Melbourne Genomics
Health Alliance

Global knowledge. Individual care.

Variant interpretation online training modules

Background

The Melbourne Genomics Health Alliance members are at the forefront of introducing genomics into diagnostic laboratory and clinical practice. From the outset it was clear there was an immediate need to instil literacy, skills and confidence in using genomics in healthcare.

'Variant interpretation' (also called 'variant curation') is the complex process of determining which DNA change (variant) is causing a patient's condition, or – in the case of cancer – is driving cancer growth and may be treatable. An internationally acknowledged 'bottle-neck' in the genomic sequencing process, variant interpretation is still largely a task for expert human minds (not computers).

A multidisciplinary team – including clinical geneticists, medical scientists, bioinformaticians, genetic counsellors and other medical specialists – need to work together to interpret and agree a patient's result.

Melbourne Genomics has been offering continuing professional development workshops in variant interpretation, but an approach that can be delivered more widely and as needed is required.

Project description and activities

The objective: a workforce competent in the interpretation, classification and reporting of variants, using an education format (or modality) that can be delivered as and where required.

A total of 10 online modules were developed from 2018 to 2019. A further seven online modules including practical exercises and cases, welcome video and a workshop preparation module were developed in early 2020.

Online modules were developed from workshop content[1]. Learning objectives and content were designed by the Melbourne Genomics education team, working with scientific and clinical leads at The Royal Melbourne Hospital, the Murdoch Children's Research Institute (Victorian Clinical Genetics Services) and the Peter MacCallum Cancer Centre.

The content covers introductory concepts and skills in variant interpretation, making the modules suitable for training a range of professions in addition to medical scientists.

The training modules developed are:

- Genetics theory for variant curation
- Introduction to variant curation
- Introduction to clinical bioinformatics
- Overview of clinical bioinformatics
- Introduction to variant QC using IGV
- Overview of variant curation
- Introduction to a curation tools and whole sample QC
- Introduction to variant filtering and rapid phenotype matching

[1] See project summary, 'Variant interpretation training through workshops'.

- Introduction to transcript selection and variant annotation checking
- Introduction to analysing variant allele frequency using gnomAD
- Introduction to practical workshops
- Practical exercises on sample and variant QC, variant nomenclature and population frequency
- Practical exercises on *in silico* scores, conservation and constraint
- Detailed solutions for four case studies

Outcomes

Training modules are available online (since August 2019), via an instance of Androgogic's Totara Learning Management System – with registrations currently administered by Melbourne Genomics.

This project has increased the efficiency in teaching variant interpretation, by reducing the need for face-to-face workshops. This alleviates demand on subject matter experts and facilitators.

A broader benefit of these modules is improved consistency in variant interpretation knowledge and skills across a range of professions in Victoria.

Impact

Melbourne Genomics member laboratories are now using the online modules for in-house training in variant interpretation (all modules are available). At the time of writing (June 2020), 37 medical scientists had accessed this material.

The modules have been incorporated into blended learning subjects within The University of Melbourne's Master of Genomics and Health[2].

Along with other Melbourne Genomics training materials, the modules were adapted for The University of Melbourne's Master of Genetic Counselling 'Advanced Clinical Genomics II' subject, with a particular focus on clinical applications of variant interpretation for genetic counsellors.

Lessons learnt

- To ensure the currency and enduring use of online variant interpretation teaching materials, consideration needs to be given to the resourcing required to maintain content, as tools and public databases are rapidly updated.
- Use of the online modules in university subjects has demonstrated that they support learning in variant interpretation. Students reported that the modules helped them prepare for, and engage with, workshops[3].
- Modules need to be 'agnostic' to the use of particular variant interpretation tools (i.e. teaching the underlying concepts, not how to use specific tools or databases), in order to reduce technical and resourcing dependencies.

[2] See separate project summary, 'Postgraduate training in variant interpretation'.
[3] See separate project summary, 'Postgraduate training in variant interpretation'.

Project team

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