SUPPLEMENTARY MATERIALS

Maher et al. Genomics education for medical specialists: case-based workshops and blended learning

Details of the co-design and delivery process

Case preparation

Each workshop has an expert team of medical specialists, clinical geneticists, genetic counsellors and/or medical scientists with experience of clinical genomic testing. Case presentations are co-developed by a Melbourne Genomics educator and these experts. Referencing the learning objectives considered important for the target audience, clinicians identify potential cases from their own experience, from which 3–4 are selected as most accessible to an 'introductory-level' audience.

Typical features of teaching cases are shown in **Box S1**. A typical case covers the full patient story, from initial presentation to genomic test result (see Case presentation description below).

Box S1. Features of teaching cases

Germline

- cases with VUS and pathogenic variants (variants classified by ACMG guidelines)
- use of microarray for conditions such as childhood syndromes before genomic sequencing
- panel vs exome/genome
- singleton and trio exome testing
- *de novo* variants
- compound heterozygous findings
- suitability of offering predictive testing for family members

Somatic (oncology)

- somatic and germline variants
- actionable vs no approved treatments or trials
- referral to a Familial Cancer Centre

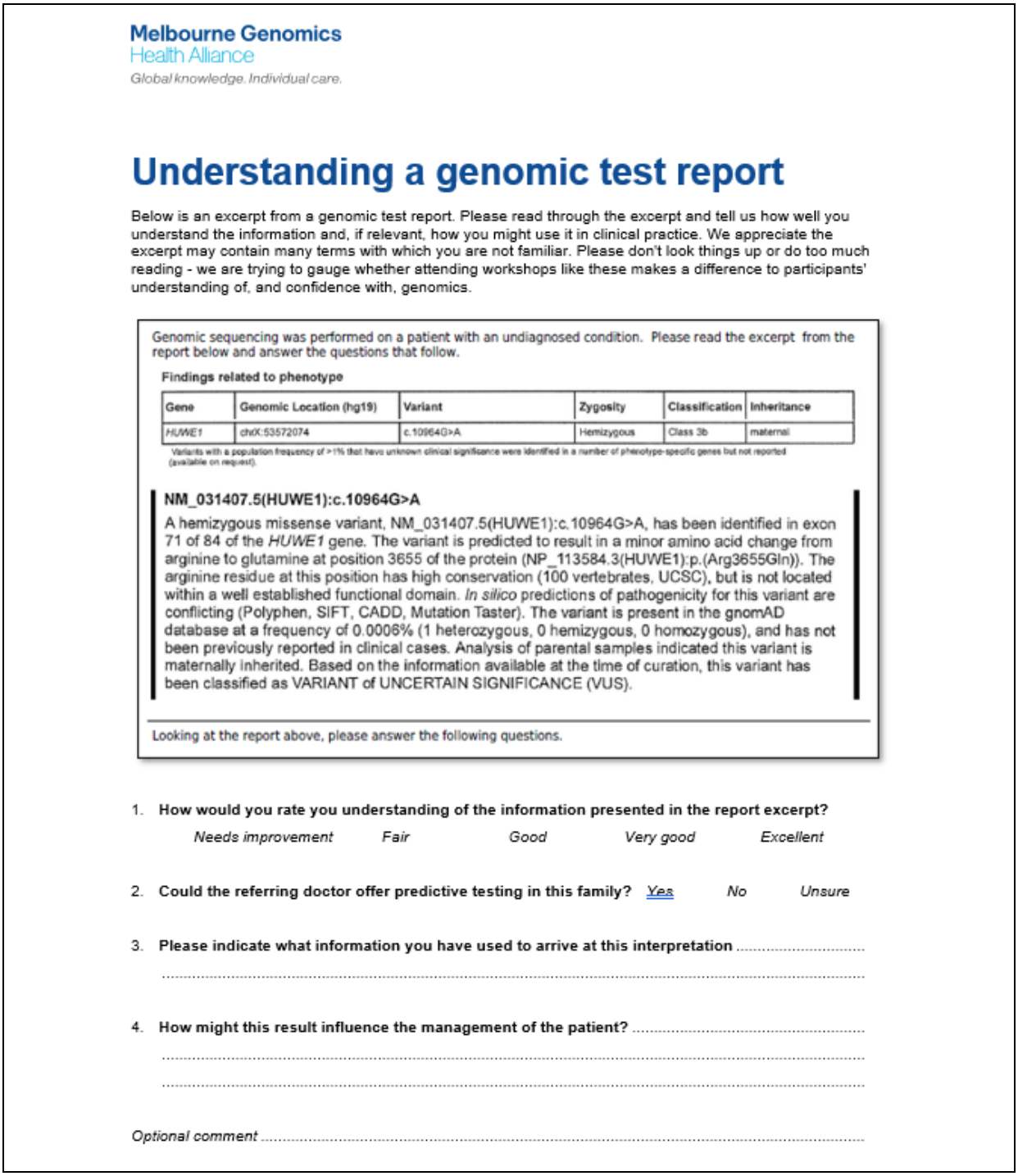
Case presentation

A typical workshop will be 2 hours duration, during which time 3 cases will be discussed for 30–40 minutes each (**Table S1**). Learner engagement is elicited by directed questioning and polls that challenge understanding, and through 1–2 small breakout groups facilitated by genomics experts and peer experts. These 10–15 minute small group discussions focus on key aspects of the case and specific learning objectives and offer opportunity for individuals to ask questions, be they technical aspects of genomic sequencing, ethics or counselling issues, or clinical implications of test results.

Table S1. Typical case presentation process

Step	Audience	Description	Engagement
1	Whole group	The presenter starts with patient clinical history, test history, tests that would/should be done before considering genomic testing	• Includes questions to engage the group e.g. 'Does the pedigree indicate a genetic cause?', 'What would you do next? • Questioning is verbal or via electronic poll (attendees respond via laptop/tablet/smart phone). • Questions and polls also provide feedback for facilitators regarding the level of understanding within the group, e.g. of testing and when to consider genomic testing.
2		Consider the use of genomic testing for the patient:	• Question/poll, 'Which test would be appropriate - select from a list of list of genetic/genomic test options [e.g. single gene test, CMA, multigene panel, exome or genome sequencing]
3	Small group break outs	10–15 min with expert facilitator to focus on a selected feature of the case, e.g., • What different tests detect (re-enforcing the pre-reading) • Reasoning for choice of tests for the patient, e.g., pros and cons of panel or exome sequencing for the patient • Interpret report (the focus for many cases): report details, identify key findings, demystify complex terminology and	• Facilitators have predetermined questions to guide the discussion of key points • Discussions should also draw on participants' clinical knowledge • Attendees receive a copy of the deidentified test report

Step	Audience	Description	Engagement
		concepts, how much variant curation detail is important for physicians to know • When to conduct segregation and cascade testing • Clinical action, change to patient management, offering family testing	
4	Whole group	Quick recap of test result or focus points	• Main message reached by each small group (discussion can get side-tracked)
Repeat Steps 3–4 to for second break out if time permits, focusing on different discussion point			
5	Whole group	Presenter concludes case, including diagnosis, clinical outcomes and patient management	• Discuss the impact of genetic diagnosis, clinical outcomes and patient management



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Understanding a genomic test report

Below is an excerpt from a genomic test report. Please read through the excerpt and tell us how well you understand the information and, if relevant, how you might use it in clinical practice. We appreciate the excerpt may contain many terms with which you are not familiar. Please don't look things up or do too much reading - we are trying to gauge whether attending workshops like these makes a difference to participants' understanding of, and confidence with, genomics.

Genomic sequencing was performed on a patient with an undiagnosed condition. Please read the excerpt from the report below and answer the questions that follow.

Findings related to phenotype

Gene	Genomic Location (hg19)	Variant	Zygosity	Classification	Inheritance
HUWE1	chrX:53572074	c.10964G>A	Hemizygous	Class 3b	maternal

Variants with a population frequency of >1% that have unknown clinical significance were identified in a number of phenotype-specific genes but not reported (available on request).

NM_031407.5(HUWE1):c.10964G>A

A hemizygous missense variant, NM_031407.5(HUWE1):c.10964G>A, has been identified in exon 71 of 84 of the *HUWE1* gene. The variant is predicted to result in a minor amino acid change from arginine to glutamine at position 3655 of the protein (NP_113584.3(HUWE1):p.(Arg3655Gln)). The arginine residue at this position has high conservation (100 vertebrates, UCSC), but is not located within a well established functional domain. *In silico* predictions of pathogenicity for this variant are conflicting (Polyphen, SIFT, CADD, Mutation Taster). The variant is present in the gnomAD database at a frequency of 0.0006% (1 heterozygous, 0 hemizygous, 0 homozygous), and has not been previously reported in clinical cases. Analysis of parental samples indicated this variant is maternally inherited. Based on the information available at the time of curation, this variant has been classified as VARIANT of UNCERTAIN SIGNIFICANCE (VUS).

Looking at the report above, please answer the following questions.

1. **How would you rate you understanding of the information presented in the report excerpt?**
   Needs improvement *Fair* *Good* *Very good* *Excellent*

2. **Could the referring doctor offer predictive testing in this family?** *Yes* *No* *Unsure*

3. **Please indicate what information you have used to arrive at this interpretation** .....
   .....

4. **How might this result influence the management of the patient?** .....
   .....
   .....

Optional comment .....

Figure S1. Subjective and objective measures used to evaluate application of clinical genomic knowledge.

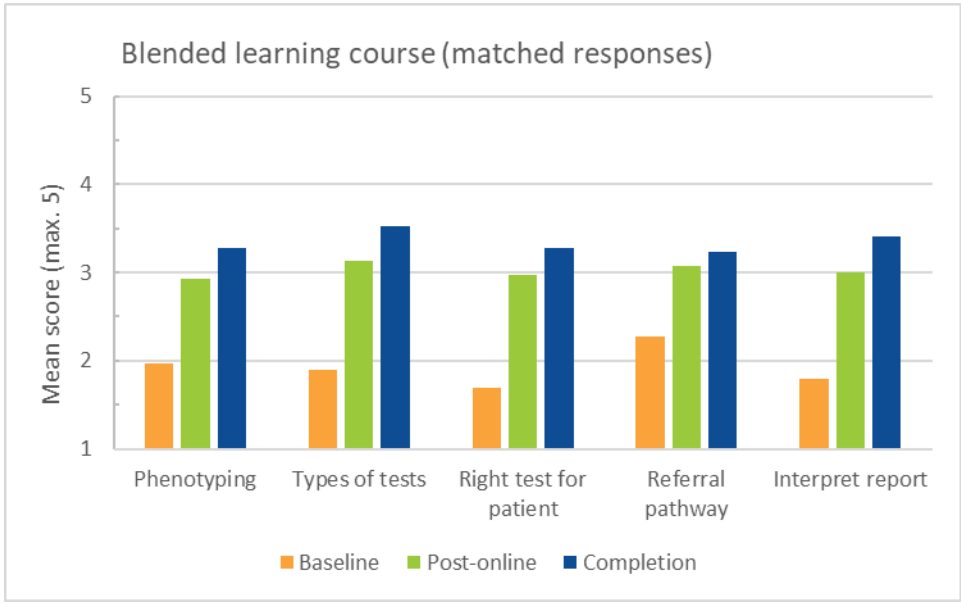

Figure S2. Matched changes in confidence in genomic processes over time for blended learning course survey respondents (n=29).

1='Needs improvement', 3='Good', 5='Excellent'. Participants in the blended learning course completed surveys at Baseline, Post-online and Completion, with 29 completing all three (n=29 matched).

Table S2. Evaluation survey domains

Domain	Specialty workshops			Blended learning course			
	Baseline	**Completion**	**Follow-up**	**Baseline**	**Post-online modules**	**Completion**	**Follow-up**
Demographics	✓			✓			
Aim vs outcome	✓	✓		✓		✓	
Previous genomic medicine practice	✓			✓			
Previous formal genetic education or training	✓			✓			
Understanding/skills	✓	✓	✓	✓	✓	✓	✓
Confidence	✓	✓	✓	✓	✓	✓	✓
Preparedness to practice		✓			✓	✓	
Feedback on workshop/ course							
Structure		✓				✓	
Content/curriculum		✓	✓		✓	✓	✓
Accessing online pre-reading/ modules		✓			✓	✓	
Application to clinical practice (intended vs actual)		✓	✓			✓	✓
Feedback on continuing education preferences (general, modes of learning)		✓				✓	

Table S3a. Baseline evaluation survey sample[a]

	Specialty workshops		**Blended learning course**	
Participants	414 attendees		104 registrants 61 accessed online modules 71 attended workshops	
Description	11 workshops across 8 specialties, with 21-43 attendees per workshop		29 completed all 4 modules 3 workshops across pediatric (n=32 attendees); adult (n=23); cancer (n=27) 11 attended two workshops	
Baseline surveys completed	**N=200**		**N=63**	
	n	**%**	**n**	**%**
Career stage	**197**		**63**	
Consultants[b]	95	48	33	52
Trainees[b]	73	37	29	46
Other	29	15	2	3
Years' experience	**199**		**63**	
0–5	53	27	14	22
6–10	70	35	25	40
11–15	30	15	8	13
16–20	11	6	5	8
>20	35	18	11	17
Previous training in genetics	**199**		**63**	
No	133	67	62	98
Yes (e.g., university lectures, basic physician training)	66	33	1	2
Previously ordered or interpreted genetic or genomic tests[d]	**199**		**63**	
Yes, ordered:	**138**	69	**43**	68
Single gene	107	78	32	74
Chromosome tests	91	66	22	51
Multigene panel	87	63	32	74
Exome/genome tests	64	46	14	33

	Specialty workshops		Blended learning course	
If not previously ordered genetic or genomic tests, anticipate using in future	**71**[e]		**20**	
Yes	50	70[d]	16	80

[a] Totals differ per question due to missing data. [b] 'Consultant' in the Australian medical system is a senior physician or surgeon who has completed all their specialist training and is approved to consult in their specialty. 'Trainee' includes Australian medical training categories of Fellow, Advanced trainee, Trainee, and Registrar (on the specialist training pathway), and more junior medical trainees – Clinical Resident, Intern and Junior Medical Officer. [c] Question was not asked for the blended learning course. [d] Respondents could select multiple options, so percentages sum to >100%. [e] Denominator for this question differs from the number answering "No" above due to variations in test use (ordering vs interpreting) and conflicting responses in category and open text entries.

Table S3b Medical specialty of baseline survey respondents for Specialty workshops and Blended learning course

	Specialty workshops		**Blended learning course**	
Baseline surveys completed	**N=198**[a]		**N=63**[b]	
Medical specialty	**n**	**%**	**n**	**%**
Anaesthesiology			2	3
Cardiology – Adult	19	10	2	3
Cardiology – Paediatric	3	2	1	2
Clinical genetics[c]	17	9		
Colorectal medicine and genetics			1	2
Dermatology – Adult	22	11		
Dermatology – Paediatric	13	7		
Gastroenterology/Hepatology			2	3
General medicine	1	1	1	2
Haematology – Adult	12	6		
Haematology – Pediatric	2	1		
Haematology – Research	1	1		
Immunology – Adult	4	2	1	2
Immunology – Pediatric	3	2		
Immunology/Allergy			1	2
Infectious diseases	2	1	1	2
Medical oncology			19	30
Metabolic medicine	2	1	1	2
Nephrology			2	3
Neurology	25	13	2	3
Neurology – sub-specialty	8	4		
Neuropsychiatry	1	1	1	2
Ophthalmology			2	3
Other (includes specialist nurses)[d]	9	5	1	2
Paediatrics and General Paediatrics	27	14	19	30

	Specialty workshops		Blended learning course	
Baseline surveys completed	**N=198**[a]		**N=63**[b]	
Medical specialty	**n**	**%**	**n**	**%**
Pediatrics – Community	1	1		
Pediatrics – Developmental	15	8	1	2
Pediatrics – Intensive Care	9	4		
Pediatrics – Neonatal intensive care	15	8		
Pediatrics – Neurodevelopment	3	2		
Pediatrics – Neurology	16	8		
Pediatrics – Rehabilitation	1	1	1	2
Pediatrics – Emergency			1	2
Palliative care	1	1		
Pathology	1	1	1	2
Psychiatry			2	3
Radiology			1	2
Rheumatology – Adult	3	2	1	2
Rheumatology – Pediatric			2	3

[a] Baseline surveys could not be deployed for the Neurology trainee workshop and Neurology conference workshop, so are not included here. Percentages are given out of 198, the number of baseline survey responses to this question. Respondents could specify multiple specialties so %s sum to >100. [b] Percentages are given out of 63, the number of baseline survey responses to this question. Respondents could specify multiple specialties so %s sum to >100%. [c] Clinical genetics includes Clinical Geneticists and Genetic Counsellors. [d] Other includes non-medical staff and allied health such as specialist nurses and psychologists, researchers, health technology adviser, and medical scientists.

Table S4. Value ratings of educational activities and learning approaches.

Rating scales for different program elements were: Needs Improvement, Fair, Good, Very good, Excellent; or Not useful, Somewhat useful, Useful, Very useful.

	Specialty workshops		**Blended learning course**	
	n	**%**	**n**	**%**
Overall value	**214**		**57**	
Very good or Excellent	186	87	46	81
Background reading and/or Introduction	**178**		**N/A**	
Useful or Very useful	162	91		
Modules	**N/A**		**38**	
Very good or Excellent			29	76
Case studies	**212**		**60**	
Useful or Very useful	209	99	60	100