

Variant interpretation training for medical scientists

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).

Medical scientists use classification schemes, scientific literature and other reference databases to interpret likely genomic variants. Then a multidisciplinary team – including clinical geneticists, medical scientists, bioinformaticians, genetic counsellors and other medical specialists – works together to interpret and agree a patient's result.

Project description and activities

The objective: diagnostic laboratory staff competent in the interpretation, classification and reporting of variants.

The purpose of this initiative was to train medical scientists in variant interpretation. Training positions were funded for 12 weeks / 48 days per trainee¹ for medical scientists within the Melbourne Genomics member organisations.

Ten medical scientists from six organisations were trained in variant interpretation, with a focus on either rare disease variants (training through the Victorian Clinical Genetics Services) or cancer variants (training through Peter MacCallum Cancer Centre).

The Melbourne Genomics members involved were: the Australian Genome Research Facility, the Peter MacCallum Cancer Centre, Monash Health, Murdoch Children's Research Institute (Victorian Clinical Genetics Services), The Royal Melbourne Hospital and Austin Health.

Evaluation interviews were conducted before and after training to better understand process and impact.

¹ This included funds to backfill the medical scientist's usual duties while they undertook training.

Outcomes

This project has enhanced the variant interpretation capability and capacity in laboratories within the Melbourne Genomics Health Alliance (Victoria's leading providers of clinical genomic testing).

Beyond the 10 formal trainees educated, a further 11 medical scientists have so far benefited from in-house variant interpretation training initiatives leveraged from enduring materials and expertise created.

Moreover, the project has produced medical scientists who are experienced in variant interpretation training and who can continue to perform this role, acting as leaders of rapid and sustained change in their workplaces and more broadly across their professional disciplines.

Broader benefits include:

- Enhanced communication and collaboration between laboratories
- Improved understanding of the key attributes required for the role of variant interpretation

Impact

This project has improved consistency in variant interpretation in Victorian laboratories.

The project has also provided insight into the prerequisite knowledge needed prior to more comprehensive (immersive) training in variant interpretation, and the level of expertise required for a medical scientist to interpret variants with minimal supervision. These are important insights for implementing genomics into regular laboratory service.

The education team also developed learning outcomes in variant interpretation, which can be used for benchmarking and developing competencies for a range of professions.

Lessons learnt

- Training had to be adapted to the background and knowledge of each trainee. Trainees benefited from prior attendance at the two-day workshop on variant interpretation.
- Part-time versus full-time placement did not seem to influence the impact of the learning experience, provided that the training period suited the trainee and trainer. However, part-time trainees experienced added pressure to meet their full-time employment responsibilities during the training period.
- Immersive training is best achieved through both independent and group-based learning, as well as a range of practical training cases complemented by relevant reading material.
- The absence of formal case-based competency assessment was a barrier to tracking trainee progress; this would be required for quality assurance of any future, formal immersive training.
- Immersive training in variant interpretation for medical scientists is required in the future. However, evaluation data did not reveal a consensus on the mechanism for delivering this (options identified include student internships, informal coordination between laboratories, or within existing professional accreditation programs).