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ORIGINAL ARTICLE

Psychiatric genetic counseling: a survey of Australian genetic counselors' practice and attitudes.

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ABSTRACT

Genetic counseling plays a critical role in supporting individuals and their families' adaption to psychiatric conditions, addressing the multifactorial nature of these conditions in a personally meaningful and empowering way. Yet data related to the practice and attitudes of Australian genetic counselors about psychiatric genetic counseling (PGC) is limited. This survey investigated the practice of Australian genetic counselors, and their attitudes towards PGC. Genetic counselors (N=393) were invited to participate in an anonymous online survey between March and May 2022. Forty-four genetic counselors (response rate = 11%) from Australia and New Zealand responded. No respondents practice in psychiatric genetics as their speciality area; most respondents do not see any patients where the primary indication is a personal and/or family history of psychiatric disorders (91%). Greater than half of respondents (56%) believed there was sufficient evidence to support PGC, and 64% enquire about personal and/or family history of psychiatric disorders, but only 25% provide genetic counseling on this topic. Most respondents do not feel confident providing risk assessments for psychiatric disorders (72%), while the majority expressed interest in attending specialist training (96%), and in incorporating PGC into future practice (77%). Australian genetic counselors would benefit from psychiatric genetic education and training, and establishment of specialised PGC services would address this gap in patient care, whilst providing opportunities for genetic counselors to gain skills and experience in PGC.

KEYWORDS: genetic counseling; mental health; psychiatric genetics; family history.

What is known about this topic?

PGC can play a crucial role in individuals and their families' adaption to psychiatric disorders (Austin 2020; Hippman et al 2016). Providing psychosocial support and etiological information in the context of family and personal history can facilitate patient empowerment, family communication and adaption, and raise awareness of primary prevention of psychiatric disorders (Austin 2020). However, genetic counselors infrequently receive referrals where the primary indication is a personal and/or family history of psychiatric disorders. There is a strong need to identify barriers to PGC and develop strategies to improve access to PGC.

What this paper adds to the topic?

This paper offers a unique perspective by describing the current landscape with respect to Australian genetic counselors' attitudes and practice towards PGC. We anticipate these results will provide a basis for discussion of this topic in both genetic counseling training programs, and in professional development; and we anticipate these results will provide further evidence to support implementation of dedicated psychiatric genetic clinics.

1. INTRODUCTION

Mental health is a key component of overall health and wellbeing (WHO 2021). During 2020- 21, an estimated 1 in 5 (5.0 million) Australians experienced a psychiatric disorder, including schizophrenia, depression, anxiety, obsessive compulsive disorder and bipolar disorder. During the same period, approximately 2.9 million Australians' received Medicare-subsidised mental health specific services through the Australian Government Medicare Benefits Schedule, which includes up to 20 Medicare-

subsidised psychological therapy sessions per year (Australian Institute of Health and Welfare, 2022; Australian Bureau of Statistics, 2022). Given the prevalence of psychiatric disorders in the Australian population, genetic counselors are likely to encounter patients with a personal and/or family history of psychiatric disorders on a daily basis. While the multifactorial nature of psychiatric disorders means a genetic test for non-syndromic psychiatric disorders is rarely available, expert genetic counseling remains a powerful, and relevant tool (Austin 2020; Hippman et al. 2016; Inglis et al. 2015; Moldovan et al. 2017; Schmitt et al. 2014).

Psychiatric genetic counseling provides psychosocial support and etiological information personalized to family and personal history, which creates a therapeutic environment characterised by empowerment, adaption and positive self-identity (Moldovan et al. 2017). It also provides an opportunity for at-risk individuals to be counseled on risk-reducing factors associated with psychiatric disorders, while focusing on strengthening protective factors (Austin 2020; Austin and Honer 2005; Semaka and Austin 2019).

Over 90% of individuals with a psychiatric disorder and their relatives' desire genetic counseling services (Lysus 2007), however few have been offered the intervention, and psychiatric disorders rarely constitute a primary indication for referral within genetic counseling (Monaco et al. 2010). For example in 2010, 83% of surveyed genetic counselors based in the US reported they '*rarely*' or '*never*' saw patients referred for psychiatric disorders (Monaco et al. 2010). This number has remained relatively static - in 2018, only 14.1% of post-graduate genetic counselors reported having received a primary referral for a psychiatric disorder (Low et al. 2018). This highlights a potentially significant service gap for individuals diagnosed with psychiatric disorders, and their relatives. To address this gap, specialized psychiatric genetic clinics have been

successfully established where dedicated genetic counselors provide PGC in the context of psychiatric disorders. However, there remains no dedicated psychiatric genetic clinic in Australia (Moldovan et al. 2019).

Without a dedicated psychiatric genetic clinic, it remains unclear whether, or how individuals with a personal or family history of psychiatric disorders needs are being met in Australia. Given the prevalence of psychiatric disorders in the general population, it is likely genetic counselors encounter patients with a personal and/or family history of psychiatric disorders on a daily basis – even where it is not the primary reason for referral (Austin et al. 2008).

The aim of this study was to understand whether genetic counselors are currently providing PGC in Australia and whether genetic counselors feel confident providing PGC, including risk assessments for psychiatric disorder. Describing the current landscape with respect to Australian genetic counselors' attitudes and practice towards PGC represents a necessary first step in understanding PGC practice in the region.

2. METHODS

This study was approved by The Royal Melbourne Health Human Research Ethics Committee (Institutional Review Board), (HREC2021.284).

2.1 Participants

Genetic counselors were recruited through the Australasian Society of Genetic Counsellors (ASGC). Genetic counselors were eligible to participate if they were

working clinically within Australia or New Zealand and able to complete the survey online in English language.

2.2 Instrumentation

The study instrument was a cross-sectional survey. The survey contained three sections. Questions from section I related to genetic counselors' demographics, including years of experience, practice speciality, region of practice, average number of patients seen per week and genetic counselor accreditation status. Section II of the survey evaluated genetic counselors' psychiatric genetic counseling practice. Likert-style questions were utilised to scale responses to questions regarding genetic counselors' practice of psychiatric genetic counseling and psychiatric illness, using 1= *always*, 2= *usually*, 3= *sometimes*, 4= *rarely*, and 5= *never*. Questions explored how frequently genetic counselors receive referrals for psychiatric genetic counseling and/or referrals where genetic testing has been facilitated for psychiatric illness, frequency genetic counselors enquire about personal and/or family history of psychiatric illness, and frequency of encounters where patients disclose a personal and/or family history of psychiatric illness secondary to their primary reason for referral. We asked respondents how they manage secondary disclosure in genetic counseling sessions, and offered the responses.

- I do not address it as it is not the primary reason for referral
- I provide genetic counseling in relation to the psychiatric history disclosed
- I rebook for a dedicated genetic counseling appointment relating to the psychiatric history disclosed
- I refer elsewhere
- Other (open ended text response)

One question about the Australian Government mental health treatment plans was included because it is significant to discussions around psychiatric illness and accessing long term psychosocial support for patients in Australia. In section III respondents' attitudes toward psychiatric genetic counseling were examined (1= *strongly disagree*, 2= *disagree*, 3= *neither agree or disagree*, 4= *agree*, 5= *strongly agree*). Questions explored genetic counselors' confidence in providing a risk assessment for an individual with a history of psychiatric illness, confidence in the data relating to the value of psychiatric genetic counseling, interest in attending further educational opportunities and in providing psychiatric genetic counseling in the future. Survey data were collected and managed using REDCap (Research Electronic Data Capture) electronic data capture tools (Harris et al. 2009) hosted at The Royal Melbourne Hospital.

2.3 Procedures

Between 1st March 2022 and 31st May 2022, an invitation to participate in this survey was distributed to genetic counselors across Australia and New Zealand through the ASGC listserv. At the time there were 397 active members. Study investigators were excluded from participation, therefore a total of 393 members received the study invitation. This email comprised a brief description of the study, a hyperlink to the anonymous online survey, and contact details for the researcher.

2.4 Data Analysis

Descriptive statistics (means, medians, and frequencies) were calculated as appropriate for close-ended responses to the online survey. Fisher's exact test of independence was used to check for significant associations ($p \leq 0.05$) between pairs

of variables. Free text responses are presented to provide context to open-ended questions (Table 2).

3. RESULTS

A total of 44 surveys were completed, yielding a response rate of 11% (N=44/393). Demographic data are listed in Table 1. Most respondents practiced within Australia, in either Victoria or New South Wales. One genetic counselor practiced in New Zealand. Respondents' genetic counseling experience ranged from less than 5 years to greater than 20 years, with a median of 5-9 years' experience. None of the respondents were working in psychiatric genetic counseling at the time of survey completion. 48% of respondents identified as certified genetic counselors, while 52% identified as having yet to complete certification (Members of the Human Genetics Society of Australasia (MHGSA) genetic counselors).

While none of the respondents practiced in psychiatric genetics as their speciality area, nine percent of respondents counseled between 1 and 4 patients per month where the primary indication is a personal and/or family history of psychiatric illness (n=4/44), and nine percent counseled one or more patient per month where genetic testing for psychiatric illness was facilitated. Of the four genetic counselors providing counseling for patients with a primary indication of psychiatric illness, two responded that they counseled patients where genetic testing for psychiatric illness was facilitated. Speciality areas of genetic counseling practice of those providing counseling in this area, included; general (n=2/44), prenatal (n=1/44) and cancer (n=1/44).

When questioned whether they routinely asked patients about personal and/or family history of psychiatric disorders, the majority reported they 'sometimes' asked (48%, n= 21/44) but 34% 'rarely' asked (n=15/44). Statistical analysis did not reveal any significant association between genetic counselors' years of experience, and/or certification level and likelihood to ask about psychiatric history ($p \geq 0.05$). When asked about addressing general mental health support for patients for any indication, over half of those surveyed reported they discussed the option of referral for a Medicare-rebated mental health treatment plan 'sometimes' with patients (55%, n=24/44), whilst 23% 'rarely' (n=10/44) or 'never' discussed this (5%, n=2/44). Despite the majority of respondents reporting they do not counsel patients where the primary indication is a personal and/or family history of psychiatric illness (91%, n=40/44), 64% 'sometimes' had patients disclose a personal and/or family history of psychiatric illness *secondary* to their primary reason for referral (n=28/44), and 25% 'usually' had patients disclose psychiatric illness (n=11/44).

Participants in the survey were asked, "If a personal and/or family history of psychiatric illness is disclosed during an appointment where psychiatric illness is not the primary reason for referral, how would you typically address it?" Twenty three percent of respondents responded they 'do not address it as it is not the primary reason for referral' (n=10/44), while 25% provide genetic counseling in relation to the psychiatric history disclosed (n=11/44). Eighteen percent of respondents reported they refer elsewhere (n=8/44), or rebook for a dedicated psychiatric genetic counseling appointment (5%, n=2/44). Thirteen participants entered open-ended responses in the free text box, which provided additional context to the quantitative data (Table 2).

Participants were asked about their level of agreement and/or disagreement with the statement "There is sufficient data to suggest that a risk assessment and genetic

counseling are of value in psychiatric illness”. Responses varied from strongly agree (16%, n=7/44) to disagree (11%, n=5/44). Whilst 41% of those surveyed agreed there was sufficient evidence, 32% responded ‘neither agree nor disagree’ (n=14/44). The majority of participants do not feel confident in providing an individual with a psychiatric risk assessment, with 59% (n=26/44) disagreeing and 14% (n=6/44) strongly disagreeing to: “I feel confident providing a risk assessment for an individual with a personal and/or family history of psychiatric illness”. Whilst, 22% ‘neither agree nor disagree’ (n=10/44). We asked participants if they would be interested in attending education sessions on psychiatric genetic counseling in the future (including web based learning), and an overwhelming 95% agreed (61% strongly agreed (n=27/44) 34%, agreed (n=15/44)) to being interested in future education opportunities. Finally, the majority of respondents either ‘strongly agree’ (32%, n= 14/44) or ‘agree’ (46%, n=20/44) to being interested in providing psychiatric genetic counseling as part of their practice. Statistical analysis did not reveal any significant association between genetic counselors’ years of experience and/or certification level and genetic counselors confidence in providing psychiatric genetic counseling ($p \geq 0.05$).

4. DISCUSSION

Despite the high prevalence of psychiatric disorders in Australia, this current study provides evidence that psychiatric genetic counseling referrals are uncommon, specialised clinics are lacking, and genetic counselors often do not ask about and/or lack confidence in addressing psychiatric disorders. Several studies have demonstrated benefit of genetic counseling in this context, regardless of the limited utility of molecular genetic testing (Costain et al. 2014a; Costain et al.; 2014b; Hippman

et al. 2013; Inglis et al. 2015; Moldovan et al. 2019). Participants with psychiatric disorders interviewed by Semaka and Austin (2019) reported that they found genetic counseling empowering through providing a new understanding of the cause and management of their disorder, reducing feelings of shame, blame and guilt, and improving family communication about their illness. Costain et al. (2014a; 2014b) showed that introducing a genetic counseling intervention in a community mental health setting had benefits in reducing patients' and family members' over-estimation of risk of schizophrenia, and decreasing stigma. Inglis et al. (2015) demonstrated that accessing specialised psychiatric genetic counseling improved outcomes of empowerment and self-efficacy at one month follow up for those with a personal history of a psychiatric disorder and increased empowerment in those with a family history of a psychiatric condition.

Despite the high demand and acceptability of PGC among individuals with psychiatric conditions, this current study provides evidence that a family history of psychiatric disorders is only discussed if raised by a patient during a session for another indication; and it rarely constitutes the reason for primary referral, which is a significant barrier to PGC. This may reflect either a lack of awareness among healthcare professionals such as psychiatrists, general practitioners and mental healthcare professionals about the relevance or availability of PGC, or a lack of awareness about how to access PGC services (Hunter et al. 2010). A second major barrier to PGC identified is that genetic counselors lack confidence in providing PGC. Other studies have suggested barriers to providing PGC may relate to: (1) limited training in PGC, (2) stigma associated with mental illness, (3) balancing existing workload demands, (4) lack of genetic testing and (5) multifactorial nature of these psychiatric illnesses (Austin et al. 2008; Austin and Honer 2005; Booke et al. 2020; Inglis, et al. 2015; Low

et al. 2018; Moldovan et al. 2017; Richter et al. 2022; Rowlatt et al. 2022). These barriers likely contribute to genetic counselors low confidence in the provision of PGC, and warrant further investigation in the local setting.

Genetic counselors are well placed to directly increase access to genetic counseling for psychiatric disorders by asking patients referred for other indications about personal and/or family psychiatric history and making follow-up appointments/referrals as appropriate (Booke et al. 2020). Previous studies have found between 35.5% and 40% of surveyed genetic counselors 'rarely' or 'never' asked about psychiatric conditions when taking a family history (Booke et al. 2020; Monaco et al. 2010) and this survey similarly found 36% of genetic counselors "rarely" or "never" asked about personal and/or family history of psychiatric conditions. This data adds weight to previous studies encouraging genetic counselors to routinely ask about personal and/or family psychiatric history in their daily interaction with patients (Low et al. 2018).

Genetic counselors in Australia have a high level of interest in receiving further training in PGC, and this is consistent with previous studies in Canada, UK and other countries (Austin et al. 2008; Austin and Honer 2005; Booke et al. 2020; Jenkins and Arribas-Ayllon. 2016; Low et al. 2018; Monaco et al. 2010; Rowlatt et al. 2022; Zhou et al. 2014). Improving the training of genetic counselors through more frequent professional development in psychiatric genetics, and more active opportunities to practice PGC (e.g. internships) has been shown to result in genetic counselors feeling more prepared to provide PGC (Dillon et al. 2022; Low et al. 2018; Van den Adel et al. 2022). PGC training and development opportunities for Australian genetic counselors have thus far been limited (Moldovan et al 2019). The strong interest (95%) in training in PGC in this study indicates that uptake of educational opportunities are likely to be

high, which would allow the outcomes of various educational formats to be evaluated and refined (Low et al. 2018).

It is within the scope of genetic counselors practice to be able to identify patients who could benefit from long-term psychological support, and identify patients who may benefit from an Australian Government Mental Health Treatment Plan (MHTP). However, there are currently no training methods or guidelines for when genetic counselors should make referrals to mental health services, or how to discuss MHTP with patients (Hayes et al 2022). Over half of those who responded, reported that they 'sometimes' discussed MHTP with their patients suggesting Australian genetic counselors feel comfortable addressing the topic of psychiatric disorders in the context of accessing long term psychological support. However, a quarter of respondents' had 'never' or 'rarely' discussed MHTP with patients. Further investigation of genetic counseling practice regarding mental health screening and referral to mental health providers would provide additional insight into genetic counselors' practice of referring patients, and factors that influence their decisions to refer and/or discuss MHTP.

It is not clear where, or to whom respondents referred when a personal and/or family history of psychiatric illness is disclosed in session particularly given there are no dedicated or specialist PGC services in Australia; and further research would be useful to gain understanding of onward referrals (Inglis et al. 2015). This finding that many genetic counselors choose to refer on for PGC is consistent with a previous Australian study, which found genetic counselors were significantly less likely to believe that it was their role to address psychiatric genetic information compared with medical geneticists and psychiatrists (Zhou et al. 2014). Understanding where Australian genetic counselors draw the line between feeling they are able to address the topic versus referring elsewhere is unknown. Further studies exploring factors which prompt

referral, and perceived differences between scopes of practice would provide further insight into the professional development needs of Australian genetic counselors.

We hope this study will prompt further awareness, education and research on addressing psychiatric disorders within genetic healthcare as well as initiating steps to establish specialised clinics and referral pathways for PGC in the region.

5. STRENGTHS AND LIMITATIONS

A limitation of this study was that only a small proportion of practicing genetic counselors (e.g. approximately 11% of the ASGC membership) participated; therefore it cannot be said to represent the views of all genetic counselors practicing in Australia. Furthermore, only one participant from New Zealand responded, therefore the findings do not represent the views of genetic counselors practicing in New Zealand. Additionally, participants of our survey may differ in important ways from non-participants. For example, there is a possibility that participant's greater interest in this topic than non-responders. This study may limit the generalizability of the findings.

6. IMPLICATIONS FOR PRACTICE

It is hoped the results of the current study create an awareness of PGC among practicing genetic counselors in Australia and elsewhere. The study highlights a major gap in genetic healthcare regarding PGC, despite the high frequency of these conditions in family histories in the general population. A direct practical implication is that genetic counselors are encouraged to ask about psychiatric conditions when gathering family histories, thus providing opportunities for patients to express questions or concerns, and opening a door for access to PGC. Another implication of this study is in highlighting a need for further genetic counselor education opportunities in PGC, in an ongoing way. Finally, growing evidence from other countries highlights

the potential benefit of dedicated clinics for PGC, and genetic counselors are well placed to advocate for this.

7. FUTURE RESEARCH

Further research is needed to explore and address the disconnection between genetic counselor perceptions of the high value of PGC and interest in providing it, yet a lack of confidence in asking and addressing this topic in practice. Studies to implement and evaluate professional education including applying theory and skills in practice will be useful in optimising and sustaining upskilling in this area. Research aimed at developing educational interventions in community (e.g. support organisations), primary (e.g. general practitioners) and specialist (e.g. psychiatry) healthcare settings to increase referrals to genetic services regarding psychiatric conditions is needed. Increased referrals will demonstrate demand and further prompt implementation at the genetic service level of models of care specific to this area of genetic counseling. Studies including patient reported outcome measures will be important in evaluating and improving models of incorporating PGC into general genetic counseling practice and into specialised PGC clinics within genetic healthcare. Longitudinal consideration of outcomes will be valuable in measuring efficacy.

8. CONCLUSION

This study provides valuable insight into the clinical practice of PGC in Australia highlighting a lack of dedicated PGC clinics, and a low level of confidence in providing genetic counseling on this topic, despite the prevalence of psychiatric conditions in the population. These findings provide a basis for initiating and trialling ongoing education in this area for genetic counselors, and for the development of specialised PGC clinics in the region.

Authors Contributions

Authors JI, AS and LF confirm that they had full access to all the data in the study and take responsibility for the integrity of the data and the accuracy of the data analysis. All of the authors gave final approval of this version to be published and agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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COMPLIANCE WITH ETHICAL STANDARDS

Approval to conduct this research was obtained by the Melbourne Health Human Research Ethics Committee (HREC2021.284). All procedures followed were in accordance with the ethical standards of the responsible committee.

Implied consent was obtained from all participants included in the study.

CONFLICT OF INTEREST

Authors JI, AS, LF, PJ, JD, JT, JA and IW declare that they have no conflict of interest.

ANIMAL STUDIES

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

DATA SHARING AND DATA ACCESSIBILITY

Data are available upon request from the authors.

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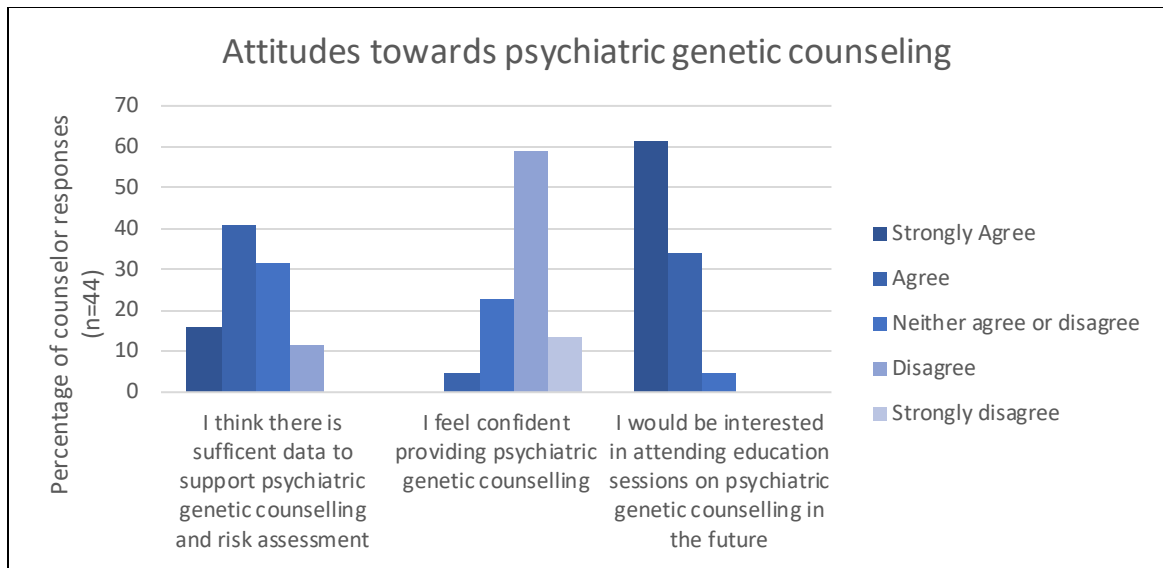


Figure 1: Participant (%; n=44) attitudes towards psychiatric genetic counseling.