, C., OshlacK, A., Thorne, N.P., *Genome Medicine* (2015) doi.org/10.1186/s13073-015-0191-x

Project description

The Melbourne Genomics prototype system was comprised of three main components:

- Leiden Open source Variation Database (LOVD+) – a variant interpretation and classification system. LOVD+ had the advantage of being widely used, customisable, easy to install and maintain, and had good security. Through collaboration with the LOVD+ developers, significant modifications were made to support the genomic variant interpretation process.
- Cpipe – an open source tool designed to provide fast, flexible, customisable, effective and reproducible genomic analysis.
- A custom-built web tool to enable data access and sharing (see separate project summary, ‘Data access and sharing’).

Activities

The Melbourne Genomics data and technology team designed and implemented prototype systems to support genomic testing in the Clinical Flagships. These activities were informed by a multidisciplinary group of users from the Melbourne Genomics members.

Key activities were:

- Genomic analysis ‘pipelines’ built in Cpipe
- Existing manual spreadsheets in use in member laboratories were replaced by LOVD+, to support variant interpretation and classification

[1] See project summary ‘Evaluating clinical utility of genomics: Overview’, as well as individual Clinical Flagship summaries for results.

- A data sharing prototype system, DASh, was built and implemented[2], to enable semi-automated applications for secure and ethical access to patient-level, consented genomic data

Using the prototype tools helped the Alliance define requirements for the development of the GenoVic system.

Lessons learnt

- Legacy tools in place in the Melbourne Genomics member organisations supported genomic testing and identified what was needed for a sustainable shared system.
- While LOVD+ met short-term needs for clinical testing in the Flagships, it did not prove scalable in the long term to support the Alliance's needs for clinical genomic testing.
- Hands-on experience from developing and using prototype systems was invaluable for procuring and building an enduring system.

Impact

Establishing prototype systems was crucial in building the capability of member hospitals and laboratories in delivering a shared system to support genomic testing within a clinical setting.

Working with Cpipe and LOVD+ enabled the Melbourne Genomics project team and the member organisations to better understand and articulate the requirements for procuring and building a shared system for genomic data – GenoVic[3].

The maturity level of Melbourne Genomics member laboratories grew across data governance, program governance, software development practices and software operations – readying laboratory experts for future implementation.

[2] See project summary, 'Data access and sharing'.
[3] See project summaries, 'GenoVic system: Selecting the best genomic tools' and 'GenoVic system: Build and implementation'.