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SPECIAL ISSUE

Role and practice evolution for genetic counseling in the genomic era: The experience of Australian and UK genetics practitioners

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Abstract

Facilitating informed decision-making regarding genetic testing is a core component of genetic counseling practice. Internationally, genetic testing is shifting toward gene panels and genomic testing, including whole exome and whole genome sequencing to improve diagnostic yield and cost-effectiveness. This study explored genetics practitioners' current experience with panels and genomic tests and the associated evolution of genetic counseling practice. Genetics practitioners with genomic testing experience, were purposively invited to participate in a semi-structured telephone interview and to snowball the invitation to colleagues. Interviews conducted with participants residing in Australia (n = 9) and the UK (n = 5) were transcribed and analyzed using an inductive thematic approach. Three themes emerged: (a) Role delineation: current roles, future roles, and the influence of increasing complexity; (b) The evolving spectrum of practice: blurred boundaries between research and clinical services; impact on facilitation of informed consent; and return of results strategies; and (c) Policy and governance needs: equality of access; achieving consistent variant interpretation, reporting, and responsibility for review; managing incidental findings; and professional regulation for Australian genetic counselors. These exploratory data highlight that genetic counseling practice and the essential role of facilitating informed consent are evolving but remain patient-centered, with core skills underpinning practitioners' capacity to adapt.

KEYWORDS

genetic counselor roles, genomic testing, informed consent, mainstreaming, multi-disciplinary teams

1 | INTRODUCTION

The speed and cost-effectiveness of next-generation sequencing (NGS) and subsequent increased feasibility of offering genetic/genomic testing in the clinical setting have led to a paradigm shift in genetic counseling practice. Where appropriate gene panels, whole exome sequencing (WES) and whole genome sequencing (WGS) are being applied in routine clinical care (Facio, Lee, & O'Daniel, 2014). While pathogenic variants in over 3,500 genes are known to cause human disease (Online Mendelian Inheritance in Man, 2016), numerous conditions have likely, yet, unresolved genetic etiology. The considerable diversity discovered in individual genomes, incomplete penetrance, and variable expressivity all complicate the process of variant classification (Bowdin, Ray, Cohn, & Meyn, 2014) and impede

expert consensus on the reportability of results (Amendola et al., 2015; Dewey et al., 2014). As a result, the scope of possible results and amount of data generated in this genomic era are placing unprecedented demands on the practitioners in the fields of medical genetics and genetic counseling (Johansen Taber, Dickinson, & Wilson, 2014). Specifically, the discovery of variants of uncertain significance (VUS), unexpected findings (UF), and incidental findings (IF) challenge patient education, facilitation of informed consent, and result(s) delivery (Facio et al., 2014; Machini, Douglas, Braxton, Tsipis, & Kramer, 2014; Ormond, 2013; Tomlinson et al., 2016). Variant reclassification also creates a need to develop pathways for clarifying responsibility in the clinical setting for reviewing genomic data (Rigter, van Aart, et al., 2014).

Preliminary evidence indicates the genomic era is impacting the practice of genetic counseling, with a shift from genetic to "genomic counseling" (Mills & Haga, 2014; Ormond, 2013) necessitating modifications to current practice (Merrill & Guthrie, 2015) and providing new practice opportunities (Kromberg, Wessels, & Krause, 2013). In the USA, genetic counselors (GCs) are involved in all stages of counseling for NGS, including the decision to offer testing, consenting with discussion of psychosocial associated issues, variant classification, and reporting results to patients, either independently or in collaboration with a Clinical Geneticist (CG) (Machini et al., 2014; Ormond, 2013). In some specialist clinics in the UK, GCs are also involved in variant classification (Middleton, Hall, & Patch, 2015). Non-clinical GC roles are also increasing in these countries (Field, Brewster, Towne, & Campion, 2016; Powell, Hasegawa, & McWalter, 2010). However, in Australia the context of the provision of genetic counseling is different: unlike the UK and the USA, genetic counseling is not a nationally regulated or registered profession. As such, GCs can only facilitate genetic/genomic testing when working under the medico-legal purview of a CG (or other medical practitioner), and counseling appointments are only billable through the national health system (Medicare) when the medical practitioner is in attendance for at least part of the consultation (Australian Institute of Health & Welfare, 2014; Australian Law Reform Commission, 2003). Fifteen years ago, Australian GCs roles included referral assessment, collecting personal and family history, risk assessment, patient education regarding genetic concepts to facilitate informed decision-making and addressing the psychosocial impacts of a diagnosis (James, Worthington, & Colley, 2003). More recent workforce reviews of GCs employed in the state of New South Wales identified that these roles continue but that experienced counselors have more autonomy and are increasingly offering services in non-genetics specialty areas (Barlow-Stewart, Dunlop, Fleischer, Shalhoub, & Williams, 2015; Urbis, 2017).

To underpin workforce planning for the genomic era there is a need to survey Australia's current approaches to genetic counseling and compare these with international best practices. This study explores current and evolving practices of genetic counseling for NGS in Australia with comparisons to the UK for two reasons. Firstly, similarities in the Australian and the UK health care systems provide opportunities for useful comparison. Citizens of Australian and the UK are provided universal health care through government funded public health systems (Medicare and the National Health Service [NHS], respectively), with optional insurance coverage or self-funding for additional investigations in a private healthcare setting (Australian Institute of Health & Welfare, 2014; National Health Service Choices, 2016). Secondly, drawing upon practitioner experience from the UK's 100,000 Genomes Project (Genomics England, 2015), provides an opportunity to reflect on the current clinical integration of genomic tests in Australia, through comparison with a system thought to be a few years ahead. Ethics approval was received from The University of Sydney's HREC (Approval number 2016/877).

2 | METHODS

2.1 | Participants and procedures

Two coauthors with longstanding connections to the genetic counseling profession in Australia (K.B.S.) and the UK (R.O.S.) identified a purposive sample of 16 genetics practitioners, who were known to have experience with gene panels and/or WES/WGS. Eligible participants included GCs, CGs, and other medical practitioners with expertise in genetics, who were residing in Australia and the UK. Both male and female practitioners with experience in either general genetics departments and/or a range of clinical or research specialisms were considered for participation. A total of 11 Australian practitioners and five UK practitioners received an email invitation, enclosing a participant information statement and consent form. The invitation included a request to snowball to colleagues with similar experience; however, snowball sampling was not tracked by the researchers. After receipt of emailed consent, the first author (T.D.) conducted semi-structured telephone interviews during January–July 2017. The interview schedule covered eight broad areas (Supporting Information Data S1).

2.2 | Data analysis

Interviews were transcribed verbatim and de-identified to ensure participant anonymity. An initial coding tree was generated by T.D. using responses obtained from the first interview. Iterative interviewing was instituted so that additional questions/prompts and codes were added to the interview schedule and coding tree as they arose. A subset of transcripts was coded for concordance by two independent researchers (B.T. and R.O.S.). The analysis was guided by an inductive thematic approach (Thomas, 2006) to identify themes encompassed within the data.

3 | RESULTS

Of 17 participants who consented, 14 were interviewed: Australia (n = 9: 8 GCs; one Clinical Genetics Fellow) and the UK (n = 5: 4 GCs; one Cancer Genetics Clinician [CGC]) (Table 1). The remaining

TABLE 1 Participant experience with panel and/or genomic testing (WES/WGS)

Participant	Role	Testing experience (Y/N)		
		Panel	WES	WGS
P01	Clinical Genetics Fellow (Aus)	Y	Y	N
P02	GC (UK)	Y	Y	Y
P03	GC (Aus)	Y	Y	Y
P04	GC (Aus)	Y	Y[a]	Y[a]
P05	GC (UK)	Y	Y	Y
P06	GC (Aus)	Y	Y	Y
P07	GC (Aus)	Y	Y[a]	Y[a]
P08	GC (Aus)	N	Y	N
P09	GC (Aus)	Y	Y	N
P10	GC (UK)	Y	Y[a]	Y[a]
P11	GC (Aus)	Y	Y	Y
P12	GC (UK)	Y	Y	N
P13	GC (Aus)	Y	Y	Y
P14	Cancer Genetics Clinician (UK)	Y	Y	Y

[a]Experience limited to pre-test counseling.

consented participants either indicated they were no longer available (n = 1) or failed to schedule an interview (n = 2). Participants reported an average of 11.8 years (range 3–28 years) experience in their profession. This included a reported average of 4.0 years (range 0–9 years) with gene panels, 3.1 years (range 0.5–6 years) with WES, and 1.4 years (range 0–6 years) with WGS, in a combination of both clinical and research settings. All participants who completed an interview were female, though to protect anonymity, additional information, including specialism and individual duration of practice are not reported here. The interview duration ranged from 38–96 min (median 64 min). Independent coding of transcripts yielded 94% concordance.

Thematic analysis identified three major themes, with representative quotations used to illustrate the key findings.

3.1 | Role delineation: Current roles, future roles, and the influence of increasing complexity

The genetic counseling profession's inception in both Australia and the UK was about 30 years ago (Ormond et al., 2018). Many GC participants indicated that over that time their autonomy and scope of their role have increased considerably. However, several GCs described a "sliding scale" between case complexity and the amount of autonomy they experience. GC-led appointments were used to facilitate informed choice and testing for previously diagnosed and more straightforward conditions and cascade testing for relatives, with the geneticist/clinician returning results with greater complexity or more immediate clinical management issues:

> I have a lot of autonomy and I think it is growing… the more straightforward a result is, the more autonomy I/we are given to deal with it and the more complex or unexpected or uncertain that it becomes, the less autonomy… or the more input from others you might have (P09, Australian GC)

Many participants reported that evolving GC roles included increasing recognition as a member of multi-disciplinary teams (MDT), with responsibilities including representing the genetics department during peer discussion, providing the family perspective, and advocating for patients:

> I certainly have more of a voice in that meeting than I used to… I am often the person who has met the patients, and I understand the family structure and who else may or may not be affected in the family, and it's then my role to contribute that information to the discussion (P12, UK GC).

All participants expressed the importance of GCs understanding the process of variant interpretation and being able to effectively communicate genomic variants. However, there was no consensus in regard to the role of GCs performing variant interpretation. Some expressed the view that all genetics practitioners should take a more active role in this process, others that this role was beyond their scope, while some were not willing to prioritize the role over counseling:

> I think it makes me a better counselor to understand that stuff, but I also don't want to prioritize curation and variant interpretation over counseling and the face-to-face or the contact that you have with patients (P09, Australian GC)

Almost all participants had views on "mainstreaming" of genetic and genomic testing into non-genetics specialties of medicine. Many asserted that "greater specialism [in GC practice] is inevitable" and would coincide with mainstreaming, enabling GCs to upskill in a specific area of practice:

> We have got to get genetic counselors employed by oncologists and cardiologists… who better than genetic counselors in these situations, to realize who needs to be referred on to clinical genetics for diagnosis… yet who can handle the straightforward things that clog up our clinical genetics clinics at the moment (P08, Australian GC)

However, ongoing links between GCs and specialist clinical genetics services were viewed by six participants (n = 3 Australia; n = 3 UK) as essential to maintain expertise, supervision, and professional development, especially for new graduates.

3.2 | The evolving spectrum of practice

Despite numerous evolving practice challenges, many participants asserted the essence of genetic counseling and the core skills required by GCs are the same, regardless of the type of test. Both the Australian and UK participants indicated that the use of gene panels was increasing while WES/WGS, at least in the clinical setting, remains limited to cases of direct therapeutic utility or to determine recurrence risk.

3.2.1 | Blurred boundaries between research and clinical services

One recurrent theme that emerged was that research initiatives influenced the type of test being offered. UK participants described an increase in WES/WGS offered as part of the 100,000 Genomes Project following uninformative clinical testing. Some also reflected on the blurred boundaries between clinical and research services generated:

> We're not quite sure whether [the 100,000 Genomes Project is] research or just part of mainstream clinical practice. It's straddling both worlds in a slightly uneasy way" (P10, UK GC).

Some Australian participants also commented on the blurring of research and clinical funding for testing:

> Some of the testing… has been done more on a research interest basis, rather than necessarily completely 100% clinical, because it may not have completely fulfilled all the criteria by the hospital (P04, Australian GC).

3.2.2 | Impact on facilitation of informed consent

Pre-test counseling to ensure informed consent was identified as an area most challenged by genomic testing. Most participants indicated that the process was considerably longer, with some practitioners offering a second appointment, and some patients were requesting more time to consider testing. However, a minority of participants indicated that their pre-test counseling sessions were shorter, as the discussion became more generalized. Ultimately participants acknowledged the importance of maintaining patient-centered practice:

> I don't know how we can do this job and think one-size-fits-all… people [have] different needs… we have to let our patients take things on at the pace that they seem ready to take, and if they need two appointments or half an appointment we should go with that (P10, UK GC)

A strategy discussed to streamline the consent process was provision of information prior to the pre-test counseling session using booklets or videos. While some participants thought this could assist in patient education, several expressed that they would struggle to identify appropriate patients in advance; others had concerns that the complexity of the information may deter some patients from pursuing genomic testing:

> A video to aid consent, where you give people all the essential information, and that then in turn will prompt them to ask the questions that they want to when they get to clinic. I honestly envision that's what would be useful in the future (P08, Australian GC)

> I'm not sure if it would be possible to identify ahead of time those that we would definitely [offer] the test to, it would be terrible to send information about a potential test to somebody that it wasn't actually relevant for. I can also see the potential of it to put people off… because not everybody does want a genetic test (P12, UK GC)

Generic consent accompanied by comprehensive post-test counseling as a strategy to decrease the duration of pre-test counseling was another strategy proposed by four participants (three Australian GCs and UK CGC). Those using this strategy were either more experienced with WES/WGS or were employed in specialty areas.

3.2.3 | Return of results strategies

Apart from the higher diagnostic yield and the greater potential for multiple or uncertain variants, many participants viewed that practice underpinning delivery of genomic test results was comparable to those from single gene tests. However, some indicated that the uncertainty inherent in many genomic results has practical implications on genetic counseling workflows. These include increased preparation time prior to results delivery, additional investigations to confirm variants (e.g., segregation studies), and prolonged contact with some families:

> [If] it's a stop/gain, it segregates with the family, it fits perfectly, it is a lot easier to prepare for that consult…. Obviously, if it's something you're not as confident with, having to go through issues of uncertainty… that does take a bit more time (P11, Australian GC)

3.3 | Policy and governance needs

3.3.1 | Equality of access

Genomic testing remains cost-prohibitive in many clinical settings in both Australia and the UK. Consequently, participants noted

considerable inequality of access to both panel and WES/WGS tests in both countries:

> I know lots of other GCs that would love to be able to offer [WES/WGS], but it's just not been a possibility (P02, UK GC)

One Australian GC stated families must fund their own segregation testing, and several Australian and one UK participant said that some families are self-funding genomic tests. Overall, some participants expressed concerns that the clinical uptake of these tests in the public system was poor, at least in some specialisms, resulting in a two-tiered system:

> Access is not equitable at the moment, at least not where I work… it might be that the test is technically available, but …. is not something you can offer as a funded test, and they are not going to be able to afford it themselves (P09, Australian GC)

3.3.2 | Achieving consistent variant interpretation, reporting, and responsibility for review

Due to a lack of standardisation in processes (e.g., thresholds for reporting VUS) between institutions and laboratories, most of the participants reported challenges in reaching consistent variant interpretation and reporting:

> You could give two different laboratories the same exome data… and there is a significant potential to get two different reports back… Organizations have different legal requirements, or different SOPs [standard operating procedures] relating to what they are allowed to do… it's difficult to get consistency (P09, Australian GC)

Most described variant review as ad hoc, driven either by the patient recontacting a genetics service or file review by the practitioner:

> We're quite clear that it's not the laboratory's job to go back and review variants. That question comes out of a clinical setting where either the patient has got back in touch, or the clinician's looked back at the file (P05, UK GC)

Only one Australian participant described a role of the laboratory in initiating result review, though this applied to uninformative results rather than VUS:

> Our lab will also come back to us if there is new technology and say they are going to look at all these test results prior to such and such date… [to] review it again (P07, Australian GC)

The resource implications of reanalysis and requirements for a structured process to inform variant reclassifications were emphasized as the major professional challenge. Development of national guidelines was suggested as a strategy to facilitate the consistent return of results:

> Everyone's still working out how they're going to handle a genomic family… I'm sure it varies, and probably will be more beneficial if there was a national policy (P02, UK GC)

> We have a system within England whereby [a BRCA1 variant reclassification from Class 4 to 1] got reported to the head of the cancer genetics group and she cascaded it down to the heads of the labs across the whole network of the country… that is vital because of the possibility that someone might be waiting for prophylactic surgery (P14, UK CGC)

3.3.3 | Managing Incidental findings

Most participants had no direct experience with IF. Many expressed relief; however, some, typically those with more experience, reiterated the rarity of IF and stated they no longer feared them. Some participants questioned whether IF are being equally reported by laboratories and others acknowledged their departments are currently finalizing discussions with laboratories regarding their procedures for reporting IF:

> [IF] caused me a lot more angst at the start. But now that we're 5 years into this [research exomes] and there have been no scary outcomes and no frightening results… it's easier to put that 1% risk in context for patients. So, you do have to warn them… [but] in all likelihood we are not going to find something inadvertently (P08, Australian GC)

3.3.4 | Professional regulation for Australian GC

The iterative nature of the interview enabled addition of the issue of professional regulation to the interview schedule with six of the Australian GCs. All of these viewed it as an important issue and discussed the challenges and potential benefits that national professional regulation will bring. Five of the six participants agreed that professional regulation would enable greater flexibility and efficiency in workflows to better meet service demands. They described that CGs or other medical practitioners are commonly required to

move between concurrent appointments (led by GCs) to enable billing for the consultations. Four also viewed that greater autonomy and recognition of GC competencies generated by regulation would also drive professional and community awareness.

> [We need] a system for regulation of the profession so that we know that someone who calls themselves a GC is in fact a GC … And there needs to be more varied pathways to certification for those who don't fulfil traditional roles, because only by doing that do we ensure that those who fulfil these emerging roles are also adequately trained and regulated (P13, Australian GC)

4 | DISCUSSION

This study explored current GC roles and evolving genetic counseling practices for genomic testing (panel/WES/WGS) in both Australia and the UK. Overall, although the essence of genetic counseling remains the same, the scope and complexity of genomic tests and increased service demands are driving the evolution of GC roles and practice. Importantly, this study extends the perspective of Australian genetics practitioners and provides a comparison between the Australian and UK practice.

4.1 | Impact on facilitation of informed consent

The area of practice most impacted in the genomic era in Australia and the UK in this study was reported to be pre-test counseling to facilitate informed choice, which is consistent with that reported by the USA GCs (Machini et al., 2014). Longer or multiple appointments to ensure adequate preparation for multiple or uncertain results were reported. While this may be regarded as "best-practice", some participants questioned the feasibility of an extended consenting approach for patients and services already burdened by long waitlists. Given the impact, developing optimal consent models is necessary for the further evolution of genetic counseling practice.

Strategies to streamline the consent processes included preferences supported by Australian patients such as sending information beforehand to inform and "activate" patients (Genetic Alliance Australia, 2016). However, this approach requires identification of appropriate patients in advance, limits provision of patient-centered information, and may only be feasible as WES/WGS becomes a more common or first-line test. Support was also indicated for the concept of generic consent, defined as the provision of "broader concepts and common-denominator issues" (Elias & Annas, 1994) to increase cost-effectiveness and efficiency. While initially controversial (Biesecker & Wilfond, 1994; Wells, 1994), this approach is increasingly being reported in the era of genomic counseling in the USA (Bernhardt et al., 2015; Merrill & Guthrie, 2015) and some participants in this study were using this strategy. Importantly, they were either more experienced with WES/WGS or were employed in specialist services that frequently engage in specific risk-management discussions (e.g., cancer) or routinely conduct targeted analysis (e.g., research), which could reduce the complexity of pre-test counseling dialogues. It has been proposed that other models of "tiered," "binned," and "layered" consent, which initially deliver essential concepts and expand information provision only if patients have additional needs (Bradbury et al., 2015; Bunnik, Janssens, & Schermer, 2013), may further optimize patient-centered service.

4.2 | Managing genomic testing challenges

Bertier, Hétu, and Joly (2016) reported that IF; the interpretation, use and review of VUS; and the cost of WES are major unsolved clinical challenges. The Australian and UK participants identified that these challenges were current factors limiting the widescale implementation of genomic testing. Although the Royal College of Pathologists of Australasia (RCPA) recommends returning only pathogenic and likely pathogenic results (The Royal College of Pathologists of Australasia, 2015), both the Australian and UK practitioners in this study were reporting VUS to patients, to enable segregation studies and/or preparation for future variant reclassification. Reporting of VUS to facilitate further delineation of pathogenicity is consistent with the UK best practice guidelines (Ellard et al., 2018b). These data are also consistent with international reporting practices for VUS, though some laboratory personnel highlighted variability in how VUS are reported and ultimately used by clinicians (Vears, Senecal, & Borry, 2017). Nonetheless, reclassification is challenged by ad hoc recontact and review of VUS—and lack of clarity around process (Carrieri et al., 2017, 2016). Carrieri et al. (2017) highlighted the resource implications of allocating time and staff, and the lack of professional consensus about responsibility for variant review. One strategy presented here is for clinicians to opportunistically review VUS relating to their patients' ongoing management, with laboratories to assume responsibility for informing clinicians of variant reclassifications. An approach to promptly inform variant reclassifications in the UK was recently proposed by the Association for Clinical Genomic Science (ACGS), though a formal process in not yet regulated (Ellard et al., 2018b). However, communication regarding variant reclassification within Australia is further complicated by the existence of independent laboratory and clinical services, and separate state-based health systems. The development of effective and efficient pathways to update genetics practitioners in regard to variant classification employed in different genetics services across Australia requires additional research.

4.3 | Strategies for genomic results delivery

Overall, the Australian and UK participants viewed the process of returning genomic results as comparable to returning single gene test results, albeit with an extended timeframe and workload. This experience supported the findings of Williams, Faucett, Smith-Packard, Wagner, and Williams (2014), who reported GCs require additional time specifically to review medical records and obtain

information about identified variants, and Sukenik-Halevy, Ludman, Ben-Shachar, and Raas-Rothschild (2016) who described that GCs, in particular, experience increased administrative burdens.

The genomic era may increase time pressures on an already insufficient GC workforce as reported in Canada (Shugar, Quercia, Trevors, Rabideau, & Ahmed, 2017) and the UK, France, and the USA (Pain, 2016). However, delegation of the more straightforward clinical or patient-focused duties to GCs offers a solution to overcoming workforce shortages. Preliminary evidence from Australia (Urbis, 2017), including the perception of the Australian participants in this study, also indicates this to be the case in Australia. Both the Australian and UK participants highlighted recent changes to clinical service structures promoting opportunities for GC-led appointments. This is consistent with a recent UK study, reporting that GCs contributed to 95% of patient contacts, and led on 46% of cases (Benjamin et al., 2015). Similarly, in the USA, Hannig et al. (2014) trialed an approach where GCs triaged clients not needing clinical examinations into a general genetic counseling clinic with a CG advising their practice.

Barlow-Stewart et al. (2015) noted that certified and more experienced GCs are increasingly offering independent services, including those working in private health services (e.g., IVF clinics) without direct supervision of a clinician. However, participants in this study noted that expansion of this approach in Australia would require regulation and recognition of GC services for remuneration of services performed by GCs independent of a clinician.

4.4 | Evolving roles of GCs in the genomic era

Genetic testing within a multi-disciplinary context is an approach recently employed in Australia (Mallett, Fowles, McGaughran, Healy, & Patel, 2016; Pokharel, Hacker, & Andrews, 2016). The growing importance of GCs within MDT was acknowledged in this study and is consistent with recent European and Australian studies (Barlow-Stewart et al., 2015; Benjamin et al., 2015; Cordier, Lambert, Voelckel, Hosterey-Ugander, & Skirton, 2012; Middleton et al., 2017; Skirton, Cordier, Ingvoldstad, Taris, & Benjamin, 2015). Pestoff, Ingvoldstad, and Skirton (2016)'s exploration of the time investment and "value-adding" by GCs, identified four key factors unique to GC service: case management, holistic care, accessibility, and continued support. These findings support the view expressed by participants in this study who felt that GCs made unique contributions to MDT, particularly as main advocates for patients and families.

Employment of GCs within mainstream health care services in order to facilitate ethical and appropriate genetic testing is occurring internationally (Shugar et al., 2017) and in Australia (Barlow-Stewart et al., 2015). Further, internationally, GCs are increasingly being employed in laboratory settings and are involved in variant curation (Waltman et al., 2016), though this is not currently common in Australia (Barlow-Stewart et al., 2015) and GCs in this study were split on their views about scope in this area. Grove, Wolpert, Cho, Lee, and Ormond (2013) proposed a need for regulation over who can legally interpret sequencing results, and this requires further research in the Australian and UK context. Moreover, participants from both Australia and the UK highlighted a need for ongoing supervision and maintenance of professional expertise, by preserving links to a genetics department in a public hospital while working in mainstream services. The practice implications of greater specialism in the genomic era, considering the potential for IF outside a GC's area of expertise, also require further evaluation. Broader assessment of service delivery models for mainstreaming, including the views of non-genetics professionals (Rigter, Henneman, et al., 2014) are also required to promote effective integration of GCs within these services.

4.5 | Australian GC regulation

There is growing recognition of GCs internationally, with recent moves to register the profession in Europe (Paneque et al., 2016; Pestoff et al., 2016). Despite this, genetic counseling is not a registered profession in many countries, including Sweden (Pestoff et al., 2016) and Canada (Shugar et al., 2017). Shugar et al. (2017) recently proposed a compelling argument for the suitability for professional regulation, noting that 15% of GCs were working within mainstream services as a primary genetics practitioner despite the absence of regulation. Many of the Australian GCs spoke in support of professional regulation. While professional "registration" for GCs is not currently possible in Australia, the Australasian Society of Genetic Counsellors is currently preparing an application for genetic counseling to become a self-regulated profession in Australia (Australasian Society of Genetic Counsellors, 2017). Regulation of GC practice is important as more roles will involve GCs to be based in MDT as the genetics expert (Middleton et al., 2017; Patch & Middleton, 2018). Importantly, professional registration and GC certification in both the UK and USA have lead to more autonomous practice opportunities and recognition of GC ability (Baty, 2018) and ensures quality and safe practice (Professional Standards Authority, 2016).

4.6 | Study limitations

This study has a small sample size with a bias of predominantly GCs in the cohort. Although the inclusion criteria stipulated experience with panel and/or WES/WGS tests, some participants had only limited experience, particularly with the latter. While the duration of practice and specialisms were varied these views may not be representative of all genetics practitioners in Australia or the UK. Participation from a larger range of genetics practitioners, including those employed in industry or laboratory environments would have strengthened the data.

4.7 | Practice implications

Core genetic counseling skills have consistently supported the adaptation to advances in genetic technologies and practice demands. Recognition of the importance of maintaining a patient-centered service remains fundamental to the development of appropriate

consent models, management of uncertainty, and provision of psychosocial support in the genomic era.

These data highlight the frequency of VUS and a need to establish responsibility for variant review, including the development of a structured approach to inform reclassifications, as a major clinical obstacle in both Australia and the UK. Inconsistent interpretation of genomic results and disrupted communication of variant reclassifications complicates the management of families living in different locations. Collaboration between laboratory and clinical services, including consideration of strategies on how best to monitor and communicate variant reclassifications should be encouraged. The ACGS proposal for variant reclassification with immediate clinical management impacts to be promptly reported (Ellard et al., 2018a) is an important first step in developing a regulated pathway.

Clearer delineation of Australian GC responsibilities and degree of autonomy, particularly in the context of their role in variant interpretation and ability to order genetic/genomic tests, is dependent on professional regulation and/or alternate practice legislation. Ultimately, the scope for UK GCs in performing variant interpretation also requires clarification, with many participants suggesting only a supporting role despite registration. While GCs have traditionally practiced solely within genetics departments in public hospitals, mainstreaming of genomic testing will increasingly extend practice opportunities in both countries, leading to new GC positions within non-genetics services (e.g., cardiac and neuroendocrine clinics) or private clinics (e.g., large primary care clinics/General Practitioner Super Clinics). Genomic technologies are also likely to differentially impact distinct specialisms, and one approach will not sufficiently meet the needs of all services. Both the Australian and UK participants demonstrated evolution of genetic counseling practices based on their experiential learning. Ultimately, greater discussion between services to draw upon the experience of other practitioners may inform future policy and facilitate appropriate practice modifications. Finally, overcoming inequality in access to genomic technologies and establishing consistent and best-practice strategies following wide-scale implementation of genomic testing needs continuing assessment.

4.8 | Research recommendations

These exploratory data highlight evolving GC roles coupled with policy and governance for variant reporting and review, and management of IF, as areas warranting further investigation in Australia and the UK. As policies to manage the return of IF and inform variant reclassifications are implemented, ongoing evaluation of their effectiveness will be required to promote best practice. Further evaluation of emerging GC roles, including involvement in variant interpretation and employment in non-genetics services, will facilitate effective mainstreaming of genomic tests and a multidisciplinary approach to patient care in both countries. More specifically in Australia, the impact of impending professional regulation on counselor autonomy and scope of practice should be monitored to promote continued quality and safe practice. Finally, implementation of a recently developed national health genomics policy framework (Australian Health Ministers' Advisory Council, 2017) may facilitate the unification of state-based health systems and overcome potential state-by-state differences in GC practices in Australia.

COMPLIANCE WITH ETHICAL STANDARDS

Conflict of interest

During this research, Bronwyn Terrill and Marcel E. Dinger held part-time roles with Genome.One, a commercial, wholly-owned subsidiary of the Garvan Institute of Medical Research that offers clinically accredited genomic testing services. Tanya Dwarte, Kristine Barlow-Stewart, and Rosie O'Shea declare that they have no conflict of interest.

Human studies and informed consent

All procedures performed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all participants included in the study.

Animal studies

No animal studies were carried out by the authors for this article.

AUTHOR CONTRIBUTIONS

Tanya Dwarte contributed to the research design; prepared documents for ethics submission; performed and transcribed all interviews; developed the coding tree and completed coding of transcripts; and drafted the manuscript. Kristine Barlow-Stewart contributed to the research design; assisted in participant recruitment; provided revisions to ethics documents, coding tree and the final manuscript. Rosie O'Shea contributed to the research design; assisted in participant recruitment; coded transcripts for concordance; provided revisions to ethics documents, coding tree and the final manuscript. Marcel E. Dinger contributed to the research design and provided revisions to ethics documents and the final manuscript. Bronwyn Terrill contributed to the research design; coded transcripts for concordance; provided revisions to ethics documents, coding tree and the final manuscript.

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SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section at the end of the article.

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