Melbourne Genomics
Health Alliance

Global knowledge. Individual care.

Variant interpretation training through workshops

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.

Variants are DNA changes. '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 – is needed to accurately determine the clinical significance of a variant and report the test result. While these multidisciplinary meetings are an important means by which professionals can learn variant interpretation, clinicians and medical scientists identified a need for workshops to educate more professionals working in genomics about the process of variant interpretation.

Project description and activities

The objective: a workforce able to incorporate genomics into patient care, and specifically, able to participate in multidisciplinary variant interpretation.

Continuing education was offered through a series of workshops focusing on either germline (inherited disease) or somatic (cancer) changes in the genome. The workshops were designed for a wide range of health professionals, to create awareness of variant interpretation and underlying concepts in clinical genomics. They were iteratively improved over four years, resulting in two-day workshops of lectures, hands-on activities and small-group case discussions.

Over the four years to 2019, 12 variant interpretation workshops were delivered. This included an Australian-first two-day national workshop, in collaboration with the Queensland Genomics Health Alliance.

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

Attendees were surveyed before, during and after the workshops, to evaluate the team's success in developing attendees' knowledge, skills and confidence in variant interpretation.

Outcomes

A total of 516 health professionals from across the clinical and diagnostic spectrum attended.

A range of professionals benefited, including clinical geneticists, genetic counsellors, medical specialists, bioinformaticians and researchers – this reflects the cross-disciplinary nature of variant interpretation. Medical scientists were the largest group but nevertheless comprised only about a third of all attendees; consultant specialists comprised 20% of attendees.

Participants on average reported a 38% increase in their understanding of variant interpretation, and the majority (89%) reported that they would incorporate these new skills into their role. Case assessments confirmed that participants' competence increased after workshop attendance.

The education team also developed learning outcomes for variant interpretation which is informing an understanding of competencies required across a range of professions.

Impact

The workshops established Victoria as a leader in variant interpretation and genomic education. They attracted national and international interest; workshops were run interstate, in Singapore and New Zealand.

Experience with face-to-face teaching informed the development of online teaching modules (see project summary, 'Variant interpretation online training modules').

Lessons learnt

- Demand for variant interpretation training is high: the germline and somatic workshops were consistently over-subscribed, with all places taken in as little as 24 hours from the release of information.
- Evaluation results indicate that while some aspects of variant interpretation are relatively easy to learn, others require two or more days for participants to gain understanding. These findings were largely consistent between germline and somatic workshops and are being used to inform design of other variant interpretation educational materials.
- Although highly effective, the intensive workshop format is not sustainable due to the level of expertise required across the three types of activities (lectures, hands-on and small-group case discussions): an experienced clinical bioinformatician, medical scientist and clinical geneticist/oncologist across two full days.

Project team

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