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Genomics education for medical specialists: case-based specialty workshops and blended learning

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Abstract

Aim: To develop and evaluate genomics education programs for health professionals to expedite the translation of genomics into healthcare.

Methods: Our co-design team of genetic specialists, expert medical specialist peers, and genomics educators developed two continuing genomics education programs for health professionals: stand-alone, specialty-specific workshops and a generic blended learning course, combining online learning with workshops. Both programs referenced adult learning theories; workshops included case-based learning and expert peer-led discussion. Longitudinal surveys evaluated changes in confidence and understanding of genomic testing processes and clinical practice.

Results: We delivered eleven specialty workshops (414 attendees) and a blended learning course comprising four self-directed online modules (61 users) and workshops (71 attendees) for mixed-specialty groups with adult, pediatric, or oncology cases. Surveys (214 workshops; 63 blended) showed that both programs significantly increased confidence and understanding of genomic testing processes. Blended learning participants showed

additional gains in confidence after attending a workshop following online learning. Workshop discussions with experts were valued, particularly regarding interpreting and applying results. At follow-up, gains in confidence and understanding were maintained for both programs and 81% of respondents had performed a new genomics activity in clinical practice.

Conclusion: Scalable education is needed. Our results suggest that specialty-specific genomics education may not be required to meet the needs of multiple specialties across a health system. Online learning can meet foundational learning needs but may not be sufficient to apply learning to practice. Blended learning offers flexible, continuing education pathways for dispersed national audiences as genomics becomes increasingly used across varied specialties.

Keywords: Workforce, genomic medicine, physician, continuing education, professional development, evaluation, case-based learning, blended learning

INTRODUCTION

Incorporating genomics into healthcare requires a health professional workforce that can apply genomic technologies translated from research settings to medicine[1,2]. Medical specialists without genetics training ("non-genetics" specialists) and other health professionals (such as nurses) need genomic literacy, skills and competencies appropriate for their professional role as genomics becomes relevant to the care of their patients[3]. Efforts to ensure graduating medical professionals have fundamental skills in genomic medicine[4,5] do not address the immediate need of practicing medical specialists to develop appropriate genomic skills. Medical professionals report a need for specialty-specific continuing medical education (CME) in genomics[6]. This approach has been adopted by genomics educators globally, with efforts focused on a single clinical area or specialty, such as oncology or primary care[7,8]. However, a specialty-specific approach to address the continuing professional development needs of "non-genetics" specialists may not be feasible across an entire health system.

In Australia, doctors complete a medical degree and then undertake specialty training through a medical college to become a Fellow (Consultant) of that college, e.g., the Royal Australasian College of Physicians. To maintain their medical registration with the national Australian Health Practitioner Regulation Agency, medical practitioners with specialist registration must self-report CME activities. Educational activities can count for up to 60% of the minimum CME requirements established by their college, with activities offered by a range of education providers. The funding and management of public health care delivery is the responsibility of each state or territory[9]. The State Government of Victoria funded the Melbourne Genomics Health Alliance ("Melbourne Genomics") to address barriers to the use of genomics in the Victorian health system. This included identifying and testing approaches to foster the understanding and skills in genomic medicine of practicing non-genetic specialists. At the time of our education programs, access to genomic testing in a clinical setting by non-genetic specialists was largely through clinical implementation research[10,11], with some having access to hospital-funded panel tests, and the role of genetic counselors varied[12].

Melbourne Genomics developed and implemented a multifaceted education strategy. The first phase of the strategy (reported elsewhere[10,13]) was to foster specialty-specific "peer" experts, potential opinion leaders who could lead change within their discipline[14] through experiences in genomics-rich workplaces[3]. Typically, these funded positions included rotation through a clinical genetics service or laboratory, or projects at the point of clinical implementation (clinical service design "flagship" projects). This resource-intensive approach is arguably best used at an early stage of translation when a cadre of early adopters needs

to be developed to support subsequent stages of implementation. To achieve the reach needed across a health system, scalable CME is needed[15]. Therefore, the second facet of our strategy was to co-design and deliver structured continuing education programs for non-genetics medical specialists to increase their understanding of genomic medicine and its application[3].

We developed and implemented two structured CME programs in genomics: face-to-face, stand-alone workshops tailored to specialist groups and a blended learning course with specialty-agnostic online content and adult, pediatric or cancer clinical cases.

Here we describe the development of these two CME programs and present the results of their evaluation. The place of specialty-specific and generalized genomics education to upskilling medical professionals across a health system is considered in light of these results.

METHODS

Program design and delivery

Audience

The target audience for both our genomics education programs were Victorian medical doctors training or qualified in specialties other than medical genetics ("medical specialists"). There were no pre-requisite genomics knowledge or skills. The specialty-specific workshops were designed to meet needs that emerged as the use of genomic testing was being evaluated in clinical care through the Melbourne Genomics program. The blended learning course was informed by the specialty workshops and designed to meet the needs of a broader range of specialists. In each case, educational programs were promoted through member hospitals and existing communication channels, including the Melbourne Genomics newsletter, website, and/or social media accounts. Advertising for specialty workshops was targeted to the relevant medical specialty. The periods of advertising for each program did not overlap; specialists could attend either program.

Theoretical frameworks, design principles and processes

All learning activities were based on theories of adult learning, that is, learners have varied prior knowledge and are self-motivated to learn when content is most relevant to their professional practice[16]. The learning design was case-based learning (CBL), with both programs including small-group discussion and learning from peer experts. The CBL approach is commonly used in medical education to link theory with practice[17]. The blended learning course also incorporated self-directed use of interactive, online resources.

The programs focused on core background knowledge and clinically-relevant practical application. This approach was based on adult learning theory and our experience that applied clinical learning, such as identifying patients suitable for genomic tests and interpreting test results, is more immediately relevant to most non-genetic specialists than technical aspects of genomics, such as test methodology and variant interpretation processes[18,19].

We used a co-design approach to develop both education programs, where the end-users of a "product" collaborate with designers and relevant stakeholders to contribute to the design process[20]. Learning objectives for the specialty workshops were developed by a co-design team that included Melbourne Genomics education staff (TC, FM, and EL), genetic specialists (clinical/medical geneticists, genetic counselors) and non-genetic "peer expert" medical specialists from the relevant specialties, many of whom had gained some expertise in genomics through Melbourne Genomics clinical flagship or immersion activities. The co-design team for the blended learning course comprised the same Melbourne Genomics

staff (TC, FM, and EL) plus a subset of peer experts to represent adult, pediatric and/or somatic (cancer) specialties. These peer experts were an integral part of the co-design process as they were able to identify key concepts to develop understanding and skills in genomic testing relevant to their specialty, with learning objectives worded in a way that was accessible to their peers. To ensure authentic, clinically-relevant learning through cases based on real-world experience, they helped develop clinical cases for the specialty workshops so that learners could apply new knowledge in relevant clinical contexts; for the blended learning course, the peer experts helped select cases to illustrate adult, pediatric or somatic application of genomic testing. They then also facilitated the CBL components of the program (details provided in Supplementary Table 1). As clinical members of the co-design team were not principally educators, the development phase included building skills in facilitating small-group discussion[21], including how to establish and build on learners' prior knowledge through guided questioning and using step-wise explanations.

Program evaluation

Each program was evaluated separately to assess effectiveness; we did not aim to compare the effectiveness of the two programs against each other or a control group. Evaluation questions aimed to assess program objectives as well as inform the design of future effective and scalable education:

1. Do the programs increase confidence, understanding and skills, and impact attitudes to practicing genomic medicine?

2. What is the value of facilitated case-based workshops for participants who have completed self-directed online modules, in addition to any gains in confidence or understanding?

We compared the results of the two program evaluations to inform broader, enduring genomics educational efforts.

In a longitudinal evaluation study design [Figure 1], we deployed surveys at up to four time points, with at least one reminder to complete each survey:

- Baseline: to all registrants, before pre-reading was provided (specialty workshop) or online modules opened (blended learning course)

- Post-online: to blended learning course participants who completed the online modules

- Completion: to all workshop attendees, after the workshop

- Follow-up: to all workshop attendees, at least 15 months later

Survey domains and outcome measures included changes in confidence, understanding, skills and attitudes relating to genomic medicine, common constructs when evaluating continuing genomics education[8] [Supplementary Table 2]. Measures were designed to assess the application of new knowledge and skills within the Australian health system context. Questions were categorical, open-text, or Likert-scale responses. To assess the ability to apply new knowledge, respondents reviewed an excerpt of a genomic test report that identified a variant of uncertain significance (VUS) and answered a question about whether predictive testing could be offered to family members [Supplementary Figure 1]. All surveys are available on request.

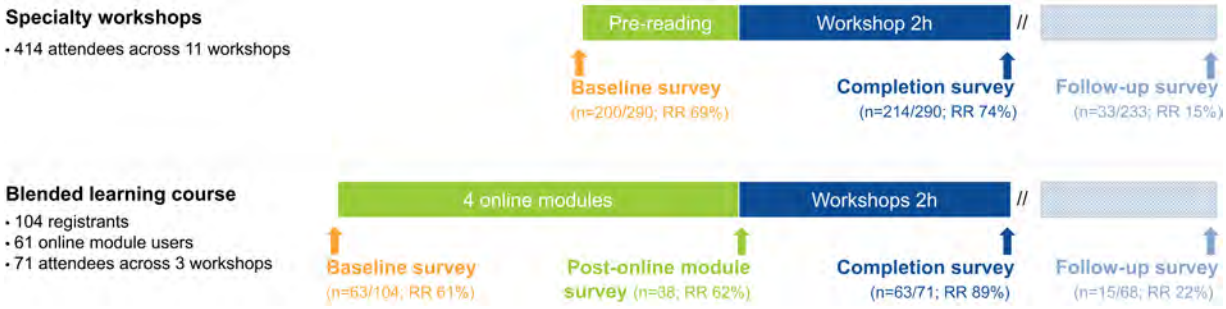

Figure 1. Longitudinal program evaluation. Specialty workshops were conducted over a 15-month period (August 2018-November 2019). The blended learning course (online and workshops) occurred in August 2019. Follow-up surveys were deployed in 2021, with responses for specialty workshops, a median of 23 months (range 16-32) and for blended learning course, a median of 20 months (range 19-20). RR: response rate. Surveys could not be deployed for two neurology workshops, so denominators differ.

Survey data were collected using REDCap[22] electronic data capture tools hosted at MCRI. Data were exported, cleaned, and analyzed using STATA 16.1[23]. All surveys and all questions were optional, so totals differed due to missing data. Percentages are reported to the nearest integer. Data were analyzed separately for each program. To assess self-rated confidence, means were calculated and data were compared between time points within each program using one-sided unpaired t-tests (normally distributed data), or Wilcoxon rank-sum tests (non-normally distributed data). Self-rated understanding of a genomic test report was assessed using chi-squared tests of pooled data at each time point to assess the change in distribution of responses across categories over time. Understanding whether predictive testing can be offered for a VUS was tested using a one-sided proportion test comparing correct with incorrect/unsure proportions across time points. A P value < 0.05 was considered significant. Open-text responses were reviewed by at least two authors both inductively and deductively for themes, then categorized[24].

RESULTS

Educational programs

When developing the learning objectives for each specialty workshop in 2018-2019, the Melbourne Genomics staff observed a common subset of learning objectives that emerged across all specialties. These objectives [Table 1] reflect concepts considered fundamental for all medical specialists to develop basic understanding, skills and confidence in genomic testing, which can then be supplemented by specialty-specific concepts for specialty workshops. The co-design team for the blended learning course deemed these common learning objectives appropriate for the blended learning course. The content of both education programs aligned with these learning objectives and was designed to relate participants' existing knowledge and clinical expertise to new learning of genomic testing relevant to their patients.

Specialty workshops

Eleven specialty workshops were held in 2018-2019 in genomics for germline (heritable) conditions in cardiology, pediatric neurology and development (twice), adult neurology (thrice), pediatric acute care, deafness, bone marrow failure, immunology, and dermatology. These specialties were targeted as they aligned with the local availability of specialty "peer" experts and priorities. Two-hour workshops for up to approximately 40 individuals included optional brief pre-reading material, a short presentation on key concepts, then 3-4 specialty-specific clinical cases presented by a genetic or peer expert, interspersed with facilitated small group case discussion (6-10 people) that addressed the learning objectives [Table 2 and Supplementary Materials].

Table 1. Common learning objectives in clinical genomics

- Identify patients suitable for genomic testing
- Understand the importance of family history
- Recognise the importance of detailed phenotyping
- Discriminate between different genomic tests, e.g., chromosomal microarray, multigene panel, NGS exome or genome sequencing, and select the right test for the patient, based on their purpose (indication), advantages and limitations
- Interpret a genomic test report
- Review the variant classification scheme (ACMG and AMP Guidelines) and the evidence-based foundation of the scheme (pathogenic; likely pathogenic; uncertain significance ("VUS"); likely benign; benign)
- Apply appropriate clinical action based on a genomic test result, e.g., changes to patient management, segregation testing, prenatal screening, or cancer therapy

Table 2. Format of the specialty workshops and blended learning course. All content included text and visuals. Online content also included animations, videos, and knowledge checks. Registrants for both programs also had access to online interactive resources and downloads available at https://learn-genomics.org.au

Content	Specialty workshops	Blended learning course
Foundational genetics and genomics theory	Pre-reading ~30 min • Introduction • DNA, chromosomes, and genes • Reading the code to make proteins • Genomic variants • Genetic and genomic tests	Module 1 ~25 min • *Genomics-Background Biology* e-book, including links to animations and glossary • Video of workshop Introductory presentation • What is genomic testing? • What is the utility of genomic testing? • Downloadable infographic of genetic and genomic testing pipelines
Foundational concepts of genomic medicine	Introductory presentation 20 min • DNA, chromosomes • Genes and proteins • Genetic and genomic tests • Genomic sequencing • Classifying variants • Interpreting reports • Clinical action	
Pre-test aspects		Module 2 ~35 min • Knowing your test options • The right patient • The right test • Pre-test counseling • Ordering genomic tests
Post-test aspects		Module 3 ~25 min • Variant identification, curation, and classification • Interpreting microarray reports • Interpreting exome reports • Post-test counseling
Somatic (cancer) genomics	-	Module 4 ~30 min • Introduction • Pre-test aspects of somatic (cancer) genomics • Variant interpretation (germline) • Variant interpretation (cancer) • Post-test aspects: interpreting somatic (cancer) reports; actionability; genetic counseling
Case study applications of genomic medicine[a]	Specialty-specific 20-30 min/case • 3-4 specialty-specific cases per 2 h workshop, interspersed with expert-facilitated small group discussions	Generic ~30 min/case • 4 cases per 2 h workshop, interspersed with expert-facilitated small group discussions

[a]More details of the case presentations are provided in Supplementary Materials.

Blended learning course

The course commenced with four online modules that covered foundational biology and genetics, and principles of genomic testing and variant interpretation in greater detail than that provided in the specialty workshop pre-reading [Table 2]. The modules addressed both germline and somatic (cancer) genomic testing. Registrants were encouraged to complete online modules over a four-week period (August 2019), with face-to-face workshops held after two weeks. Registrants could choose to attend one or two of three two-hour workshops. Each consisted entirely of CBL using "generic" cases designed to be accessible to a range of specialists while illustrating the principles of genomic testing: germline pediatric genomics, germline adult genomics, and/or somatic (cancer) genomics.

Education participants and evaluation sample

All education activities reached capacity.

Specialty workshops

A total of 414 health professionals attended a specialty workshop (20-43 per workshop). Respondents to the baseline workshop survey (n = 200) were mainly Consultants (48%) or Trainees (37%) in each specialty [Supplementary Table 3]. Two-thirds (67%) reported having no formal genetics training; those who did cite lectures at university (65%) or during basic medical training (26%). Over two-thirds (69%) had ordered or interpreted genetic or genomic tests in their practice, most often ordering single gene (78%) or chromosome tests (66%), with 46% ordering an exome/genome test. Of those who had not previously used genetic or genomic testing, 70% anticipated doing so in the future.

Workshop participants also responded to surveys at the completion of education (n = 214) and follow-up (n = 33; Figure 1).

Blended learning course

One hundred and four health professionals registered for the blended learning course, with all 86 engaging with at least one part of the program. Sixty-one registrants accessed the online modules, averaging 26 min per module; 29 participants completed all four modules. Seventy-one registrants attended workshops (pediatric germline, n = 32; adult germline, n = 23; cancer, n = 27), with 11 of those attending two workshops (pediatric plus cancer; adult plus cancer). Those who completed baseline surveys (n = 63) were mainly Consultants (52%) and Trainees (46%) from varied specialties [Supplementary Table 3]. In contrast to specialty workshop attendees, most (98%) blended learning course participants reported no formal genetics training. Approximately two-thirds (68%) had previously used genetic or genomic testing in their practice, most often ordering single gene tests (74%) or multigene panel tests (74%), and 33% had ordered exome/genome tests. 80% of those who had not previously used genomic testing anticipated doing so in the future.

Blended learning course participants also responded to surveys after accessing the online modules (post-online, n = 38), completion of education program (n = 63), and follow-up (n = 15; Figure 1). As paired survey responses were only completed by 29 participants, unpaired analyses were used. (Paired survey data are provided in Supplementary Figure 2 and showed the same trends as unmatched responses, suggesting that our findings may reflect individual gains.)

Impact of the education programs on confidence, understanding, skills and attitudes

Confidence in understanding and following genomic processes

Respondents rated their confidence in the understanding of five aspects of genomic testing processes [Figure 2]. Confidence increased from baseline in all aspects after both the specialty workshop and blended learning course. For specialty workshops, confidence increased from baseline to completion ($P < 0.001$ for all processes). For the blended learning course, confidence increased from baseline to post-online ($P < 0.001$ for all processes), with further post-workshop gains in confidence for all processes ($P < 0.02$) except "referral pathways" ($P = 0.057$). At follow-up, mean confidence remained above baseline levels for all processes for both programs (workshops $P < 0.001$; blended $P < 0.02$). Comparing change in confidence from completion to follow-up after specialty workshops, there was no significant decrease in confidence for phenotyping ($P = 0.208$) and referral pathways ($P = 0.280$; Figure 2A). For blended learning, confidence was maintained only for referral pathways ($P = 0.193$; Figure 2B).

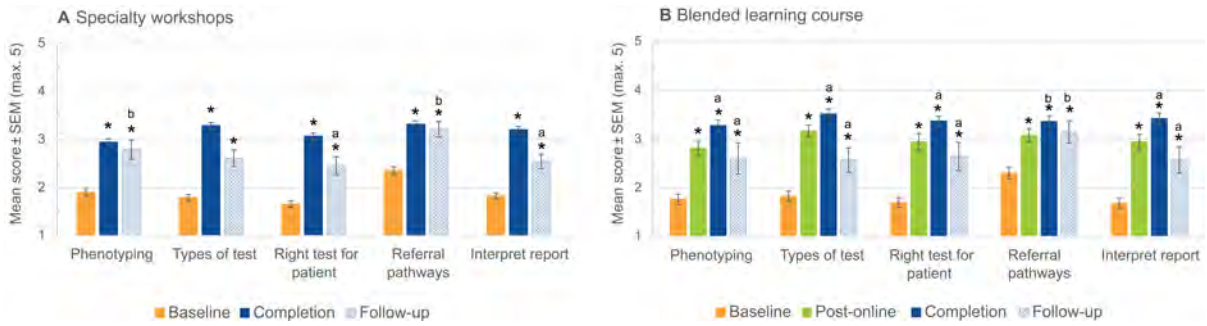

Figure 2. Changes in confidence in genomic processes. (A) Specialty workshops, (B) Blended learning course. 1 = "Needs improvement", 3 = "Good", 5 = "Excellent". SEM: Standard error of the mean. Sample size for each item differs. For specialty workshops, n at baseline, completion, and follow-up, respectively: Phenotyping, 198, 210, 33; Types of test, 198, 211, 33; Right test for patient, 198, 211, 33; Referral pathways, 197, 180, 33; Interpret report, 195, 212, 33. For blended learning course, n at baseline, post-online, completion, and follow-up, respectively: Phenotyping, 63, 38, 62, 15; Types of test, 63, 38, 62, 14; Right test for patient, 63, 38, 62, 14; Referral pathways, 63, 38, 61, 14; Interpret report, 63, 38, 62, 14. *Increased above baseline, $P < 0.02$; [a]increased from previous time point, $P < 0.02$; [b]no significant difference from previous time point (Wilcoxon rank-sum or one-sided t-test as appropriate).

Understanding of genomic testing and skills in interpreting test results

Survey respondents in both education programs showed a marked increase in self-rated understanding of the genomic test report from baseline to completion [Figure 3]. "Good" or higher self-ratings tripled for both specialty workshops (from 28% to 85%; Figure 3A) and blended learning (27% to 87%; Figure 3B), with substantial declines in "Fair" and lower ratings. "Very good" self-ratings increased incrementally through the different components of the blended learning course [Figure 3B]. At follow-up, self-rating of "Good" was maintained for specialty workshop respondents, while self-rating of "Good" or higher declined overall compared to completion (85% to 63% workshops; 87% to 58% blended).

Objective understanding of the clinical implications of a VUS result improved after both programs [Figure 4]. The proportion of respondents that correctly identified that predictive testing cannot be offered based on a VUS finding increased substantially through both programs (workshops 66%, $P < 0.001$, Figure 4A; blended 82%, $P = 0.001$, Figure 4B). Correct responses remained at similar levels at follow-up (72% workshops; 75% blended). However, some respondents remained "Unsure" after program completion (22% workshops; 5% blended).

Changing genomic practice

At program completion, most specialty workshop respondents (90%; 186/206) and all blended learning course respondents (62/62) anticipated incorporating skills into their professional roles:

"*[I can now make a] more informed choice of genetic testing in neuromuscular disease" (Consultant neurologist, blended learning course, completion*)

"*[I can now give] more consideration of the limitations of [genomic] testing" (Consultant pediatrician, blended learning course, completion)*

"*[I now have] confidence to convey information and interpret reports" (Acute care specialist, specialty workshop, completion)*

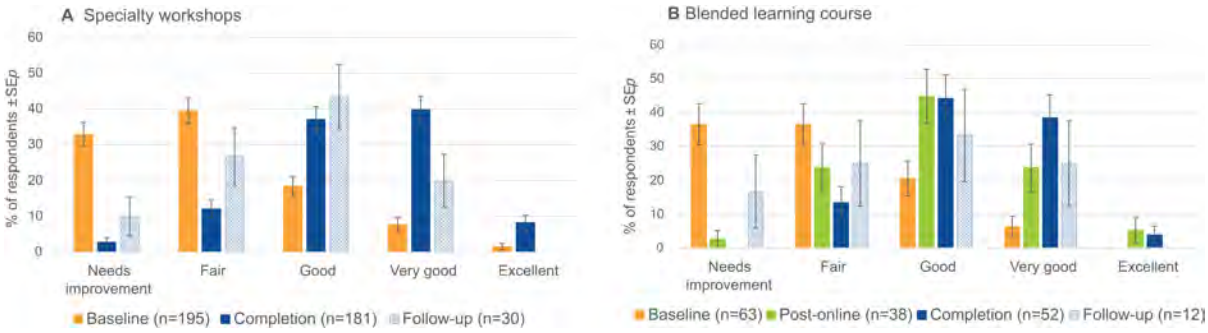

Figure 3. Self-rated understanding of a genomic test report excerpt. (A) Specialty workshops, (B) Blended learning course. Survey question, including a report excerpt with VUS result: "How would you rate your understanding of the information presented in the report excerpt?" Error bars: standard error of the proportion (SE*p*). For both programs, there was a significant difference in the distributions of responses, shifting right from "Needs improvement" towards "Good"/"Very good", from baseline to completion (specialty workshops: $X^2 = 135.35$, df = 4, $P < 0.001$; blended learning course: $X^2 = 46.35$, df = 4, $P < 0.001$), and also from baseline to follow-up for specialty workshops ($X^2 = 17.75$, df = 4, $P = 0.001$) but not for the blended learning course ($X^2 = 6.05$, df = 3, $P = 0.109$).

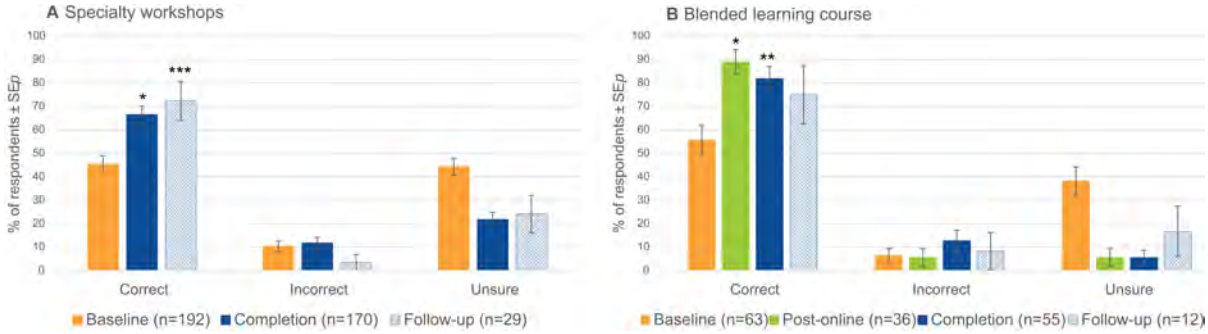

Figure 4. Understanding the implications of a genomic test result of a variant of unknown significance. (A) Specialty workshops, (B) Blended learning course. Survey question, including a report excerpt with VUS result: "Could the referring doctor offer predictive testing in this family?". Error bars: standard error of the proportion (SE*p*). *Increase from baseline (using one-sided proportion test, comparing the proportion correct versus incorrect/unsure combined): *$P < 0.001$, **$P = 0.0012$, ***$P = 0.003$.

Some respondents acknowledged that the education program illuminated gaps in their understanding:

"*I still feel overwhelmed by the amount of knowledge WES [whole exome sequencing] gives and feel I need to improve my knowledge of which panels to focus on. If anything I feel less confident but more aware of gaps to address." (Pediatric neurologist, specialty workshop, completion)*

"*[I have] better knowledge of when to refer to [discuss] with geneticist and interpretation of test results, but still a lot to learn." (Consultant Ophthalmologist, blended learning course, completion)*

An actual change in practice was reported by 81% of respondents to the follow-up surveys ($N = 291$; response rate 16.5%; Table 3); 76% of those who had attended a specialty workshop and 93% of those who had attended the blended course. New activities since attending our education programs included referring patients to a clinical genetics service, consulting a clinical genetics service for advice, requesting exome/genome tests, and educating others about genomics.

Education aims and program feedback

At baseline, respondents were asked what they hoped to gain from completing a workshop or blended

Table 3. New activities undertaken since attending a specialty workshop or blended learning education program[a]

	Specialty workshops	Blended learning course	Total
N	33	15	48
At least one new activity	25 (76%)	14 (93%)	39 (81%)
Type of new activity	**n (% of 25)**	**n (% of 14)**	**n (% of 39)**
Referred patients to a Clinical Genetic Service	15 (60)	12 (86)	27 (69)
Consulted a Clinical Genetic Service for advice	15 (60)	9 (64)	24 (62)
Requested exomes/genomes	9 (36)	8 (57)	17 (44)
Educated others in my discipline about genomics	10 (40)	6 (43)	16 (41)
Requested multigene panel tests	9 (36)	5 (36)	14 (36)
Interpreted multigene panel test reports	10 (40)	4 (29)	14 (36)
Requested single gene tests	7 (28)	4 (29)	11 (28)
Requested single gene tests	7 (28)	4 (29)	11 (28)
Requested chromosome tests	6 (24)	4 (29)	10 (26)
Interpreted exome/genome test reports	5 (20)	4 (29)	9 (23)
Interpreted chromosome reports	5 (20)	3 (21)	8 (21)

[a]If a respondent had attended both a specialty workshop and the blended course (n = 5), they were categorized by the most recent activity. Respondents could select multiple responses, so percentages sum to > 100%.

learning course. The most common responses related to: understanding the clinical utility of genomics in their field; understanding the types, limitations, and appropriate use of genomic tests; ability to interpret genomics test reports; and broader genomics knowledge.

"*[To gain] A better understanding of the genetic testing available, advantages, disadvantages, what some of the latest tests won't pick up etc" (Consultant neonatologist, specialty workshop, baseline)*

"*To gain more confidence in ordering and interpreting relevant genetic tests for my patients and be able to discuss these results with my patients" (Trainee medical oncologist, blended learning course, baseline)*

At completion, most respondents rated the educational activities overall as "Excellent" or "Very good" (87% workshops; 81% blended). Preparatory materials were rated highly, with 91% of specialty workshop respondents and 76% of blended learning course respondents rating these as "Very useful"/"Useful" [Supplementary Table 4]. Case studies were considered the most useful aspect; 99% workshop and 100% blended, rating them "Very useful"/"Useful" [Supplementary Table 4].

When asked to reflect on the value of the different modes of learning, the majority of blended learning respondents indicated a blended approach was more valuable than either online modules or workshops alone (61%-83% across all learning objectives; Figure 5). Respondents considered online modules more valuable than workshops for learning about pre-test consent and counseling (22% *vs.* 12%) and test requests, administration, and referral (25% *vs.* 14%). Workshops were more valued for all other learning objectives, particularly interpreting test reports (29% *vs.* 2%) and post-test counseling (21% *vs.* 7%). The most beneficial aspects of workshops included discussing result interpretation and application with experts.

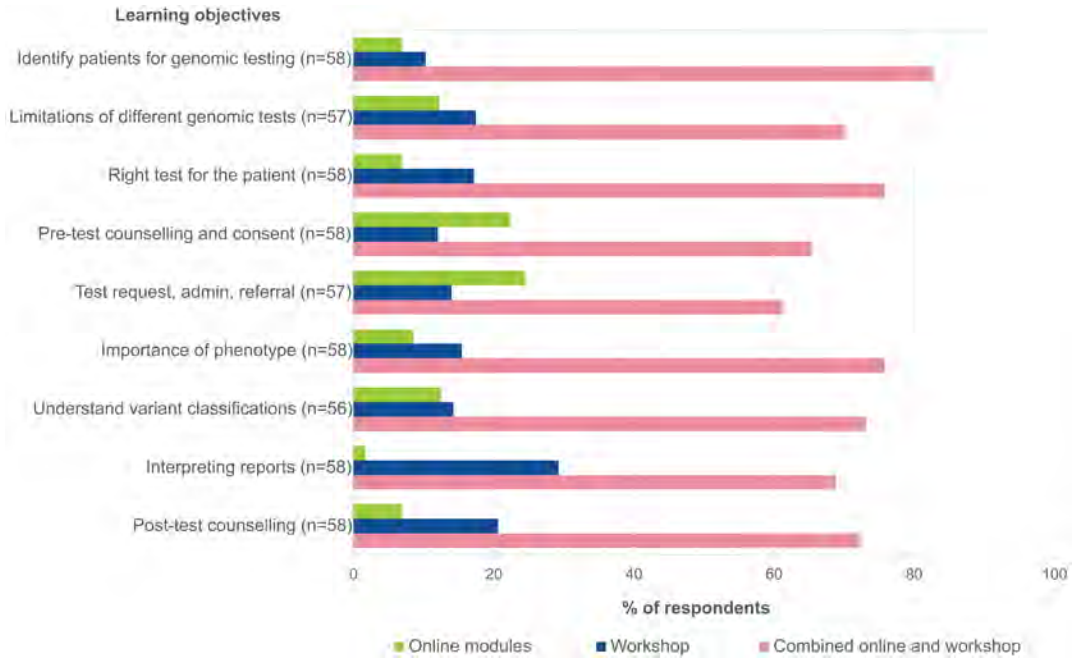

Figure 5. Perceived value of separate or combined components of the blended learning course for each learning objective. Survey question: "Reflecting on both the online content and workshop activities please indicate which component was most helpful in your learning in each of the course objectives (select one option for each objective)".

"*The cases and walking through the reports" (Hematologist, specialty workshop, completion)*

"*Small group discussion with an expert to answer questions and pre-empt discussion after/during each case is very useful." (Consultant cardiologist, blended learning course, completion)*

"*WES explanation interpreting results" (Pediatrician, specialty workshop, completion)*

At program completion, respondents indicated the most valued aspects across both programs were specific content (e.g., understanding genomic tests and appropriate use, interpreting test reports), as well as the CBL format and networking with peer experts.

"*Networking, learning from the best with clinical scenarios, updates in research and clinical management" (Adult neurologist, specialty workshop, follow-up comment)*

The most frequent suggestions for improvement across both programs focused on cases: discussing more cases for longer periods of time or more complex cases.

"*Longer session and more cases" (Neurodevelopmental pediatrician, specialty workshop, completion)*

"*[I would have liked] more complex cases" (Pediatric cardiologist, blended learning course, completion)*

DISCUSSION

Our education programs introduced the principles and processes of genomic medicine to non-genetic medical specialists across a range of disciplines and career levels. Longitudinal evaluation shows real-world changes in practice following our education programs, with an improved understanding of the relevance of genomic medicine, and the types of genomic tests available. The inclusion of non-genetic specialists (peer

experts) in design and delivery was a crucial element of our programs. As a result, we have identified a common set of learning objectives required for "entry level" education, irrespective of medical specialty. These are potentially useful to other education programs, particularly where there are no agreed national standards (e.g., competencies) to guide education for non-genetic medical specialists (e.g., Australia). Our evaluation results provide insights into the place of online learning and workshops as implementation strategies to translate the use of genomics from research settings to health systems.

Medical specialists' preference for CME specific to their specialty and patient group[6] would suggest that education programs need to be designed for a specific discipline. Our results challenge that assumption. Firstly, a common set of core learning objectives could be defined across the specialty-specific workshops, similar to the common "clinical goals" identified across multiple specialties for a continuing genomics education program at a US hospital[25]. Secondly, there was little difference in learner confidence or understanding between survey respondents from our specialty-specific workshops versus generic blended course, with similar ratings on the usefulness of the cases in each program. This suggests that, with an appropriate selection of cases and the inclusion of experts for group facilitation, similar outcomes can be achieved.

There is potentially an inherent tension between continuing genomics education tailored to a specialty and more readily scalable online education approaches to meet the needs of a growing number of many medical specialties across a health system. The reliance on the availability of experts with genomics, specialty, and/or teaching expertise to deliver specialty-specific workshops limits their potential to scale across a health system. However, the step-wise increases observed in the blended learning course respondent data, where participants first completed the online modules and then subsequently attended a workshop, suggest there is an additional benefit in attending a CBL workshop. Workshops can provide additional benefits that support the adoption of genomic medicine by practitioners. Respondents' preferences for the different modes of delivery in our programs suggest that online learning can be satisfactory for education relating to pre-test counseling and test request procedures, which are generally common to all patients. In contrast, workshops may be more suitable for education about post-test counseling, perhaps reflecting the nuance inherent in interpreting and applying the test results for an individual patient. Australian medical specialists prefer a service model of providing genomic medicine with support from clinical genetics services[19]. Multidisciplinary workshops involving genetic specialists as educators can also facilitate the formation of networks with clinical genetics services to foster this support. While workshops are resource intensive, as the practice builds, expert guidance will be increasingly available within the workplace or networks, potentially reducing the need for this approach over time.

Genomics education programs are typically designed and delivered by specialists with up-to-date expertise in genomics[25-28]. A critical success factor for both our genomics education programs was to include non-genetic yet "expert" medical specialists (peer experts), alongside genetic specialists, educators and evaluators. Our peer experts gained specialty-specific genomics experience through periods of immersion in genomics-rich environments and received training in case-based learning before facilitating workshops. Reports of using peer experts to co-design and deliver continuing education are limited, with few relevant to genetics/genomics[15,29-32]. Peer experts view the application of genomic medicine through their discipline-specific lens while also retaining the perspective of a genomics novice[33]. In both our programs, peer experts were crucial in defining learning objectives and content that introduced genomics in a relevant and accessible way to non-genetic medical specialists. Involving genetics specialists as well as peer experts in case-based group discussions at workshops creates an opportunity to establish communication channels between health professionals with different levels of expertise in genomics and to develop new connections

between genetic services and medical specialty services across hospitals. Both these elements support the diffusion of innovation[34]. The extent to which genomics education workshops do support the development of new relationships and thus support the implementation of genomics warrants further investigation.

Assessing the broader impacts of genomics education is a well-recognized challenge[32,35-37]. Evaluation of participants' self-reported changes to practice through follow-up surveys is a common approach[32,38-42]. Survey respondents in our education programs did report undertaking new genomic activities in their real-world practice, including educating others in their discipline about genomics, and patient-related activities. However, we cannot confidently ascertain the magnitude of change to practice. As is common with follow-up surveys, we had a low response rate. In addition, many of our program participants worked in specialties where opportunities to engage with genomic medicine might be limited. If we take a conservative approach and assume all those who made a change responded to the follow-up survey, nearly 15% of participants changed their practice within two years. Audits of clinical practice provide an objective measure of impact[26,41,42]. However, the breadth of hospitals employing participants, diversity of referral pathways, and limited electronic data sources rendered audits following our programs unfeasible. We therefore relied on self-reported behavior change at long-term follow-up.

Although objective measures of application of knowledge following education have been developed for use within a single specialty or setting[25,32,40,43,44], it is challenging to design scenarios and questions that adequately reflect clinical decisions relevant to diverse specialties and contexts, especially in a rapidly-developing field such as genomic medicine. We co-developed a scenario and question to assess the application of genomic knowledge to clinical care to evaluate our workshops. In common with other continuing genomics education programs, we also included subjective measures of evaluation[8,28,29,40,44]. The long-term follow-up survey was deployed during the COVID-19 pandemic. The additional demands on health professionals at this time may have contributed to the lower response rate than the baseline and completion surveys. Survey responses at completion and follow-up may also be biased towards participants who are more confident in their genomic practice.

Education has a key role to play in the implementation and adoption of genomic medicine within a health system. We intentionally describe the theories and design principles referenced during the development of our education programs, and include detailed descriptions and Supplementary Materials for both the education and its evaluation, to provide insights into potentially effective and widely-applicable workforce development strategies[45]. Online modules provide a highly scalable approach to workforce education, including supporting access by health professionals who work in regional towns or remote areas. We hypothesize that wholly online learning may be sufficient preparation for medical specialists to offer genomic testing to patients who meet clear diagnostic criteria, such as hematuria with hearing and vision involvement (Alport syndrome). However, nuanced clinical decision-making is required for pre-testing for complex patient presentations and often post-testing - where test results must be interpreted in the context of the patient's presentation and may influence patient management. Participants in our programs particularly valued workshop discussions with specialty and genetics experts for these aspects. Some respondents also wanted more complex cases, which could be provided as optional program extensions in the future.

Adoption of genomic medicine requires more than just genomic literacy. A key feature of our program was the use of "peer experts" to co-design and deliver education. Peer experts can mediate and "translate" the evidence for the use of genomics in a specialty and adapt clinical genetics practice as appropriate to the specialty; this makes them important mediators of change[33]. Using peer experts in both co-design and

delivery of education, particularly in case discussions, not only builds capability in genomics, but also increases motivation through articulation and illustration of the relevance of genomics to the specialty. Strengthening cross-specialty relationships also provides an opportunity to practice genomic medicine. We would encourage more people to use peer experts purposively in both co-design and delivery of education programs, and explicitly report how they leverage specialist knowledge to bridge gaps between clinical genetics and other medical specialties.

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Authors' contributions

Contributed to all stages of this work and manuscript preparation: Maher F, Lynch E
Made substantial contributions to the design of the work and data interpretation, plus drafting or critically revising the manuscript for important intellectual content: Martyn M, Nisselle A, Gaff C
Contributed to the development of educational materials for the blended learning course and critically reviewed the manuscript: Charles T
Contributed to data cleaning, analysis and manuscript preparation: Tytherleigh R

Availability of data and materials

The surveys and datasets generated during and/or analyzed during the current study are available from the corresponding author upon reasonable request. Some education materials are available at https://learn-genomics.org.au.

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Conflicts of interest

All authors declared that there are no conflicts of interest.

Ethical approval and consent to participate

This study was approved by the Melbourne Health Human Research Ethics Committee (HREC/13/MH/256). All education participants were informed that completing survey questions would be taken as implied consent to participate in the evaluation program.

Consent for publication

All education participants were informed that completing survey questions would be taken as implied consent to participate in the evaluation program.

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