Horizon Scanning Series
The Future of Precision Medicine in Australia

Professional Development

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1. Introduction

The successful implementation of precision medicine in health care will depend on the availability of a workforce able to deliver both genomic medicine and precision medicine. Underpinning this is the recognition around the world that professionals working in health care will need to have sufficient competencies (knowledge and skills) around genomics (and other 'omics') to support implementation. Workforce development requires education and training (including accreditation) for both new and established professionals and faces a number of challenges going forward.

1.1 Current literacy levels and need for professional development

The need for genetic/genomic education of non-genetic specialist health professionals has been the subject of discussion for more than 20 years (Collins, 1997). Despite substantial resources and activities in this area over the years (Bennett et al, 2017; Talwar et al, 2017), there are concerns that health professionals are still not adequately prepared for the rapid pace of advances in, and expectations around, genomics and precision medicine. This is compounded by acknowledgement that, both internationally (Bennett et al, 2017) and within Australia (Australian Government Department of Health, 2017), the limited number of existing genetic and other expert health professionals cannot meet demand for services.

Precision medicine can offer tailored diagnoses, prognoses and therapies to individual patients but will also generate large volumes of complex information for health professionals to interpret and communicate with patients. Recent international literature has identified that professional development for health professionals is still required to address gaps in genomic literacy and confidence/competence in requesting testing and interpreting results (Feero and Green, 2011; McInerney et al, 2012; McCarthy et al, 2013; Haspel and Saffitz, 2014; Alyass et al, 2015). While provision of education and training will contribute to increased competence, it is also important that health professionals see a need for education and accept they have a role to play in implementing genomic and precision medicine. However, health professionals may lack awareness of these imminent and increasing demands on their practice (Mirnezami et al, 2012; Jameson and Longo, 2015). Therefore, for education and training of professionals to be acceptable, and undertaken, requires that education providers have an understanding of perspectives of professionals in terms of their attitudes and needs, rather than developing educational programs from solely a top-down viewpoint (Feero and Green, 2011).

In addition, there are calls for clinical decision support tools (point-of-care/guidelines) to be developed and made available for genomic and precision medicine to be successfully implemented (Feero and Green, 2011; Mirnezami et al, 2012; McCarthy et al, 2013; Mikat-Stevens et al, 2015).

1.2 International professional development activities

In the UK, Health Education England's Genomics Education Program (GEP) has a suite of postgraduate and continuing professional development courses, both in-person and online, aimed at a broad range of disciplines, spanning genetic and non-genetic health professionals, medical scientists and allied health. The recent annual report of the Chief Medical Officer highlighted the role of education by Genomics England in integrating genomics into Britain's National Health Systems (NHS) by providing health care staff "with relevant data science expertise" (Ch1, p22). Through the Wellcome Genome Campus, the Wellcome Trust hold numerous conferences and courses, from laboratory to

computational to clinical aspects of genomics, and their Connecting Science initiative aims to translate Wellcome Trust research to health professionals and the public.

In Europe, the Gen-Equip program, a consortium of health professionals and educators across six countries, also provides resources and workshops for continuing professional development of primary care health professionals across Europe and practical clinical tools. The French Plan for Genomic Medicine 2015 describes the "establishment of the necessary training in genomic and digital health in universities and schools to meet the challenge of exploiting and interpreting data".

In the United States, one of the aims of the Precision Medicine Initiative at the National Institutes of Health (NIH)'s National Human Genome Research Institute (NHGRI) is to provide clinicians with the knowledge to select the most appropriate tests and treatments for each patient. The All of Us cohort study of one million Americans is examining how genetics, environment and lifestyle can influence how best to prevent or treat disease, particularly in oncology and pharmacogenomics, and the NHGRI website provides links to numerous genomic education materials. The NHGRI also hosts the Implementing Genomics in Practice (IGNITE) Network, which uses an implementation science framework to analyse six demonstration projects – ranging from family histories and monogenetic disorders to pharmacogenomics – where participants provide feedback on work practices, processes and utility of genomic testing, plus training, knowledge, self and group efficacy, beliefs and attitudes (Addie et al, 2016). The NHGRI also convenes the Intersociety Coordinating Committee for Practitioner Education in Genomics (ISCC), which aims to improve genomic literacy of health professionals through shared needs assessments and educational approaches and resources. Members include representatives from NIH institutes and professional medical organisations, plus key genomic educators from many countries, including the United States, Canada, United Kingdom and Australia. An example of resources shared through this committee include the Training Residents in Genomics (TRIG) online modules for pathologists, which were developed based on a robust needs assessment, use a 'train the trainer' model of education to increase capacity, and are formally evaluated.[1] The curricula are now being expanded to American undergraduate medical students. The ISSC has also developed a framework to develop genomic practice competencies for a health professionals that can be tailored as appropriate to different disciplines (Korf et al, 2014).

The mission of the American Society of Human Genetics (ASHG) is to discover, educate and advocate; their education portfolio spans formal K–12 and university institutions, medical training and continuing professional development for health professionals, plus public outreach. The ASHG recently prioritised education of non-genetic health professionals and formed the Genomic Medicine Education Consortium, which provides courses, blogs, podcasts and other resources.

The Jackson Laboratory provides continuing medical education and training modules via the Precision Medicine for Your Practice program, in collaboration with Scripps Translational Science Institute and the American Medical Association. The modules emphasise real-world examples of applying genomic information, assessing utility and being aware of both the benefits and limitations of new technologies. A number of other leading American academic medical centres run annual one–two day symposia aimed at educating health professionals and the biomedical sector.

In Canada, the Genetics Education Canada – Knowledge Organization (GEC-KO) was founded by the Children's Hospital of Eastern Ontario (CHEO) Department of Genetics to enhance genetics literacy of health care professionals by curating, developing, promoting and evaluating genetics and genomics educational materials and building competencies for primary care, medical specialties and allied health professions. More locally, regional genetics services run workshops for medical specialists, such as the Workshop in Genomic Medicine for Paediatric Specialists at The Hospital for Sick Children, Toronto, held in February 2017.

In Asia, the University of Malaya and Chinese University of Hong Kong are planning to offer a Certificate Course in Clinical Genetics and Genomics in conjunction with the Baylor College of Medicine for non-

[1] Noted in www.genome.gov/pages/research/researchfunding/dgm/sixth_isccmeeting_minutes_01242017.pdf

genetic health professionals. A limited number of Australian-based education programs are also offered in Asia (detailed below).

Several countries have started producing massive open online courses (MOOCs), many of which are targeted to international audiences as well as providing information about local contexts around health care systems, governance, etc. Popular MOOCs include series produced by St George's University London, Health Education England, University of California, Stanford University and Johns Hopkins University. Curricula range from introductory, top-level concepts, explanations of next generation sequencing in clinical contexts, detailed bioinformatics analyses and variant curation, through to ethical, legal and social issues (ELSI) around genomic and precision medicine. Target audiences range from novice through expert health professionals, medical scientists and the public.

Two global consortia – the Global Alliance for Genomics and Health (GA4GH) and the Global Genomic Medicine Collaborative (G2MC) – aim to align the varied international approaches into flexible frameworks to foster sharing and implementing genomic tools and knowledge. The GA4GH held their first biannual symposium in London in May 2017, where 15 national genomics initiatives shared data, resources and experience, including educational research and programs. Expected outcomes include a catalogue of educational resources (including approaches to development, needs assessments and evaluation) and an evaluation framework to measure outcomes of educational activities, led by the Australian Genomics Health Alliance (Australian Genomics).

Australian Genomics Program 4 (see below) is collaborating with international senior academics in education provision and evaluation from the Genomics Education Program, the Genomic Medicine Division of the NHGRI and GEC-KO, as well as national experts to develop a generic program logic and evaluation framework. These tools will be applied internationally to develop, deliver and evaluate genomic education programs in the future.

Several countries already have, or are developing, National Genomics Frameworks that cite the importance of creating genomic literate workforces, including Australia, Canada, France and the United Kingdom. International collaboration is vital, and Australian Genomics will maintain strong and active international collaborations across genomic workforce education, training and development.

1.3 National professional development activities

The recent draft Australian National Health Genomics Policy Framework rated "a skilled and literate genomics workforce" as a priority area, defining this as both genomic specialists (clinical geneticists, genetic counsellors, genetic pathologists and clinical bioinformaticians) and multidisciplinary teams that include non-genetic health professionals. The framework envisions a future in which a well-trained and certified genomics workforce has clear roles and responsibilities based on core competencies. The draft framework noted there is only limited information on the scope of the workforce and low genomic literacy levels in the non-genetic health professional workforce.

The development and support of a skilled and literate genomics workforce is a fundamental tenet of Australian Genomics. Program 4 research within Australian Genomics focuses on mapping current education and training programs to identify gaps and opportunities, assessing the needs of relevant stakeholders, and formulating guidelines to evaluate education and training programs. It will also contribute to the updating of core capabilities in genetics for medical graduates that the Human Genetics Society of Australasia (HGSA) developed in 2008. Overall, this program of work is designed to inform and assist education providers in genomics throughout Australia and potentially internationally.

The genetic and genomic specialist workforce in Australia includes clinical geneticists, genetic counsellors, genetic pathologists, medical diagnostic scientists and clinical bioinformaticians.

- Clinical geneticists and genetic pathologists must have completed a medical degree, internship and residency, before commencing training through the Royal Australian College of Physicians (RACP) or Royal College of Pathologists of Australasia (RCPA) to gain their Fellowship in Clinical Genetics and/or Genetic Pathology (also offered as combined Advanced Training). The process

of specialising typically takes 6–8 years and Fellows must then comply with continuing medical education requirements.

- Genetic counsellors complete a postgraduate Master of Genetic Counselling then undergo certification through the HGSA by being employed clinically as a genetic counsellor and submitting cases, log books, supervision reports, literature review or publications, a reflective essay and evidence of continuing education for review by a Board of Censors. The certification process typically takes 3–5 years and genetic counsellors must then comply with maintenance of professional standards (MOPS) to maintain certification.
- Medical scientists working in diagnostic laboratories typically undertake a science/biomedical science undergraduate degree for the initial part of their training. Depending on the situation and laboratory discipline they may then undertake a Masters level degree and/or PhD. In genetic/genomic diagnostics, scientists have traditionally specialised in cytogenetics, molecular genetics or biochemical genetics, and then taken examinations through HGSA in the specific discipline (MGHSA). Further on-the-job training was required to be eligible for fellowship examinations (FHGSA). In recent years, the disciplines have merged to an extent and fellowship training is now available through RCPA Faculty of Science (FFSc RCPA) specialising in either medical genomics or biochemical genetics, taking 5 years while working in a genetic pathology laboratory with examination. Another pathway allows scientists to gain FFSc through research (published works). From 2018, the entry point into FFSc RCPA will be to undertake a new Master of Diagnostic Genomics while working in a genetic pathology laboratory (see below).
- Clinical bioinformaticians typically gain an undergraduate degree including computer science or software engineering, a Masters and/or PhD of Bioinformatics – such as the Master of Science (Bioinformatics) at the University of Melbourne – then several years' experience with exposure to clinical work. The role and job title are not currently formally certified through a college or professional organisation.

Australian Genomics Program 4 recently completed a professional status survey of clinical geneticists and genetic counsellors to help map the genetic health professional workforce practice and readiness for genomics (Nisselle et al, 2017). To date, 677 people have graduated with an Australian genetic counselling degree (Grad Dip or Master) and approximately 150 have completed advanced training in clinical genetics through the RACP. Of 354 survey respondents, currently two-thirds (67%) of genetic counsellors work in clinical roles; of those, one-third (31%) incorporate genomics into their practice. In contrast, almost all (97%) clinical geneticists work clinically, with three-quarters (75%) incorporating genomics into practice. Encouragingly, a higher proportion of all genetic counsellors and clinical geneticists have completed genomics continuing professional development in the last two years (74% and 87%, respectively), attending predominantly workshops and conferences.

The draft Australian framework noted while state and national education and accreditation initiatives exist – spanning undergraduate through to postgraduate education and continuing professional development – a more cohesive national approach is needed. Australian Genomics Program 4 mapped available **genomics-specific** education programs in 2016 through desktop research and interviews with genomic education providers across Australia. The audit found 49 activities offered, or in development, across the spectrum of formal award programs (offered by professional colleges and universities) and accredited continuing professional education programs through more informal workshops and online activities (Table 1; Prichard et al, 2017).

Category	Description	n
Postgraduate course/ subject	Masters (2 existing, 3 new to start in 2018–19) GradDip/Cert or individual subjects (may be 3rd year u'grad)	5 15
Substantive program	Substantive ongoing program (workshops, podcasts, resources, case studies) suitable for CPD	27
MOOC	Massive Open Online Course	2
	TOTAL	**49**

Table 1. Genomics-specific education activities available and/or produced in Australia in 2016.

The activities are offered by a range of institutions which have local and national reach, such as the universities in each state, Melbourne Genomics Health Alliance ('Melbourne Genomics') and the Centre for Genetics Education (CGE), Bioplatforms Australia, and the Garvan Institute of Medical Research ('the Garvan'), all based in NSW. There are only two Masters-level courses at present, the existing Master of Genetic Counselling courses offered at the University of Melbourne and University of Sydney (intake frozen in 2018 for review of course). Three new courses are due to commence in 2018–19: Master of Genomics and Health, with nested grad Dip and Grad Cert (University of Melbourne), Master of Diagnostic Genomics (Queensland University of Technology), and Master of Genetic Counselling (University of Technology, Sydney). In addition there are some university Masters-level subjects in bioinformatics. Some substantive courses are available solely online, e.g., a couple in nutritional genomics, another in personalised medicine, while many substantive programs are workshops, often embedded in professional activities such as annual conferences (for example, the annual scientific meeting of the HGSA, the Pathology Update or medical college conferences). Successful programs that have been repeated locally, nationally and internationally include:

- *Annual Clinical Genomics Symposium* – developed by the Garvan and CGE in 2016, first hosted in Sydney and now held annually around Australia from 2017 with Australian Genomics support
- *Clinical Genomic Data Analysis Workshop* – developed by the Garvan in 2015, first hosted in Sydney, then Hong Kong
- *Variant Curation Workshop* – developed by Melbourne Genomics, first hosted in Melbourne in 2016, then Singapore and New Zealand, and repeated in Melbourne.

Regular seminars are also held by genetics and health care services, such as the Genetics and Genomics Lecture Series hosted by the University of New South Wales at St Vincent's Hospital, Sydney.

The Queensland Genomics Health Alliance, Sydney Genomics Collaborative, Canberra Clinical Genomics, South Australian Genomics Health Alliance and Melbourne Genomics are all actively investing in and developing the genomics workforce in their states. For example, several activities identified in Table 1 were developed and delivered by Melbourne Genomics, which has provided continuing professional development to over 800 health professionals, scientists and other stakeholders. Other education resources and activities have been developed based on curricula and lessons learned from Melbourne Genomics deliverables.

In tertiary care settings, another avenue for continuing professional development for health professionals is multidisciplinary team (MDT) meetings, which can include genetic and non-genetic health professionals, scientists and bioinformaticians. For example, Melbourne Genomics and Australian Genomics both currently use MDT meetings to triage patients for whole exome/genome sequencing of germline variants and to discuss and confirm results. At molecular tumour board (MTB) meetings, oncology patients are triaged for appropriateness of sequencing somatic driver variants and subsequently prognosis and treatment options (Harada et al, 2017). Peer-to-peer, cross-disciplinary knowledge integration occurs at these meetings, and is an important educational activity.

In the primary care setting, the *Genetics in Family Medicine: The Australian Handbook for General Practitioners* resource is hosted by the NHMRC and now managed by the Royal Australian College of

General Practitioners. The resource has been recently updated to include genomics, *Genomics in General Practice*, and modified to become a point-of care tool for launch in early 2018.

To date, many genomic education programs have been developed on topics for which technology is being implemented, rather than as a comprehensive approach based on a national needs assessment of health professionals. Therefore, as noted above, Australian Genomics Program 4 research also includes developing an evaluation framework to ensure high-quality, evidence-based genomic education activities moving forward. A draft program logic is currently in progress and will be developed into an evaluation framework and piloted by an Advisory Board of international experts in genomic education and evaluation throughout 2018–19.

Identifying educational needs, successful training approaches and evaluation strategies will ultimately improve the outcomes of genetics and genomics education. Australian Genomics Program 4 will also consider the patient perspective on genomic testing, along with the ethical, legal and policy issues regarding data sharing, and a theoretical ethical analysis of genomic testing. A series of national, deliberative workshops with the Australian public is gathering perspectives on personal genomics, in collaboration with an ARC-funded research study (www.genioz.net.au), to make recommendations for education, consumer information and public engagement strategies in this area. Results from all these projects will feed into recommendations and guidelines for effective professional education in genomic and precision medicine.

1.4 Issues moving forward

There are a number of challenges around professional education that are ongoing and will not fall in the remit of the work undertaken by Australian Genomics Program 4. Some of these are discussed below.

- **Competencies.** Although work is underway regarding developing up-to-date competencies in genomics for medical graduates (and also for continuing professional education) to inform medical curricula, the challenge will be to actually see these implemented within Australian medical schools. The previous core competencies developed in 2008 by HGSA were not endorsed by the Medical Deans of Australia and New Zealand. To date, this body has endorsed a national set of competencies in indigenous health only. In moving forward it will be necessary to engage with this body to guarantee that modern medical curricula reflect the importance of genomics and precision medicine and ensure a suitably trained medical workforce.
- **Genomic specialist workforce.** The limited existing genomic specialist workforce is a potential barrier to implementing genomic and precision medicine in Australia. There is a need to attract more medical graduates into clinical genetics training programs, yet this also requires sufficient training positions to be available, and hence is a financial resource issue. In genetic counselling, the issue is not of attracting potential students (e.g., there have been more than 150 enquiries for 20 places in the Master of Genetic Counselling at the University of Melbourne in 2018 – the number of places has increased from 12). The HGSA Board of Censors sets clinical placement requirements for genetic counsellors and a major bottleneck to increasing the numbers of genetic counsellors in training at the Masters level has been the number of clinical placements available. These are likely to increase but again, expanding these has resource implications for clinical genetics services providing placement opportunities.
- **Role of genetic counsellors.** Another consideration is how genetic counsellors might practice in the future. One important aspect is that of recognition of the profession, which is under debate. Self-regulation has commenced through the National Alliance of Self-Regulating Professions (NASRHP), although this process will take time. Some genetic counsellors are already working in private practice, either with clinical geneticists or in ultrasound and IVF practices. Indeed, some are working with other health professionals in primary care, which may be a model for how genomics and precision medicine become integrated and managed in primary care in the future, notwithstanding the need for further education of primary care practitioners, such as GPs. However, once again funding for such roles needs to be considered,

and also whether genetic counsellors (in public and private practice) will in the future be able to order genetic, genomic and other tests on the Medicare Benefits Schedule.

- **Conflicts of interest.** There are some concerns around a potential conflict of interest when genetic counsellors work for private genetic testing companies, especially when undertaking pre-test counselling (Minear et al, 2015). There are also potential ethical issues around conflicts of interest when commercial testing companies themselves provide 'education' of health professionals who would be ordering tests, which may or may not have clinical validity and utility. It is therefore important to provide health professionals with independent training through colleges, professional societies, and other accredited education providers, using best practice guidelines and minimising the perception of coercion.

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