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When to break the news and whose responsibility is it? Genetic health professional focus groups on disclosure of BRCA genetic cancer risk

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1. ABSTRACT

Objectives: Disclosure of a hereditary condition in the family poses notable challenges for patients, who often seek the assistance of genetic health professionals (GHPs). This study aimed to investigate GHPs' opinions about the ideal time for disclosure to offspring and their responsibility to at-risk relatives.

Method: GHPs (N=73) from genetic clinics covering all states of Australia participated in semi-structured focus groups and interviews to explore the topic of disclosure to at-risk offspring, using *BRCA1* and *BRCA2* families as an example. Transcriptions were analysed thematically.

Results: GHPs perceived that life-stage, maturity, parents' knowledge, and capacity to disseminate information influenced parent-child disclosure. In general, GHPs recommended early informal conversations with offspring about a family illness. GHPs considered that facilitation of disclosure to relatives using counselling strategies was their responsibility, yet there were limitations to their role (e.g., legal and resource constraints).

Variability exists in the extent to which genetic clinics overcome challenges to disclosure.

Conclusions: GHPs' perspective towards the ideal time for disclosure is generally dependent on the patient's age and relative's ability to disclose information. A responsibility towards the patient and their at-risk relative was widely accepted as a role of a GHP but views vary depending on legislative and specialty differences. Greater uniformity is needed in genetic procedural guidelines and the importance of each discipline (e.g., geneticists, oncologists, genetic counsellors, nurses, and psychiatrists/psychologists) in genetic clinics to manage disclosure challenges.

Strengths and limitations of this study

- This study provides one of the largest cohort of Australian genetic health professionals, with a detailed, in-depth approach to responsibility and confidentiality concerns.
- The findings extend on previous literature by focusing on two major genetic disclosure issues: ideal age of disclosure and the extent to which health professionals are responsible to warn at-risk relatives of their risk.
- The study was limited to focus primarily on the disclosure of BRCA1 or BRCA2 genetic test results.
- A qualitative approach to understanding the challenges of disclosure limited generalisability but highlighted the variability in clinical practices across different legislative contexts and a need for clearer policies and role definitions.

Keywords: duty to warn; genetic privacy; genetic testing; genetics; disclosure; ethical issues

2. BACKGROUND

Identifying a *BRCA1* or *BRCA2* (hereafter *BRCA1/2*) pathogenic variant in the family and informing relatives can be a challenge, since most people do not want to be a 'bearer of bad news'. Yet, such information can have far-reaching implications for a relative's decision regarding risk management, lifestyle, and family planning.

Genetic health professionals (GHPs) often work with families trying to navigate parent-child communication about genetic risk, commonly around when, how and what information to give, particularly the ideal age for disclosure (Metcalfe, Coad, Plumridge, Gill, & Farndon, 2008). Factors such as age, gender and the type of genetic condition can influence a child's understanding of genetic information (Rowland & Metcalfe, 2013; Vears, Delany, Massie, & Gillam, 2016). GHPs are also concerned about non-disclosure to children, which does occur, albeit less commonly than to extended relatives (Aktan-Collan et al., 2011; Healey et al., 2017). Reasons for non-disclosure include: parental guilt, fear of burdening others and a relatives' inability to cope (Healey et al., 2017). Approximately one third of patients want GHP involvement during family communication (Aktan-Collan et al., 2011; Pentz et al., 2005), especially when families are emotionally and geographically distant (Pentz et al., 2005). Offspring have also reported a preference for GHPs to disclose a hereditary condition in the family as opposed to parents (Klitzman, Thorne, Williamson, Chung, & Marder, 2007). Yet, the extent of GHP responsibility for ensuring appropriate disclosure is a matter of debate.

Despite the prevalence of studies exploring family communication of genetic information (Gaff et al., 2007), very few studies explore GHPs' opinions on their responsibility in disclosure. A recent systematic review (Dheensa, Fenwick, Shkedi-Rafid, Crawford, & Lucassen, 2016) found that across eight countries and varying heritable illnesses, GHPs generally felt some sense of

responsibility to inform their patients' relatives about their genetic risk. Yet, a range of moral, legal, and practice-related arrangements reportedly made it challenging to act on their perceived responsibility. Of the nine studies in the review, none specifically explored GHPs' responsibility towards patients' relatives. A recent study therefore, aimed to address this gap by conducting focus groups with UK GHPs (Dheensa, Fenwick, & Lucassen, 2017). GHPs in the UK were concerned about the difficulty in distinguishing between genetic and personal information therefore potentially breaching confidentiality through disclosure and more broadly, reported a need for national consensus on following the UK guidelines from the Joint Committee on Medical Genetics. According to these guidelines, GHPs explore family relationships, encourage family communication, and assume that responsibility of disclosure lies with the patient.

Under the legislative guidelines of some countries, when patients do not provide consent for the disclosure of genetic information, GHPs can make contact with at-risk relatives. Across 10 countries, there are eight that accommodate exceptions to confidentiality, with Australia, Canada, Israel and Japan providing explicit circumstances surrounding disclosure without consent (i.e., serious, treatable or preventable) (Wolf et al., 2015).

Unlike the UK, Australia has clearer guidelines on genomic disclosure, but with elusive governance. According to current Australian guidelines from the National Health and Medical Research Council, a GHP can disclose genetic information to an at-risk relative without the patient's consent in specific circumstances. This exemption applies for "incurable" conditions which are "preventable" or include "treatable manifestations" (e.g., depression), in which "specific management" or "treatment" can "lessen or prevent" the threat of disease or distress National Health and Medical Research Council (2014; p.42)**.** Nevertheless, both Australian and UK guidelines strongly encourage GHPs to take reasonable steps to obtain consent and consider

the potential consequences of disclosure when consent is not provided (National Health and Medical Research Council, 2014; Royal College of Physicians Royal College of Pathologists and British Society for Human Genetics, 2011). However, there is a lack of uniformity across Australia in how these guidelines are followed and upheld in clinical practice (Otlowski, 2015). South Australian (SA) genetic services (Suthers, Armstrong, McCormack, & Trott, 2006), for example, provide family letters to at-risk relatives to inform them of an increased risk, with the patient's consent but without the recipient's consent, whereas the rest of Australia do not make provisions for direct contact with relatives. The extent to which Australian GHPs within public hospitals consider it their role to assist families with disclosure is currently unclear. The purpose of the current study was to understand the role of genetic health professionals in assisting families with disclosure of genetic cancer risk. Specifically, two research questions guided the study: (1) When is the best time to tell offspring about their genetic risk? and, (2) Who is responsible to inform relatives of their genetic risk?

3. METHODS

Patient and Public Involvement statement: The study involved GHPs but no patients.

Study participants and recruitment: Eligible GHPs (e.g. geneticist, genetic counsellors, medical specialists, nurses, surgeons, and psychiatrist/psychologists) who had worked with *BRCA1/2* families from familial cancer centres within all Australian states. A PhD candidate with several years' experience in qualitative research (A.L.Y.) presented the study aims to GHPs during family cancer clinic meetings and/or emailed study details. Interested GHPs were then re-contacted to arrange a suitable time for participation in a focus group or interview. Recruitment continued until theoretical saturation was achieved (Namey et al., 2016). Informed consent was obtained from all participants and the study has the approval of institutional ethics committees.

Procedures: GHPs completed a questionnaire primarily collecting demographic data, and then took part in a focus group or interview. Focus groups (2-8 individuals) were held in-person or via video-teleconferencing in 2017, during familial cancer clinic meetings or at a time convenient for participants. Semi-structured telephone or face-to-face interviews were completed with participants unable to attend a focus group. Interviews and focus groups ranged in duration from 15-77-minutes, depending greatly on the time-availability of participants.

Data analysis: Focus groups and interviews were transcribed verbatim and underwent data-driven analysis by three authors (A.L.Y., P.N.B., R.W.) guided by thematic analysis (Braun & Clarke, 2006) using NVivo 11 computer software to map themes. Consideration was given to whether individuals participating in one method (i.e., focus groups) differed in relation to the experiences discussed in the other method (i.e., interviews). Emphasis was placed on the themes mentioned by the majority of participants and data that raised novel lines of inquiry, reflecting unique sub-themes (e.g., rurality, specialists). Three authors (A.L.Y., P.N.B., R.W.) analysed the first six transcripts by re-reading each transcript, generating codes, and developing overall themes which were then organised into a thematic 'map'. Differences in coding were resolved by consensual discussion. Subsequent transcripts were analysed according to the 'map' resulting in a final set of themes. Focus group or individual interview identification (e.g., FG4 or II4) are provided below.

4. RESULTS

Sample characteristics: Of 91 eligible GHPs invited, 73 consented and participated in the study. Demographic characteristics are provided in Table I.

4.1. When is the best time to tell offspring about their genetic risk?

4.1.1. Offspring-dependent: GHPs had different arguments for and against disclosing genetic status to offspring earlier than 18, at the age of 18 and at the age of 25 onwards. Most GHPs reported that earlier was better to allow time for the offspring to adjust to, process and research information about their genetic risk before make decisions about medically mitigating their risk. Informing children as the conversation arises, in an age-appropriate manner, was commonly encouraged: "*in an ideal situation it should be a progressive discussion over time*" (FG1). GHPs argued that if information about genetic risk is withheld, offspring might hear it inadvertently from relatives and through GHPs during unrelated appointments, placing strain on parent-child relationships. A few GHPs said that informing children in their mid-teenage years (15-17 years old) was ideal: "*probably mid-teens, and the reason why [is] to be aware [that] they can be breast aware, not breast alarmed and breast paranoid"* (II15). Planning to inform children at the age of 18, or "*saving it up as an 18th birthday present*" (FG9), was considered unhelpful and described as *"dropping the bombshell*" (FG3).

In contrast, some GHPs felt that disclosure should be related to **when it could inform testing/screening behaviour or decision-making**, and therefore advocated disclosure at an older age (>20 years)**.** The recommended breast screening age for *BRCA1/2* carriers is 30 within Australia (Cancer Institute NSW, 2018). In relation to the patient's mental health, some GHPs said that disclosure at a young age can lead to prolonged worry, since the time between disclosure and screening is longer compared to their older counterparts. However, others disagreed with this stance stating that older patients can be more anxious if their parents informed them later due to the immediacy of action needed to mitigate risk. Some GHPs noted that parents may not be alive when their child reached the recommended age for screening or testing, and therefore disclosure, and possibly testing, should occur earlier. Subsequent, shifts in

opinions were discussed, "*it used to be you don't do anything until you're going to use the information. And my original teaching was that you don't do genetic testing till a month before they're due to start screening…we now know that that isn't necessarily the best way of offering genetic testing*" (FG1).

Other reasons some GHPs advocated disclosure **at an older age** were that young adults are considered generally more mature and receptive towards genetic information than younger offspring. Furthermore, the parent-child relationship is likely to change to an adult-adult pattern of relating by older ages, which can be considered *"on a more even level"* (FG4), allowing the younger person to be more autonomous in their responses to genetic information and testing decisions. Disclosure at an older age was considered advantageous to avoid having the child incorporate the pathogenic variant into their identity. A final justification for withholding information till adulthood was the potential for inappropriate medical management of young adults by GHPs in response to anxious, insistent younger adults: "*they can be inappropriately managed if they're aware of this information from a young age…privately…[and] publicly… a breast surgeon will often screen younger women*" (FG7).

Some GHPs recommended disclosure **at key points related to genetic risk** when the child was in any case probably aware of health problems and emotional distress in the family, such as when a parent was diagnosed and being treated for cancer, or having surgery.

Other GHPs said they **did not recommend a time or age for disclosure**, rather they spoke about taking a case-by-case approach to families, taking into account the unique characteristics of each individual, life experiences and family dynamics.

4.1.2. Parent-dependant: Some GHPs emphasised that disclosure should depend on the parent's decision about when and how they want to tell their children: "*parents know their child*

best of all and they would be in the best position to judge" (FG22). Parents can use their intuition to decide on the timing of disclosure and skilfully navigating around stressful events (e.g., cancer diagnosis or anniversaries). Yet, other GHPs felt that some parents' negative experience with genetic services could hinder timely and effective disclosure. For example, "*it's often a red flag when you have someone who's not [coped with the testing process]…if they've got a lot of emotional turmoil going on they kind of can project that and expect that their children will react the same way and perhaps think that their kids can't cope"* (FG4). GHPs also reported that parents may not understand the seriousness of sharing hereditary cancer information with families, may have forgotten about their results from a research study, and can potentially still be trying to process the information for themselves.

4.2. Who is responsible to inform relatives about their genetic risk?

4.2.1. GHPs are responsible to facilitate and support family communication: Many GHPs agreed that they were responsible for facilitating family communication by using a range of strategies to support probands (i.e., during diagnostic testing) and relatives (i.e., during predictive testing) to talk to their children and relatives (See Table II for full list of strategies). Factsheets were referred to most often as a resource to provide probands to assist with disclosure but was not considered user-friendly or too generic by some. GHPs also admitted to treating families differently depending on the type of test, "*I'm probably not as active in my making sure the information gets out there with the predictive*[1] *as I am [with probands who are the first individual with a pathogenic variant to be identified in the family]*" (FG7).

GHPs reported that assessment of family communication processes should start early in the consultation; for geneticists/genetic counsellors this involved pretest counselling, for nurses/psychologists this was during the first consultation and for oncologists this was at the time

of talking about genetic testing to cancer patients. Assessment included exploring family dynamics (e.g., estrangement, lack of communication), emotional responses that could impede communication (e.g., guilt, fear) and assessing the proband's ability to disclose information to relatives (e.g., clarify their understanding, coping skills). Building rapport with the patient is an important "*initial foundation*" (FG20) to help patients with their communication with families. GHPs advocated for gently preparing probands for the possibility that they could have a positive test result, and if so, to consider to whom, what and when they would disclose their results. When parents were finding it difficult to communicate to their children, GHPs offered to have the offspring join the parent's consultations, or provide subsequent over-the-phone consultations with the offspring or a separate consultation for the offspring to obtain more information. Family group consultations were also recommended to facilitate communication and address concerns with the relevant relatives present. Such consultations allow all members of the family to be informed simultaneously. Having another family member can lead to greater clarification of information: "*someone [can] obviously pick something up but they then explained it in a way that helps*" (FG21). However, some GHPs were also concerned about the practicality of implementing family group consultations in the current public health model. In some cases when disclosure in families were not occurring, some GHPs were willing to "*take it upon themselves" to see that the adult children involved [were] informed of their risk, "So you're protecting the rights of the child as well*" (FG12).

4.2.2. Different clinics, different responsibility: The work culture, resources and expectations within particular genetic services influenced GHPs' views about disclosure to relatives: "*the scope of your role changes with whatever clinical service you are working with*" (FG2). Some clinical teams placed greater emphasis on disclosure to at-risk relatives. For example, working

within high-risk clinics provided some opportunities for nurses to explore disclosure, whereas some genetic counsellors reported, "*working in a busy clinical service in the public system really limits us in terms of our capacity of what we can do"* (II14). Emphasise was placed on young at-risk relatives' personal responsibility, with one geneticist saying, *"It's going to become too big for familial cancer centres to be able to hold onto these families and do the follow-up. I think it's going to have to shift out to personal responsibility*" (FG10).

4.2.3. Ultimately it is the families' responsibility: Families were considered ultimately responsible for what they want to do with their own medical information. Confidentiality and autonomy were upheld by GHPs and if a patient choose to be private, this was respected. Some felt it was not their role to ensure disclosure beyond providing a family letter.

Conversely, others were of the mind that they would like to assist families with communication but were limited by time constraints and procedural barriers. Some GHPs believed GHPs should not feel guilty if disclosure did not occur in a family particularly since families do not always tell GHPs the truth and are unwilling to discuss family dynamics. Some familial issues are beyond a medical GHP's capacity (or consultation time) to discuss and requires psychological assistance. For example, a genetic counsellor said, *"Sometimes I think whatever's going on in their families is beyond what we as genetic counsellors can actually help with, which is unfortunate, but…considering the workload…you can only pour so much of your energy into one family"* (FG20). Another common limitation GHPs discussed was having no control over what happens after a consultation, "*you've got no control over what's passed on and what isn't or how it's passed or whether facts and figures [are] mixed up*" (FG10). Moreover, GHPs were also aware that advocating for disclosure was not beneficial in all cases, "*it's important to be aware of the fact that there could be positives and negatives [in] telling, but also positives and negatives in*

not telling" (II18). Situations in which information is withheld or difficult to navigate include cases when an at-risk relative has a mental health concern and/or cognitive disability.

5. DISCUSSION

The age at which disclosure should ideally begin is not a concern of *BRCA1/2* families alone but is common amongst families with a hereditary condition (Rowland & Metcalfe, 2013). GHPs in this study recommended optimal timeframes for disclosure of genetic risk to offspring, with the majority favouring early disclosure tailored to individual circumstances. Hereditary cancer can be introduced into the family story with a simple explanation about genetics, cancer and the benefits of testing (Werner-Lin, Merrill, & Brandt, 2018). Similarly, families with Cystic Fibrosis normalised their condition by informing children that "everyone possesses disease causing genes" (Cavanagh, Compton, Tluczek, Brown, & Farrell, 2010; p. 206). This method of dissemination is modelling to children that coping and adjustment to such information is possible. Having more time to process, talk and ask questions during casual conversations is less anxiety-provoking than being informed unexpectedly at an age when immediate medical action is required (Dennis, Howell, Cordeiro, & Tartaglia, 2015).

According to Klitzman and colleagues (2007), the reasoning behind GHPs' perspective about the ideal age for genetic testing and subsequent disclosure can fall under two categories: 1) the *life stage* or maturity of the child and 2) the *medical time course* and benefit of the information at a given time. Age-appropriateness was a key feature of early disclosure. According to Piaget's theory of cognitive development, children at approximately 11 years old reach the stage of 'formal operational thought', at which hypothesis testing and abstract reasoning develop (Piaget, 1964). In theory, children at this stage can make inferences that if their parent is ill, then they too could become ill with the same illness (Biba ce & Walsh, 1980; Metcalfe, Plumridge, Coad,

Shanks, & Gill, 2011). Thus, parents will benefit from considering their offspring's cognitive and emotional capacity before informing them about their risk (Werner-Lin et al., 2018), which may have different developmental trajectories depending on the temperament of the offspring (Perlman & Pelphrey, 2010). Parental consideration of disclosure of genetic status with young adults involves consideration of poignant life-stage changes or communicating at certain junctures (e.g., impending marriage or pregnancy (Gaff et al., 2007)). Parental capacity to inform offspring (Cavanagh et al., 2010; Rowland & Metcalfe, 2013) and their own experience or level of satisfaction with genetic testing may hinder communication (Lieberman et al., 2018), warranting the facilitation of communication by GHPs (Peshkin, DeMarco, & Tercyak, 2010). GHP's facilitation of disclosure is generally agreed to be incorporated in their clinical practices (i.e., Table II), expounding upon previously reported strategies (Dheensa et al., 2017). Contrastingly, the opinions of some GHPs suggest that facilitation of disclosure is a peripheral requirement of their practice, other than providing a family letter to be passed from patient-to-relative. Patient autonomy, confidentiality and/or the law can also contribute to the reluctance to facilitate disclosure (Dheensa et al., 2016). Follow-up calls/appointments to address disclosure is considered worthwhile to revisit the topic of disclosure with families, particularly when legislative changes occur (Derbez, de Pauw, Stoppa-Lyonnet, & de Montgolfier, 2017), yet resource and time constraints can make this impracticable (Forrest, Delatycki, Curnow, Skene, & Aitken, 2010).

In SA, the genetic services send letters directly to at-risk relatives, with the patient's consent. Other research has shown benefits from direct contact with at-risk relatives (Schwiter, Rahm, Williams, & Sturm, 2018). Studies involving a range of illnesses (e.g., *BRCA1/2*, Lynch, Cowden Syndrome), have shown that when GHPs mediated contact, uptake of testing was

greater amongst at-risk relatives compared to when contact was patient-mediated (Sermijn et al., 2016; Suthers et al., 2006). A Western Australian study adopted a cascade screening process derived from an Australasian model of care for familial hypercholesterolaemia in accordance to local and national guidelines. Nurse-led initiation of contact with at-risk relatives, despite non-consent from probands, was effectively completed (Bell et al., 2014). Recently GHPs working in French genetic clinics are legally permitted to offer a written document informing at-risk relatives of their risk, yet guidance about to whom this requirement extends to and how GHPs responsibility will be defined remains elusive (d'Audiffret Van Haecke & de Montgolfier, 2018).

In this study, we found that GHPs' opinions regarding their responsibility towards at-risk relatives differed depending on four factors. First, a GHP's role and opinion were informed by the attitudes and expertise of the genetic clinic in which they worked, which varied between local health districts and states. Second, GHPs in each speciality were governed by their own legislation and ethical guidelines, including the overall framework and ethos in which they practice, which emphasised family communication to a lesser or greater extent depending on the profession (e.g., genetic counsellor vs. medical oncologist). Third, GHPs generally reported a greater need to facilitate communication for probands who are the first in the family to be identified as a carrier since the burden of sharing information appears greater than those who have cascade testing, and therefore can also be dependent on whether the patient has cancer or not. Fourth, state-wide differences in health provision and legislation meant that clinics in SA can send genetic letters to relatives effectively (Suthers et al., 2006), but GHPs in New South Wales are still cautious of potential litigation issues despite amendment of privacy principles in attempt to uniform Australian genetic practices (Otlowski, 2015).

GHPs working in genetic clinics, many of whom are genetic counsellors, are governed by their respective professional guidelines and respective health district legislation. 'Non-directiveness' is a term used to describe GHP practices that are patient-centred and uphold the autonomous decisions of the patient (Elwyn, Gray, & Clarke, 2000). Yet, within the context of cancer genetics in which evidence-based surgical treatments exist that effectively mitigate risk, adherence to such principles is questionable (Koch & Nordahl Svendsen, 2005). The Task Force of the National Society of Genetic Counsellors has consequently excluded the term 'non-directiveness' in their definition of genetic counselling, emphasizing instead on educating patients about testing implications for themselves and relatives (Resta, 2006). A shared decision-making approach is currently favoured (Forbes Shepherd, Browne, & Warwick, 2016).

Greater clarity is needed on the definition of 'at-risk relatives', and the extent to which GHPs are responsible to inform them. With the rise of genomic medicine and the subsequent need to educate the general public of the potential benefits and limitations of such knowledge, the question of who is responsible to inform the public is a wider healthcare concern. Contrary to the opinions of a small minority of GHPs in the current study who predict a shift to personal responsibility when managing at-risk relatives, the European Breast Cancer Council (Rutgers et al., 2019) argues that the healthcare system will need to rise to the challenge and support future families in obtaining high-quality and timely information. It is already evident that without the input of cancer genetic clinics, at-risk relatives are not considering the potential limitations of a proposed test before choosing direct-to-consumer genetic testing (Roberts et al., 2017). The lack of guidance and advice from medical professionals about their genetic risk (Roberts et al., 2017), further reinforces the need for genetic cancer clinics to clarify their responsibility for at-risk relatives.

Practice Implications: A multidisciplinary approach to genomic medicine has been proposed to be effective in tackling the challenge of disclosure (Derbez et al., 2017; Metcalfe, 2018). GHPs are currently supporting young at-risk relatives within high-risk clinics in Australia, but can also potentially allow for ongoing support of families struggling with disclosure difficulties. Funding regulators are to emphasis fiscal and institutional backing of genetic clinics in order to sustain a multidisciplinary team and to manage the future role of GHPs in the preventive health of their patient's relatives.

Limitations: This study focused primarily on GHPs' view of *BRCA1/2* families' disclosure of genetic risk. Nevertheless, the topics covered were broad enough to have applicability and transferability to other adult-onset genetic health conditions (e.g., Familial Hypercholesterolaemia) in which parents are also faced with the dilemma of disclosure, and in some cases they have similar consequences and risk-management options available (e.g., Lynch syndrome). Genetic counsellors (80%) provided the most input into discussions and therefore their practices were emphasised the most in the results. Further research is required, with larger samples of geneticists, surgeons, nurses and psychologists in order to elucidate whether their opinions about their role in disclosure differs.

Conclusion: This Australian study reports on the ideal age of parent-child disclosure of genetic risk and GHPs' responsibility towards at-risk relatives. Our findings highlight the need for clearer policies regarding GHP's responsibility to relatives and to the community in terms of preventive health, including the need for more staff and fiscal support to sustain disclosure initiatives (e.g., direct-contact letters).

Footnotes:

Predictive testing is the testing of a relative of after a pathogenic variant has already been identified in the proband.

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CONFLICT OF INTEREST

A.L.Y., P.N.B., K.M.T., C.E.W., E.H., and R.W declare that they have no conflict of interest.

ETHICS APPROVAL

The project was approved by the University of Sydney Human Research Ethics Committee (HREC 2017/011) which covered the participating hospitals.

INFORMED CONSENT

All procedures followed were in accordance the University of Sydney Human Research Ethics Committee) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Signed or electronic consent was obtained from all participants. Informed consent was obtained from all patients for being included in the study.

AUTHOR'S CONTRUBUTION STATEMENT

A.L.Y made a substantial contribution to the study design, acquisition of data, analysis, interpretation, revisions for important intellectual content.

P.N.B. made a substantial contribution to the conception, acquisition of data, analysis interpretation and drafting the work for important intellectual content.

K.M.T. made a substantial contributed to the study design, acquisition of data, analysis interpretation and drafting the work for important intellectual content.

C.E.W. made a substantial contributed to the acquisition of data, analysis, interpretation, and revisions for important intellectual content.

E.H. made a substantial contributed to the acquisition of data, analysis, interpretation, and revisions for important intellectual content.

R.W. made a substantial contributed to the study design, acquisition of data, analysis interpretation and drafting the work for important intellectual content.

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Table I. Sample characteristics

HEALTH PROFESSIONALS (N=73)	**No. (range)**
Mean age in years at interview (range)	39.81 (23-64)
Average years practicing (range)†	8.55 (0.50-23)
Average hours each week in direct contact with patients at-high risk of breast/ovarian cancer (range)	7.53 (0.05-27.50)
Cultural Background‡	**N (%)**
Caucasian	63 (86.30)
Other	9 (12.33)
Marital Status‡	
Single	19 (26.03)
Married	45 (61.64)
De facto / Partnered / Engaged	7 (9.59)
Other	1 (1.37)
Employment‡	
Genetic counsellor	59 (80.82)
Other (e.g., geneticists, nurses, oncologists, psychologists/psychiatrists)	13 (17.81)

†Missing demographic data (n=3)
‡Missing demographic data (n=1)

Table II. Spontaneously reported techniques used by genetic health professionals to facilitate disclosure about a hereditary condition within the family

TECHNIQUES OR RESOURCES USED BY GHPS	EXAMPLES	ILLUSTRATIVE QUOTE
Educate and correct misconceptions	Emphasis the significance of genetic results for the families' healthcare, emphasis their right to know. Assess what information the patient has retained and capable of re-telling others.	*"[Patient's come thinking there is] a pre-determined or that a concrete plan has been set in place of what [testing's] going to mean for them"* (FG23)
Assess motivation or reasons for disclosure	Patients may want their relatives to test to relive themselves of guilt; potential for relatives not to make autonomous decisions	*"I think about what motivates someone to want to tell their family early and what motivates them to withhold information, [it] is really important"* (FG3)
Hypothetical scenarios & benefit/cost of non-disclosure	Used when patients are reticent or actively non-disclosing to their at-risk relatives	*"…try [to] think about the consequences of not disclosing to [your] daughter"* (FG24)
Frame	Perceive genetic testing as helpful	*"I often say to people, 'You don't*

positively	and beneficial in leading to risk-reduction of cancer through screening and surgery	*have any choice about what genes you pass on, but you do have a choice to share this information…this is something you can do…[that] you are in control of"* (FG4)
Normalise	Reassure that others commonly experience the same emotional responses and barriers. Provide examples of other families' experiences and the strategies used to overcome similar barriers.	*"Everybody brings in a different attitude to this from sort of incredibly pragmatic to incredibly emotional and that must provide the way you deal with that information in your family. There's no right or wrong it's just how you're wired to move forward*" (FG4)
Identify another relative to disclose information to family members	Used particularly when the proband/patient is unable to disclose to relatives.	*"If they can delegate the task…get your brother who is in touch with all these people or cousin…give the job of disseminating information to somebody else and then that way the patient can concentrate on their own health"* (FG1)

Discuss "when", "who", "how" they will disclose	Identify ideal timing (e.g., casually, avoid anniversaries/major events), who is at risk and what modality to use to communicate to relatives (e.g., face-to-face, letter, online)	*"Determined whether we know which side of the family the mutation is coming from"* (FG12)
Role play	Re-enact the discussion parents would have with their relative, provide the vocabulary, develop a plan, and draw upon how parents' have disclosed difficult information in the past.	*"I often say, 'You best have…a phrase or something you're going to say that you feel is age-appropriate for your child'…you want to have a scenario whereby you can communicate something that feels safe for the child that is age-appropriate in terms of the language and you probably don't want to minimise it or just brush it under the carpet. You want to try and be honest."* (FG4)
Family letters	Helpful when proband/patient is unable to disclose to relatives or fearful of forgetting important information.	*"The letter help[s] them to share it with their family and that kind of externalises it from them"* (FG21)
Follow-up phone	Provided: 1) to clarify information provided by proband/patient, 2)	*"I say, 'Make a plan…we'll discuss it over dinner or when you*

calls/letters/appointments	when the relative is reaching an age when medical management is recommended (e.g., screening at 30), 3) when proband/patient needs time to process information (cognitively, emotionally)	*are on a family outing and then I check in a few weeks later, 'Did you do that', 'How did it go', 'Is there any way you can think of doing it another way if you didn't get opportunity to discuss it?'"* (FG2)
Booklets, pamphlets, websites, factsheets	Provides information, techniques, vocabulary, and examples of other families' experiences.	Younger generation: *"Anything online probably would be a great resource for that age group*" (FG20) Older generation: *"...I think if people are handing things to the family, they still want it as more a physical thing like I think from my experience I guess the people maybe that are sharing that information are little bit older themselves..."* (FG7)
Social media	Helpful if relatives are estranged, live overseas or have minimal contact.	*"They will say 'Oh, actually my relatives are overseas' it's almost as though they're not part of the*

family anymore, 'They're so distant from me'" (FG4)

Table 1 Consolidated criteria for reporting qualitative studies (COREQ): 32-item checklist	**Manuscript meets criteria (Yes / No)**
No Item Guide questions/description	
Domain 1: Research team and reflexivity	
Personal Characteristics	
1. Interviewer/facilitator Which author/s conducted the interview or focus group?	Yes
2. Credentials What were the researcher's credentials? E.g. PhD, MD	Yes
3. Occupation What was their occupation at the time of the study?	Yes
4. Gender Was the researcher male or female?	Yes
5. Experience and training What experience or training did the researcher have?	Yes
Relationship with participants	
6. Relationship established Was a relationship established prior to study commencement?	Yes
7. Participant knowledge of the interviewer What did the participants know about the researcher? e.g. personal goals, reasons for doing the research	Yes
8. Interviewer characteristics What characteristics were reported about the interviewer/facilitator? e.g. Bias, assumptions, reasons and interests in the research topic	Yes
Domain 2: study design	
Theoretical framework	
9. Methodological orientation and Theory What methodological orientation was stated to underpin the study? e.g. grounded theory, discourse analysis, ethnography, phenomenology, content analysis	Yes
Participant selection	
10. Sampling How were participants selected? e.g. purposive, convenience, consecutive, snowball	Yes
11. Method of approach How were participants approached? e.g. face-to-face, telephone, mail, email	Yes
12. Sample size How many participants were in the study?	Yes
13. Non-participation How many people refused to participate or dropped out? Reasons?	Yes
Setting	
14. Setting of data collection Where was the data collected? e.g. home, clinic, workplace	Yes
15. Presence of non-participants Was anyone else present besides the participants and researchers?	Yes

16. Description of sample What are the important characteristics of the sample? e.g. demographic data, date	Yes
Data collection	
17. Interview guide Were questions, prompts, guides provided by the authors? Was it pilot tested?	Yes
18. Repeat interviews Were repeat interviews carried out? If yes, how many?	NA
19. Audio/visual recording Did the research use audio or visual recording to collect the data?	Yes
20. Field notes Were field notes made during and/or after the interview or focus group?	Yes
21. Duration What was the duration of the interviews or focus group?	Yes
22. Data saturation Was data saturation discussed?	Yes
23. Transcripts returned Were transcripts returned to participants for comment and/or correction?	NA
Domain 3: analysis and findingsz	
Data analysis	
24. Number of data coders How many data coders coded the data?	Yes; 3
25. Description of the coding tree Did authors provide a description of the coding tree?	Yes
26. Derivation of themes Were themes identified in advance or derived from the data?	Yes
27. Software What software, if applicable, was used to manage the data?	Yes
28. Participant checking Did participants provide feedback on the findings?	NA
Reporting	
29. Quotations presented Were participant quotations presented to illustrate the themes / findings? Was each quotation identified? e.g. participant number	Yes
30. Data and findings consistent Was there consistency between the data presented and the findings?	Yes
31. Clarity of major themes Were major themes clearly presented in the findings?	Yes
32. Clarity of minor themes Is there a description of diverse cases or discussion of minor themes?	Yes

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When to break the news and whose responsibility is it? A cross-sectional qualitative study of health professionals' views regarding disclosure of BRCA genetic cancer risk

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When to break the news and whose responsibility is it? A cross-sectional qualitative study of health professionals' views regarding disclosure of BRCA genetic cancer risk

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1. ABSTRACT

Objectives: Disclosure of a hereditary condition in the family poses notable challenges for patients who often seek the assistance of genetic health professionals (GHPs). This study aimed to investigate GHPs' opinions about the ideal time for disclosure to offspring and their responsibility to at-risk relatives.

Design: Cross-sectional qualitative study.

Setting: Genetic familial cancer clinics related to mostly secondary and tertiary care hospitals and centres in urban, regional and rural areas across all states of Australia.

Participants: GHPs (N=73) including clinical geneticists, genetic counsellors, medical specialists, nurses, surgeons, and psychiatrist/psychologists who had worked with *BRCA1* and *BRCA2* families for an average of 9 years.

Results: Focus groups and interviews were transcribed and analysed thematically. GHPs perceived that life-stage, maturity, parents' knowledge, and capacity to disseminate information influenced parent-offspring disclosure. In general, GHPs recommended early informal conversations with offspring about a family illness. GHPs considered that facilitation of disclosure to relatives using counselling strategies was their responsibility, yet there were limitations to their role (e.g., legal and resource constraints).Variability exists in the extent to which genetic clinics overcome challenges to disclosure.

Conclusions: GHPs' views on the ideal time for disclosure of genetic risk is generally dependent on the patient's age and relative's ability to disclose information. A responsibility towards the patient and their at-risk relative was widely accepted as a role of a GHP but views vary depending on legislative and specialty differences. Greater uniformity is needed in genetic procedural guidelines and the role of each discipline (e.g., geneticists, oncologists, genetic

counsellors, nurses, and psychiatrists/psychologists) in genetic clinics to manage disclosure challenges.

Strengths and limitations of this study

- This study provides one of the largest cohort of Australian genetic health professionals, with a detailed, in-depth approach to responsibility and confidentiality concerns.
- The findings extend on previous literature by focusing on two major genetic disclosure issues: ideal age of disclosure and the extent to which health professionals are responsible to warn at-risk relatives of their genetic cancer risk.
- The study was limited to focus primarily on the disclosure of *BRCA1* or *BRCA2* genetic test results.
- A qualitative approach to understanding the challenges of disclosure
- Highlights the variability in clinical practices across different legislative contexts and a need for clearer policies and role definitions.

Keywords: duty to warn; genetic privacy; genetic counselling; genetic testing; Genes, *BRCA1*; Genes, *BRCA2*; disclosure; ethical issues; genetic services

2. BACKGROUND

Identifying a *BRCA1* or *BRCA2* (hereafter *BRCA1/2*) pathogenic variant in the family and informing relatives can be a challenge, since most people do not want to be a 'bearer of bad news'. Yet such information can have far-reaching implications for a relative's decision regarding risk management, lifestyle, and family planning.

Genetic health professionals (GHPs) often work with families trying to navigate parent-offspring communication about genetic risk, commonly around when, how and what information to give, particularly the ideal age for disclosure [1]. Factors such as age, gender and the type of genetic condition can influence offspring's understanding of genetic information [2, 3]. GHPs are also concerned about non-disclosure to offspring, which does occur, albeit less commonly than to extended relatives [4, 5]. Reasons for non-disclosure include: parental guilt, fear of burdening others and a relatives' inability to cope [5]. Approximately one third of patients want GHP involvement during family communication [4, 6], especially when families are emotionally and geographically distant [6]. Offspring have also reported a preference for GHPs to disclose a hereditary condition in the family as opposed to parents [7]. Yet the extent to which GHPs are responsible for ensuring appropriate disclosure is a matter of debate. According to Parker and Lucassen [8] considering who owns genetic information is a matter of two viewpoints, namely, as belonging to the individual (personal account model) or belonging to the family (joint-account model). From a personal account standpoint, genetic information is *confidential* unless there is strong reason for disclosure, whereas from a joint-account viewpoint, genetic information is *familial* information, assuming justice to all members, and is communicable unless there is strong reason for non-disclosure. In Australia, the latter is not a widespread viewpoint.

Despite the prevalence of studies exploring family communication of genetic information [9], very few studies explore GHPs' opinions on their responsibility to at-risk relatives. A recent systematic review [10] found that across eight countries and varying heritable illnesses, GHPs generally felt some sense of responsibility to inform their patients' relatives about their genetic risk. Yet a range of moral, legal, and practice-related arrangements reportedly made it challenging to act on their perceived responsibility. Of the nine studies in the review, none specifically explored GHPs' responsibility towards patients' relatives. A recent study therefore, aimed to address this gap by conducting focus groups with UK GHPs [11]. GHPs in the UK were concerned about the difficulty in distinguishing between genetic and personal information thereafter potentially breaching confidentiality through disclosure and more broadly, reported a need for national consensus on following the UK guidelines from the Joint Committee on Medical Genetics. According to these guidelines, GHPs explore family relationships, encourage family communication, and assume that responsibility of disclosure lies with the patient.

Under the legislative guidelines of some countries, when patients do not provide consent for the disclosure of genetic information, GHPs can make contact with at-risk relatives.

Both Australian and UK guidelines encourage GHPs to take reasonable steps to obtain consent and consider the potential consequences of disclosure when consent is not provided [12, 13]. According to current Australian guidelines from the National Health and Medical Research Council, a GHP can disclose genetic information to an at-risk relative without the patient's consent in specific circumstances. This exemption applies for "incurable" conditions which are "preventable" or include "treatable manifestations" (e.g., depression), in which "specific management" or "treatment" can "lessen or prevent" the threat of disease or distress [12; p.42]**.** Nevertheless, there is a lack of uniformity across Australia in how these guidelines are followed

and upheld in clinical practice [14]. South Australian genetic services [15], for example, provide family letters to at-risk relatives to inform them of an increased risk, with the patient's consent but without the recipient's consent, whereas the rest of Australia do not make provisions for direct contact with relatives. The extent to which Australian GHPs within public hospitals consider it their role to assist families with disclosure is currently unclear. The purpose of the current study was to understand the role of GHPs[1] in assisting families with disclosure of genetic cancer risk. Specifically, two research questions guided the study: (1) When is the best time to tell offspring about their genetic risk? and, (2) Who is responsible to inform relatives of their genetic risk?

3. METHODS

Patient and Public Involvement statement: No patient involved.

Study participants and recruitment: Eligible GHPs (e.g. clinical geneticist, genetic counsellors, medical specialists, nurses, surgeons, and psychiatrist/psychologists) who had worked with *BRCA1/2* families from familial cancer centres within all Australian states. A PhD candidate with several years' experience in qualitative research (A.L.Y.) presented the study to potential GHP participants during family cancer clinic meetings and/or emailed study details. Interested GHPs were then re-contacted to arrange a suitable time for participation in a focus group or interview. Recruitment continued until theoretical saturation was achieved [16]. Informed consent was obtained from all participants and the study was approved by the University of Sydney Human Research Ethics Committee (HREC 2017/011) which covered the participating hospitals.

Procedures: GHPs completed a questionnaire primarily collecting demographic data, and then took part in a focus group or interview. Focus groups (2-8 individuals) were held in-person or via

video-teleconferencing in 2017, during familial cancer clinic meetings or at a time convenient for participants. Semi-structured telephone or face-to-face interviews were completed with participants unable to attend a focus group. Three qualitative researchers (A.L.Y., P.N.B., C.E.W) conducted interviews and focus groups that ranged in duration from 15-77 minutes, depending on the time-availability of participants.

Data analysis: Focus groups and interviews were transcribed verbatim and underwent data-driven analysis by three authors (A.L.Y., P.N.B., R.W.) guided by thematic analysis [17] using NVivo 11 computer software to map themes. Consideration was given to whether individuals participating in one method (i.e., focus groups) differed in relation to the experiences discussed in the other method (i.e., interviews). Emphasis was placed on the themes mentioned by the majority of participants and data that raised novel lines of inquiry, reflecting unique sub-themes (e.g., rurality, specialists). Three authors (A.L.Y., P.N.B., R.W.) analysed the first six transcripts by re-reading each transcript, generating codes, and developing overall themes which were then organised into a thematic 'map'. Differences in coding were resolved by consensual discussion. Subsequent transcripts were analysed according to the 'map' resulting in a final set of themes. Focus group or individual interview identification (e.g., FG4 or II4) are provided below.

4. RESULTS

Sample characteristics: Of 91 eligible GHPs invited, 73 consented and participated in the study. Demographic characteristics are provided in Table I.

4.1. When is the best time to tell offspring about their genetic risk?

4.1.1. Depending on the offspring: GHPs had different arguments for and against disclosing genetic status to offspring earlier than the age of 18, at the age of 18 and at the age of 25 onwards. Most GHPs reported that earlier was better to allow time for the offspring to adjust to,

process and research information about their genetic risk before making decisions about medically mitigating their risk. Informing offspring as the conversation arises, in an age-appropriate manner, was commonly encouraged: "*in an ideal situation it should be a progressive discussion over time*" (FG1). GHPs argued that if information about genetic risk is withheld, offspring might hear about it inadvertently from relatives and through GHPs during unrelated appointments, placing strain on the parent-offspring relationship. A few GHPs said that informing offspring in their mid-teenage years (15-17 years old) was ideal: "*probably mid-teens, and the reason why [is] to be aware [that] they can be breast aware, not breast alarmed and breast paranoid"* (II15). Planning to inform offspring at the age of 18, or "*saving it up as an 18th birthday present*" (FG9), was considered unhelpful and described as *"dropping the bombshell*" (FG3).

In contrast, some GHPs felt that disclosure should be related to **when it could inform testing/screening behaviour or decision-making**, and therefore advocated disclosure at an older age (>20 years)**.** The recommended breast screening age for *BRCA1/2* carriers is 30 within Australia [18]. In relation to the patient's mental health, some GHPs said that disclosure at a young age can lead to prolonged worry, since the time between disclosure and screening is longer compared to their older counterparts. However, others disagreed with this stance stating that older patients can be more anxious if their parents informed them later due to the immediacy of action needed to mitigate risk. Some GHPs noted that parents may not be alive when their offspring reached the recommended age for screening or testing, and therefore disclosure, and possibly testing, should occur earlier. Subsequent, shifts in opinions were discussed, "*it used to be you don't do anything until you're going to use the information. And my original teaching was*

that you don't do genetic testing till a month before they're due to start screening…we now know that that isn't necessarily the best way of offering genetic testing" (FG1).

Other reasons some GHPs advocated disclosure **at an older age** were that young adults are considered generally more mature and receptive towards genetic information than younger offspring. Furthermore, the parent-offspring relationship is likely to change to an adult-adult pattern of relating as offspring age, which can be considered *"on a more even level"* (FG4), allowing the young adult to be autonomous in their responses to genetic information and testing decisions. Disclosure at an older age was considered advantageous to avoid having offspring incorporate the pathogenic variant into their identity. A final justification for withholding information till adulthood was the potential for inappropriate medical management of young adults by GHPs in response to anxious younger adults insistent to have testing: "*they can be inappropriately managed if they're aware of this information from a young age…privately…[and] publicly… a breast surgeon will often screen younger women*" (FG7).

Some GHPs recommended disclosure **at key points related to genetic risk** when offspring was in any case probably aware of health problems and emotional distress in the family, such as when a parent was diagnosed and being treated for cancer, or having surgery.

Other GHPs said they **did not recommend a time or age for disclosure**, rather they spoke about taking a case-by-case approach, taking into account the unique characteristics of each individual, life experiences and family dynamics.

4.1.2. Depending on the parent: Some GHPs emphasised that disclosure should depend on the parent's decision about when and how they want to tell their offspring: "*parents know their child best of all and they would be in the best position to judge*" (FG22). Parents can use their intuition to decide on the timing of disclosure and skilfully navigating around stressful events (e.g., cancer

diagnosis or anniversaries). Yet other GHPs felt that some parents' negative experience with genetic services could hinder timely and effective disclosure. For example, "*it's often a red flag when you have someone who's not [coped with the testing process]…if they've got a lot of emotional turmoil going on they kind of can project that and expect that their children will react the same way and perhaps think that their kids can't cope"* (FG4). GHPs also reported that parents may not understand the seriousness of sharing hereditary cancer information with families, may have forgotten about their results from a research study, and can potentially still be trying to process the information for themselves.

4.2. Who is responsible to inform relatives about their genetic risk?

4.2.1. GHPs are responsible to facilitate and support family communication: Many GHPs agreed that they were responsible for facilitating family communication by using a range of strategies to support probands (i.e., during diagnostic testing) and relatives (i.e., during predictive testing) to talk to their offspring and relatives (See Table II for full list of strategies). Factsheets were referred to most often as a resource to provide probands to assist with disclosure but was considered too generic or not user-friendly by some. GHPs also admitted to treating families differently depending on the type of test, "*I'm probably not as active in my making sure the information gets out there with the predictive[2] as I am [with probands who are the first individual with a pathogenic variant to be identified in the family]*" (FG7).

GHPs reported that assessment of family communication processes should start early in the consultation; for geneticists/genetic counsellors this involved pre-test counselling, for nurses/psychologists this was during the first consultation and for oncologists this was at the time of talking about genetic testing to cancer patients. Assessment included exploring family dynamics (e.g., estrangement, lack of communication), emotional responses that could impede

communication (e.g., guilt, fear) and assessing the proband's ability to disclose information to relatives (e.g., clarify their understanding, coping skills). Building rapport with the patient is an important "*initial foundation*" (FG20) to help patients with their communication with families. GHPs advocated for gently preparing probands for the possibility that they could have a positive test result, and if so, to consider to whom, what and when they would disclose their results. When parents were finding it difficult to communicate to their offspring, GHPs offered to have the offspring join the parent's consultations, or provide subsequent over-the-phone consultations with the offspring and/or a separate consultation for the offspring to obtain more information. Family group consultations were also recommended to facilitate communication and address concerns with the relevant relatives present. Such consultations allow all members of the family to be informed simultaneously. Having another family member can lead to greater clarification of information: "*someone [can] obviously pick something up but they then explained it in a way that helps*" (FG21). However, some GHPs were also concerned about the practicality of implementing family group consultations in the current public health model. In some cases when disclosure in families were not occurring, some GHPs were willing to '"*take it upon themselves" to see that the adult children involved [were] informed of their risk, "So you're protecting the rights of the child as well*"' (FG12).

4.2.2. Different clinics, different responsibility: The work culture, resources and expectations within particular genetic services influenced GHPs' views about disclosure to relatives: "*the scope of your role changes with whatever clinical service you are working with*" (FG2). Some clinical teams placed greater emphasis on disclosure to at-risk relatives. For example, working within high-risk clinics provided some opportunities for nurses to explore disclosure, whereas some genetic counsellors reported, "*working in a busy clinical service in the public system really*

limits us in terms of our capacity of what we can do" (II14). Emphasis was placed on young at-risk relatives' personal responsibility, with one clinical geneticist saying, *"It's going to become too big for familial cancer centres to be able to hold onto these families and do the follow-up. I think it's going to have to shift out to personal responsibility*" (FG10).

4.2.3. Ultimately it is the families' responsibility: Families were considered ultimately responsible for what they want to do with their own medical information. Confidentiality and autonomy were upheld by GHPs and if a patient choose to be private, this was respected. Some felt it was not their role to ensure disclosure beyond providing a family letter.

Conversely, others were of the mind that they would like to assist families with communication but were limited by time constraints and procedural barriers. Some GHPs believed GHPs should not feel guilty if disclosure did not occur in a family particularly since families do not always tell GHPs the truth and are unwilling to discuss family dynamics. Some familial issues are beyond a medical GHP's capacity (or consultation time) to discuss and requires psychological assistance. For example, a genetic counsellor said, *"Sometimes I think whatever's going on in their families is beyond what we as genetic counsellors can actually help with, which is unfortunate, but…considering the workload…you can only pour so much of your energy into one family"* (FG20). Another common limitation GHPs discussed was having no control over what happens after a consultation, "*you've got no control over what's passed on and what isn't or how it's passed or whether facts and figures [are] mixed up*" (FG10). Moreover, GHPs were also aware that advocating for disclosure was not beneficial in all cases, "*it's important to be aware of the fact that there could be positives and negatives [in] telling, but also positives and negatives in not telling"* (II18). Situations in which information is withheld or difficult to navigate include cases when an at-risk relative has a mental health concern and/or cognitive disability.

5. DISCUSSION

The age at which disclosure should ideally begin is not a concern of *BRCA1/2* families alone but is common amongst families with a hereditary condition [2]. GHPs in this study recommended optimal timeframes for disclosure of genetic risk to offspring, with the majority favouring early disclosure tailored to individual circumstances. Hereditary cancer can be introduced into the family story with a simple explanation about genetics, cancer and the benefits of testing [19]. An example of such an explanation is that used for families with Cystic Fibrosis, terminology that normalises their condition such as, "everyone possesses disease causing genes" [20; p. 206]. This method of dissemination is modelling to offspring that coping and adjustment to such information is possible. Having more time to process, discuss, and ask questions during casual conversations is less anxiety-provoking than being informed unexpectedly at an age when immediate medical action is required [21].

According to Klitzman and colleagues [7], the reasoning behind GHPs' perspective about the ideal age for genetic testing and subsequent disclosure can fall under two categories: 1) the *life stage* or maturity of offspring and 2) the *medical time course* and benefit of the information at a given time. Age-appropriateness was a key feature of early disclosure. According to Piaget's theory of cognitive development, children at approximately 11 years old reach the stage of 'formal operational thought', at which hypothesis testing and abstract reasoning develop [22]. In theory, children at this stage can make inferences that if their parent is ill, then they too could become ill with the same illness [23, 24]. Thus, parents are recommended to consider their offspring's cognitive and emotional capacity before informing them about their risk [19], which may have different developmental trajectories depending on the temperament of the offspring [25]. Parental consideration of disclosure of genetic status with young adults involves

consideration of poignant life-stage changes or communicating at certain junctures (e.g., impending marriage or pregnancy) [9]. Parental capacity to inform offspring [2, 20] and their own experience or level of satisfaction with genetic testing may hinder communication [26], warranting the facilitation of communication by GHPs [27].

GHP's facilitation of disclosure is generally agreed to be incorporated in their clinical practices (i.e., Table II), expounding upon previously reported strategies [11]. Contrastingly, the opinions of some GHPs suggest that facilitation of disclosure is a peripheral requirement of their practice, other than providing a family letter passed from patient-to-relative. Patient autonomy, confidentiality and/or the law can also contribute to the reluctance to facilitate disclosure [10]. Follow-up calls/appointments to address disclosure is considered worthwhile to revisit the topic of disclosure with families, particularly when legislative changes occur [28], yet resource and time constraints can make this impracticable [29].

In South Australia, the genetic services send letters directly to at-risk relatives, with the patient's consent. Other research has shown benefits from direct contact with at-risk relatives [30]. Studies involving a range of illnesses (e.g., *BRCA1/2*, Lynch, Cowden Syndrome), have shown that when GHPs mediated contact, uptake of testing was greater amongst at-risk relatives compared to when contact was patient-mediated [15, 31]. A Western Australian study adopted a cascade screening process derived from an Australasian model of care for familial hypercholesterolaemia in accordance to local and national guidelines. Nurse-led initiation of contact with at-risk relatives, despite non-consent from probands, allowed for the identification of carriers in first-, second- and third-degree relatives [32]. Recently GHPs working in French genetic clinics are legally permitted to offer a written document informing at-risk relatives of their risk, yet guidance about to whom this requirement extends to and how GHPs responsibility will be

defined remains elusive [33]. Yet GHPs are still apprehensive about changing their practices [34], highlighting that a shift towards a 'joint-account model' is not only a matter of legislative changes but also a matter of shifting viewpoints.

In this study, we found that GHPs' opinions regarding their responsibility towards at-risk relatives differed depending on four factors. First, a GHP's role and opinion were informed by the attitudes and expertise of the genetic clinic in which they worked, which varied between local health districts and states. Second, GHPs in each specialty were governed by their own legislation and ethical guidelines, including the overall framework and ethos in which they practice, which emphasised family communication to a lesser or greater extent depending on the profession (e.g., genetic counsellor vs. medical oncologist). Third, GHPs generally reported a greater need to facilitate communication for probands who are the first in the family to be identified as a carrier since the burden of sharing information appears greater than those who have cascade testing, and therefore can also be dependent on whether the patient has cancer or not. Fourth, state-wide differences in health provision and legislation meant that clinics in South Australia can send genetic letters to relatives effectively [15], but GHPs in New South Wales are still cautious of potential litigation issues despite amendment of privacy principles in attempt to uniform Australian genetic practices [14].

GHPs working in genetic clinics, many of whom are genetic counsellors, are governed by their respective professional guidelines and respective health district legislation. 'Non-directiveness' is a term used to describe GHP practices that are patient-centred and uphold the autonomous decisions of the patient [35]. Yet within the context of cancer genetics in which evidence-based surgical treatments exist that effectively mitigate risk, adherence to such principles is questionable [36]. The Task Force of the National Society of Genetic Counsellors has

consequently excluded the term 'non-directiveness' in their definition of genetic counselling, emphasising instead on educating patients about testing implications for themselves and relatives [37]. A shared decision-making approach is currently favoured [38].

With the rise of genomic medicine and the subsequent need to educate the general public of the potential benefits and limitations of such knowledge, the question of who is responsible to inform the public is a wider healthcare concern. Contrary to the opinions of a small minority of GHPs in the current study who predict a shift to personal responsibility when managing at-risk relatives, the European Breast Cancer Council [39] argues that the healthcare system will need to rise to the challenge and support future families in obtaining high-quality and timely information. It is already evident that without the input of cancer genetic clinics, at-risk relatives are not considering the potential limitations of a proposed test before choosing direct-to-consumer genetic testing [40]. The lack of guidance and advice from medical professionals about their genetic risk [40], further reinforces the need for genetic cancer clinics to clarify their responsibility for at-risk relatives.

Practice Implications: A multidisciplinary approach to genomic medicine has been proposed to be effective in tackling the challenge of disclosure [28, 41]. GHPs are currently supporting young at-risk relatives within high-risk clinics in Australia, but can also potentially allow for ongoing support of families struggling with disclosure difficulties. It is possible that during the consent conversation with an index patient GHPs can provide the joint-account viewpoint towards familial information using the analogy of family members owning a joint bank account and having equal rights to the funds (information) [8].

Funding regulators are to emphasis fiscal and institutional backing of genetic clinics in order to sustain a multidisciplinary team approach and to manage the future role of GHPs in the

preventive health of their patient's relatives. GHPs need to be having discussions, amongst themselves, but ideally nationally with policymakers, legal services and government, to advocate for more clarity about who owns genetic information – the patient or the family (personal vs joint-account model) and greater clarity is needed on the definition of 'at-risk relatives', and the extent to which GHPs are responsible to inform them. The current approaches recommended to address disclosure of cancer risk in Australia are ad hoc; more guidance and standardisation of practices is needed by modifying guidelines that are better suited to local regulatory needs.

Limitations: This study focused primarily on GHPs' view of *BRCA1/2* families' disclosure of genetic risk. Nevertheless, the topics covered were broad enough to have applicability and transferability to other adult-onset genetic health conditions (e.g., Familial Hypercholesterolaemia) in which parents are also faced with the dilemma of disclosure, and in some cases they have similar consequences and risk-management options available (e.g., Lynch syndrome). Genetic counsellors (80%) provided the most input into discussions and therefore their practices were emphasised the most in the results. Further research is required, with larger samples of geneticists, surgeons, nurses and psychologists in order to elucidate whether their opinions about their role in disclosure differs.

Conclusion: This Australian study reports on the ideal age of parent-offspring disclosure of genetic risk and GHPs' responsibility towards at-risk relatives. Our findings highlight the need for clearer policies regarding GHP's responsibility to relatives and to the community in terms of preventive health, including the need for more staff and fiscal support to sustain disclosure initiatives (e.g., direct-contact letters).

Footnotes:

[1]*GHPs,* in the context of this study, refers to clinical geneticists and genetic counsellors, and more broadly, health professionals who have worked closely with patients with a *BRCA1* or *BRCA2* genetic risk.

[2]*Predictive testing* is the testing of a relative after a pathogenic variant has already been identified in the proband.

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CONFLICT OF INTEREST

A.L.Y., P.N.B., K.M.T., C.E.W., E.H., and R.W declare that they have no conflict of interest.

ETHICS APPROVAL

The project was approved by the University of Sydney Human Research Ethics Committee (HREC 2017/011) which covered the participating hospitals.

DATA SHARING STATEMENT: Data are available upon reasonable request to the authors.

INFORMED CONSENT

All procedures followed were in accordance the University of Sydney Human Research Ethics Committee) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Signed or electronic consent was obtained from all participants. Informed consent was obtained from all patients for being included in the study.

AUTHOR'S CONTRUBUTION STATEMENT

A.L.Y made a substantial contribution to the study design, acquisition of data, analysis, interpretation, revisions for important intellectual content.

P.N.B. made a substantial contribution to the conception, acquisition of data, analysis interpretation and drafting the work for important intellectual content.

K.M.T. made a substantial contributed to the study design, acquisition of data, analysis interpretation and drafting the work for important intellectual content.

C.E.W. made a substantial contributed to the acquisition of data, analysis, interpretation, and revisions for important intellectual content.

E.H. made a substantial contributed to the acquisition of data, analysis, interpretation, and revisions for important intellectual content.

R.W. made a substantial contributed to the study design, acquisition of data, analysis interpretation and drafting the work for important intellectual content.

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Table I. Sample characteristics

HEALTH PROFESSIONALS (N=73)	**No. (range)**
Mean age in years at interview (range)	39.81 (23-64)
Average years practicing (range)†	8.55 (0.50-23)
Average hours each week in direct contact with patients at-high risk of breast/ovarian cancer (range)	7.53 (0.05-27.50)
Cultural Background‡	**N (%)**
Caucasian	63 (86.30)
Other	9 (12.33)
Marital Status‡	
Single	19 (26.03)
Married	45 (61.64)
De facto / Partnered / Engaged	7 (9.59)
Other	1 (1.37)
Employment‡	
Genetic counsellor	59 (80.82)
Other (e.g., geneticists, nurses, oncologists, psychologists/psychiatrists)	13 (17.81)

†Missing demographic data (n=3)
‡Missing demographic data (n=1)

Table II. Spontaneously reported techniques used by genetic health professionals to facilitate disclosure about a hereditary condition within the family

TECHNIQUES OR RESOURCES USED BY GHPS	EXAMPLES	ILLUSTRATIVE QUOTE
Equip parents with terminology to discuss genetic risk with offspring in an age-appropriate manner	Be mindful of an individual's cognitive capacity, emotional maturity and external factors before informing offspring. Use concepts that are easily understood by a particular age group. • **Children:** Use basic concepts; brief explanations; be open about your medical appointments • **Adolescence:** casual conversations; allow for collaborative decision making • **Young Adults:** provide details of genetic clinics;	*"…every family has certain health issues…this is just one thing that our particular family [has to do, e.g. check-ups, etc.]…"* (FG9) *"Just briefly mention 'In our family there is an increased cancer risk, [e.g.] Aunty Stella has decided that she's going to look after herself this way [prophylactic surgery]. But you've seen me I go to my screening every year and that's what I do'. If [parents] go for annual screening…don't just hide that appointment or go when the children are at school so they*

	allow for an autonomous decision making	*don't know, just be open about the fact 'Oh I'm off for my annual screening today'"* (FG9)
Educate and correct misconceptions	Emphasis the significance of genetic results for the families' healthcare, emphasis their right to know. Assess what information the patient has retained and capable of re-telling others.	*"[Patient's come thinking there is] a pre-determined or that a concrete plan has been set in place of what [testing's] going to mean for them"* (FG23)
	When patients discuss reasons for not wanting to inform relatives gently question for their reasons; such questioning might reveal myths that can be dispelled.	*"[Some] people…[with] no breast cancer in the family…still can't talk about it because, 'We might all get breast cancer', even though no one has had breast cancer before… I don't think they can talk about [a] gene mutation or the cancer because I think for many people there are very intimately associated"* (FG25)
Provide reassurance and encourage patients to ask	At-risk relatives can make contact or attend an appointment at the genetic clinic for information-	*"I say, 'You don't need to worry about giving them all the ins and outs of what it means and what it means for them, they can speak to*

their at-risk relatives to make contact with a genetic service	gathering purposes and not only for genetic testing.	*the local genetics service…so I reassure them that they're not expected to be the expert for the family just to be the source of information about it'"* (FG9)
Assess motivation or reasons for disclosure	Patients may want their relatives to test to relieve themselves of guilt; potential for relatives not to make autonomous decisions	*"I think about what motivates someone to want to tell their family early and what motivates them to withhold information, [it] is really important"* (FG3)
Hypothetical scenarios & benefit/cost of non-disclosure	Used when patients are reticent or actively non-disclosing to their at-risk relatives	*"…try [to] think about the consequences of not disclosing to [your] daughter"* (FG24)
Frame positively	Perceive genetic testing as helpful and beneficial in leading to risk-reduction of cancer through screening and surgery	*"I often say to people, 'You don't have any choice about what genes you pass on, but you do have a choice to share this information…this is something you can do…[that] you are in control of"* (FG4)

Normalise	Reassure that others commonly experience the same emotional responses and barriers. Provide examples of other families' experiences and the strategies used to overcome similar barriers.	*"Everybody brings in a different attitude to this from sort of incredibly pragmatic to incredibly emotional and that must provide the way you deal with that information in your family. There's no right or wrong it's just how you're wired to move forward*" (FG4)
Alerting at-risk relatives by not disclosing the identity of an index patient	At-risk relatives can be alerted about their cancer risk by informing them that they might be at risk of a condition without disclosing personal medical information	*"[e.g.,] a woman does not want her family to know her [identity but wants them to know they are at risk, so making a] letter with de-identified information, which we can have them check [and written collaboratively with the patient was] was done routinely."* (FG19)
Identify another relative to disclose information to	Used particularly when the proband/patient is unable to disclose to relatives.	*"If they can delegate the task…get your brother who is in touch with all these people or cousin…give the job of disseminating information to somebody else and then that way the patient can*

family members		*concentrate on their own health"* (FG1)
Discuss "when", "who", "how" they will disclose	Identify ideal timing (e.g., casually, avoid anniversaries/major events), who is at risk and what modality to use to communicate to relatives (e.g., face-to-face, letter, online)	*"Determined whether we know which side of the family the mutation is coming from"* (FG12)
Role play	Re-enact the discussion parents would have with their relative, provide the vocabulary, develop a plan, and draw upon how parents' have disclosed difficult information in the past.	*"I often say, 'You best have…a phrase or something you're going to say that you feel is age-appropriate for your child'…you want to have a scenario whereby you can communicate something that feels safe for the child that is age-appropriate in terms of the language and you probably don't want to minimise it or just brush it under the carpet. You want to try and be honest."* (FG4)
Family letters	Helpful when proband/patient is unable to disclose to relatives or fearful of forgetting important information.	*"The letter help[s] them to share it with their family and that kind of externalises it from them"* (FG21)

Follow-up phone calls/letters/appointments	Provided: 1) to clarify information provided by proband/patient, 2) when the relative is reaching an age when medical management is recommended (e.g., screening at 30), 3) when proband/patient needs time to process information (cognitively, emotionally)	*"I say, 'Make a plan…we'll discuss it over dinner or when you are on a family outing and then I check in a few weeks later, 'Did you do that', 'How did it go', 'Is there any way you can think of doing it another way if you didn't get opportunity to discuss it?'"* (FG2)
Booklets, pamphlets, websites, factsheets	Provides information, techniques, vocabulary, and examples of other families' experiences.	Younger generation: *"Anything online probably would be a great resource for that age group*" (FG20) Older generation: *"…I think if people are handing things to the family, they still want it as more a physical thing like I think from my experience I guess the people maybe that are sharing that information are little bit older themselves…"* (FG7)

Social media	Helpful if relatives are estranged, live overseas or have minimal contact.	*"They will say 'Oh, actually my relatives are overseas' it's almost as though they're not part of the family anymore, 'They're so distant from me'"* (FG4)

Table 1 Consolidated criteria for reporting qualitative studies (COREQ): 32-item checklist	Manuscript meets criteria (Yes / No)	Page no.
No Item Guide questions/description		
Domain 1: Research team and reflexivity		
Personal Characteristics		
1. Interviewer/facilitator Which author/s conducted the interview or focus group?	Yes	7
2. Credentials What were the researcher's credentials? E.g. PhD, MD	Yes	7
3. Occupation What was their occupation at the time of the study?	Yes	7
4. Gender Was the researcher male or female?	Yes	1, 7
5. Experience and training What experience or training did the researcher have?	Yes	7
Relationship with participants		
6. Relationship established Was a relationship established prior to study commencement?	Yes	7
7. Participant knowledge of the interviewer What did the participants know about the researcher? e.g. personal goals, reasons for doing the research	Yes	7
8. Interviewer characteristics What characteristics were reported about the interviewer/facilitator? e.g. Bias, assumptions, reasons and interests in the research topic	Yes	7
Domain 2: study design		
Theoretical framework		
9. Methodological orientation and Theory What methodological orientation was stated to underpin the study? e.g. grounded theory, discourse analysis, ethnography, phenomenology, content analysis	Yes	8
Participant selection		
10. Sampling How were participants selected? e.g. purposive, convenience, consecutive, snowball	Yes	7
11. Method of approach How were participants approached? e.g. face-to-face, telephone, mail, email	Yes	7
12. Sample size How many participants were in the study?	Yes	8
13. Non-participation How many people refused to participate or dropped out? Reasons?	Yes	8
Setting		
14. Setting of data collection Where was the data collected? e.g. home, clinic, workplace	Yes	7
15. Presence of non-participants Was anyone else present besides the participants and researchers?	Yes	7

16. Description of sample What are the important characteristics of the sample? e.g. demographic data, date	Yes	8
Data collection		
17. Interview guide Were questions, prompts, guides provided by the authors? Was it pilot tested?	No included in this paper specifically but is elsewhere	
18. Repeat interviews Were repeat interviews carried out? If yes, how many?	NA	
19. Audio/visual recording Did the research use audio or visual recording to collect the data?	Yes	8
20. Field notes Were field notes made during and/or after the interview or focus group?	Yes	8
21. Duration What was the duration of the interviews or focus group?	Yes	7
22. Data saturation Was data saturation discussed?	Yes	7
23. Transcripts returned Were transcripts returned to participants for comment and/or correction?	NA	
Domain 3: analysis and findingsz		
Data analysis		
24. Number of data coders How many data coders coded the data?	Yes; 3	8
25. Description of the coding tree Did authors provide a description of the coding tree?	Yes	8
26. Derivation of themes Were themes identified in advance or derived from the data?	Yes	8
27. Software What software, if applicable, was used to manage the data?	Yes	8
28. Participant checking Did participants provide feedback on the findings?	NA	
Reporting		
29. Quotations presented Were participant quotations presented to illustrate the themes / findings? Was each quotation identified? e.g. participant number	Yes	Page 8-13
30. Data and findings consistent Was there consistency between the data presented and the findings?	Yes	Page 8-13
31. Clarity of major themes Were major themes clearly presented in the findings?	Yes	Page 8-17
32. Clarity of minor themes Is there a description of diverse cases or discussion of minor themes?	Yes	Page 8-17