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ARTICLE

Connecting patients, researchers and clinical genetics services: the experiences of participants in the Australian Ovarian Cancer Study (AOCS)

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Population-based genetic research may produce information that has clinical implications for participants and their family. Researchers notify participants or their next of kin (NoK) about the availability of genetic information via a notification letter; however, many subsequently do not contact a family cancer centre (FCC) to clarify their genetic status. Therefore, the purpose of this study was to examine research participants' experience of receiving a notification letter and the factors that influenced contact with an FCC. Twenty-five semi-structured interviews were conducted with research participants ($n = 10$) or their NoK ($n = 15$) who had received a notification letter following participation in the Australian Ovarian Cancer Study. There were a number of factors which impacted participants' access to genetic counselling at an FCC. Some participants had unmet information and support needs, which were addressed by their participation in this psychosocial interview study. Recruitment and participation in this study therefore inadvertently increased a number of participants' intention to contact an FCC. For others, participation in this study facilitated access to an FCC. Recommendations are proposed regarding future notification as well as implications for clinical practice. An approach that also provides opportunity to address research participants' support and informational needs before contacting a clinical genetics service as well as practical guidance for accessing genetic services would facilitate timely and smooth access for research participants who are interested in following up clinically relevant genetic test results.

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INTRODUCTION

Researchers have some responsibility to notify research participants of their personal clinically significant genetic research results.[1,2] The Australian National Health and Medical Research Council mandates that participants must be asked whether they wish to be notified of clinically significant research results and recommend clinical advice and counselling at minimum if information is disclosed.[3] Many empirical studies suggest that participants want to receive genetic results following research participation.[4,5] There is proven clinical utility (through early detection, prevention and mortality reduction) of acting on one's *BRCA1* or *BRCA2* gene mutation status,[6,7] making notification of these mutations to participants or their families useful. Ninety-three percent of women who participated in a UK *BRCA* prevalence study opted to receive feedback if a mutation was detected.[4] All who received a notification letter informing them of the finding accepted the invitation for genetic counselling.[4] Similarly, the majority of participants from the Colon Cancer Family Registry (Colon CFR) multinational study, in which genetic testing for mismatch repair or *MutYH* gene mutations was performed, accepted the opportunity to receive personal genetic results.[8]

In contrast, various Australian studies have found that less than half of research participants chose to attend a family cancer centre (FCC) to find out about their cancer risk after being notified that a genetic mutation had been identified.[9,10] Although overall uptake of receiving results was high in the Colon CFR study, Australia had the lowest uptake (56%) compared with other participating centres in the USA and Canada, where uptake was 72–86%.[8] Wakefield *et al*[11] found that many Australian research participants do not necessarily understand, value or act upon the information they received in the notification letter.[11] Alternatively, the lower uptake rated witnessed in Australia may be due to economic and geographical factors, such as misconceptions about the costs of genetic testing, concerns about insurance implications or the fact that clinics may be geographically distant and, therefore, difficult to attend.[8,12,13]

Concern that few participants pursued genetic counselling after receiving a notification letter regarding the identification of

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a *BRCA1/2* mutation led the Australian Kathleen Cunningham Foundation Consortium for Research into Familial Aspects of Breast Cancer (kConFab) to implement an intensive notification system to ensure participants were adequately informed about the available genetic information.[10] This involved a research nurse telephoning research participants who had not acknowledged receiving their notification letter.[10] Intensive follow-up did not increase uptake of genetic testing compared with usual notification; however, there was no exploration of why uptake remained low.[10] Although individuals are initially receptive and consent to receive notification of personal clinically significant research results at the time of study enrolment, there is little evidence of participants' experience of receiving notification and why many subsequently do not take up the offer of genetic counselling and testing in Australia. This qualitative exploration of research participants' and their next of kin's (NoK) experiences of receiving a notification letter from the Australian Ovarian Cancer Study (AOCS, refer to Box 1) was undertaken to address this gap. An earlier publication arising from this study has reported on the emotional response of receiving a notification letter.[14] This paper examines the factors that influenced individuals' contact with an FCC after receipt of a notification letter, specifically the factors influencing failure to access FCCs and a subsequent unintended intervention by experienced genetic counsellor researchers.

MATERIALS AND METHODS

Recruitment

Ethical approval was obtained from the Peter MacCallum Cancer Centre's Human Research Ethics Committee. Criteria for inclusion included previously receiving a notification letter about genetic research results, being >18 years of age and English speaking. Eligible participants were sent an invitation to participate in an interview with an opt-out card. A researcher telephoned the recipient to arrange an interview 2 weeks later.

Box 1 Description of the Australian Ovarian Cancer Study (AOCS)

The Australian Ovarian Cancer Study (AOCS) is a population-based study that recruited women diagnosed with ovarian cancer throughout Australia between 2002 and 2006, collecting a number of biospecimens as well as clinical and epidemiological data. Twelve hundred and seventy three women provided blood samples and mutation testing was undertaken to determine the prevalence of BRCA1 and BRCA2 mutations in Australian ovarian cancer patients (see Figure 1). At enrolment, participants consented to be notified if findings were identified with implications for themselves or their families. Women in whom a pathogenic mutation was identified, or their next of kin in the case where the woman was deceased, were notified in writing by the AOCS researchers about the finding of a mutation:

"...our research **has identified information relevant to your family**. This means that **a genetic change has been found in your family** which may account for the family's experience of cancer" [AOCS Notification letter, original emphasis].

In addition, the letter encouraged recipients to seek further information from an FCC.

"**We strongly recommend that you discuss this letter with a genetic counselor or doctor at a family cancer clinic**. This information may be very important for you or for other family members in reducing the risk of cancer" [AOCS Notification letter, original emphasis].

A list of contact details for all FCC's throughout Australia was included to facilitate the follow-up process. The data reported below were collected in interviews with a subset of participants or their nominated next of kin who had received this notification letter.

Data collection and analysis

Between May 2010 to September 2011, in-depth semi-structured telephone interviews were conducted by two student researchers (AC and LP) and two experienced cancer genetic counsellor psychosocial researchers (MG and MAY). The following themes were explored in the interviews: recollection of receiving the notification letter, understanding and responses to the letter, family communication and factors influencing decision making about follow-up with an FCC. A topic guide informed the interviews incorporating questions and probes relating to themes emerging in earlier interviews.

With consent, all the interviews were tape-recorded and transcribed verbatim. The interviews were anonymised and pseudonyms allocated. An inductive approach was used for analysis: the transcripts were coded according to the method of constant comparison.[15] This iterative process involves systematically identifying, comparing and coding themes within and across interviews and accounts for deviant cases. Emerging patterns and relationships between the codes led to the development of second-order categories. Analytical rigor was achieved by other members of the research team reading the interviews to confirm the themes and coding.

The preliminary data analysis resulted in a publication reporting the experiences and responses of the AOCS participants and their NoK to receiving notification letters.[14] However, an emergent theme was evident where participation in an interview for many participants acted as an intervention to facilitate the process of accessing an FCC. Therefore, a secondary analysis of the data was undertaken by AC, LP, LF and MAY, re-examining and coding the transcripts to clarify the process and barriers that participants' experienced when acting upon the information in the notification letter and contacting an FCC.

RESULTS

Participants

The AOCS study identified 109 participants with a *BRCA1* or *BRCA2* pathogenic mutation. Seventy-eight notification letters were sent to 33 participants and 45 NoK (Figure 1). In 27 cases, a notification letter was not sent either because participants had opted for no follow-up ($n=2$), had deceased and not nominated a NoK to be contacted ($n=3$) or because the family was already known to a FCC and were already aware that a mutation segregated in their family ($n=22$). Finally, we were unable to contact/trace four participants or their nominated NoK. Thirty-five (45%) recipients contacted an FCC for genetic counselling and testing to confirm their carrier status. Three (5%) were already known to an FCC or knew their carrier status. Thirty-nine (50%) have not contacted an FCC to date. One notification letter was returned to sender. Of the 78 participants sent a notification letter, 59 were eligible to participate in this psychosocial study and were sent an invitation letter. Twenty-five individuals from 25 families were interviewed (30–90 min in duration; 42% uptake). Ten were AOCS participants (AOCS) and 15 were NoK (see Table 1).[14]

Outcome following receipt of the notification letter

Seventeen (68%) of the 25 individuals in this analysis had contacted an FCC, and 12 (48%) subsequently made an appointment (Figure 2). Of the 5 (20%) who contacted an FCC but did not make an appointment, 2 (8%) were in the process of making an appointment in terms of returning missed telephone calls or obtaining a GP referral, and 3 (12%) had attempted to make an FCC appointment although were unable to do so.

Eight participants (32%) had not contacted an FCC after receiving the notification letter: one said they explicitly did not want to explore this option, one said they could not recall receiving the notification letter, and one regretted not following up the information in the letter but thought she had missed her only opportunity to contact an FCC.

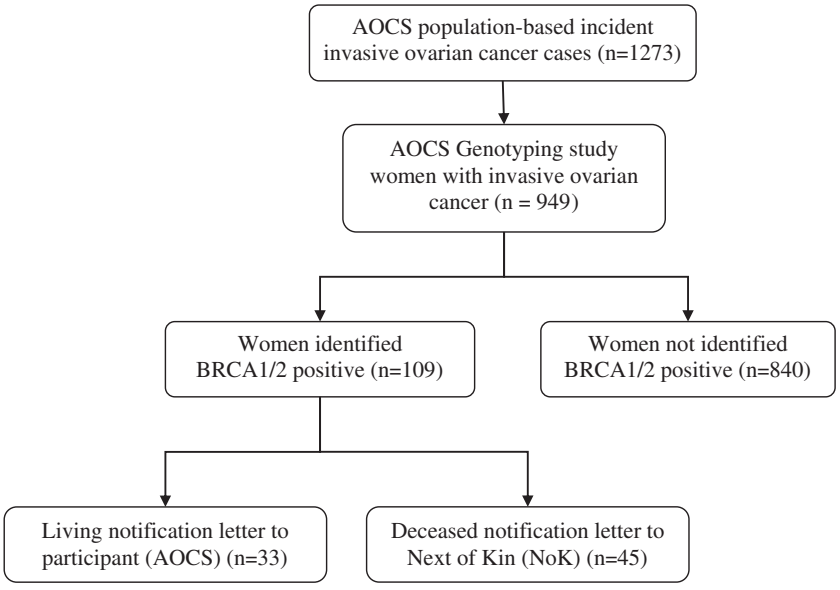

Figure 1 AOCS main study participant's flow diagram.

Table 1 Interviewees' demographic characteristics[12]

	AOCS (n = *10*) *Mean (range)*	*Next of kin (NoK)* (n = *15*) *Mean (range)*
Age (years)	65 (53–80)	42 (28–62)
Years since diagnosis	6 (5–8)	NA
	n	n
NoKs' relationship to AOCS participant		
Daughters	—	12
Husband/partner	—	2
Daughter-in-law	—	1
Educational level		
No post-school qualifications	2	4
Post-school qualifications	8	11
Marital status		
Married or living as married	7	11
Divorced or separated	2	2
Widowed or never married	1	1
Children	8	13
No. with daughters	6	10
No. with sons	7	0

The remaining 5 (20%) had not yet acted on the letter, although said they intended to do so.

Although it was not the initial intention of the researchers at the conception of this study, at the completion of the research interview, five participants (20%) were actively referred to FCCs by the genetic counsellor psychosocial researchers, MG and MAY. These participants included three who had unsuccessfully attempted to make an FCC appointment and two who had not yet contacted an FCC.

The results are presented in two overarching themes: factors influencing failure to access an FCC, and the psychosocial research process acting as an intervention to facilitate action after receiving a notification letter.

Factors influencing failure to access FCCs

There were three factors impacting participants' access to genetic counselling at an FCC: (1) lack of knowledge about AOCS research results among health professionals staffing the FCCs, (2) organisational health system pathway to access a genetic counselling appointment, and (3) geographical locations of the FCCs.

FCC health professionals' lack of knowledge about AOCS notification letter. The AOCS researchers had notified all FCCs who were detailed in the notification letter about the research before commencing notification. Despite this, three participants recalled they perceived a lack of knowledge about the AOCS among health professionals at the FCCs and/or the FCC staff failed to follow up participants' enquiries.

> 'well the person I rang, they were… a bit at a loss to what the letter was about and so they… did they tell me to ring somebody else? I think something like that happened.'
>
> Minnie, 43, NoK, daughter

> 'The girl I spoke to said I'm sorry, I don't know anything about that. I explained where it [the letter] was from. I said I've been given an identification number to quote and she said that really doesn't mean anything to me. She said I will go and check. She came back and said nobody here seems to know anything about this. I said well can you get someone to look into it and get back to me… She took my details and I've never heard from them again'
>
> Edna, 56, AOCS

Health professionals' lack of knowledge led one participant to doubt the validity of the study.

> '[the genetic counsellor had] never heard of the research… I wondered whether [the study] actually did exist'.
>
> Ella 50, NoK, daughter

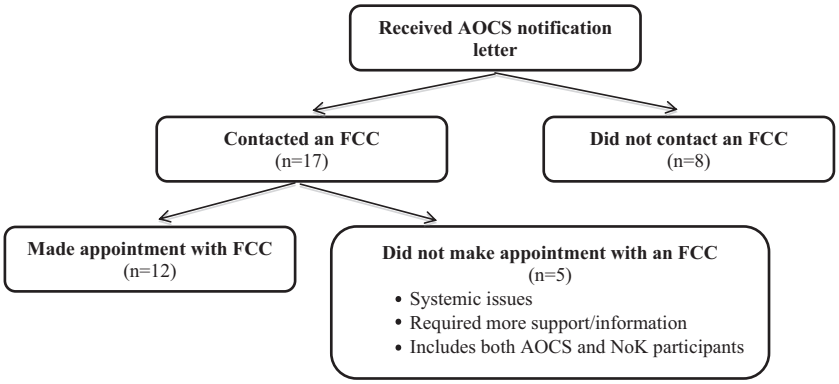

Figure 2 AOCS psychosocial interview study participant's flow diagram.

Organisational health system pathway to access a genetic counselling appointment. The notification letter mentioned a referral may be necessary to attend an FCC appointment. Still, many participants talked about the difficulties of navigating complex clinic referral pathways.

'….the genetic counsellor said you've got to go back to your doctor and get a referral, and then you get an appointment to see a genetic nurse, or something or a counsellor, before we go on any further. I started getting really frustrated because I thought, …wouldn't now be the time to do it rather than waiting to go through all these steps?'

Ella, 50, NoK, daughter

'I've written some things on this letter, go to GP, get referral to Doctor … send that in.'

Minnie, 43, NoK, daughter

Some women were also put off by the lengthy delays in obtaining a clinic appointment.

'I have spoken to them [FCC], and they said to go and get a referral to this doctor and they'll fax it down and then I'll go on a waiting list to be looked at……but I mean, how long's the waiting list … it could be 12 months or so….'

Sarah, 50, NoK, daughter

Geographical locations of the FCCs. A number of participants while interested in attending an FCC were concerned by the clinic location and the distance.

'The letter I got was… saying that if I wanted to know the results of my research…to go into one of those hospitals but of course I can't get there … it's just the hours and the location.'

Louise, 62, AOCS

'You'd have to go into town and then from town you'd have to catch another bus, so that means I'd have to catch four buses'

Ruth, 70, AOCS

'I followed it for so long and then…I had to go to [state capital]… to have this discussion… and I didn't do it.'

Margaret, 82, AOCS

The psychosocial research process: an unintended intervention

Recruitment and participation in a psychosocial interview inadvertently increased a number of participants' intention to contact an FCC. For some participants who had not previously made contact with an FCC, receiving an invitation to participate in the psychosocial study reminded and encouraged them to follow up the AOCS notification letter.

'[After receiving recruitment letter to the AOCS psychosocial study] well that's when I made contact again and I thought… I really shouldn't ignore it… it's just a reminder… and it's something I should do'.

Felicity, 41, NoK, daughter-in-law

For other participants, the act of taking part in an interview influenced their reengagement with the information contained in the notification letter.

'[The interview] has gone and opened my eyes up a little bit…Probably shouldn't be so scared of it, actually go and do it…I'm going to dig out that paperwork tonight… I'm going to call tomorrow.'

Josephine, 29, NoK, daughter

Some participants did not completely understand all of the content of their notification letter, and the psychosocial researchers were able to clarify the meaning of the letter.

'I wasn't quite sure what it was about… whether it was … trying to tell me that the family will get cancer or whether it was the reason why I got it or it might come back…

Louise, 62, AOCS

For other participants, very practical information was clarified about the provision of cancer genetic services and costs associated with genetic testing.

'I didn't even know about these places [FCCs]…I don't even know where they are, are they at the hospital or..?'

Ruth, 70, AOCS

'So, Mum and I, we did talk all about this when, before Mum passed away… should go and have this and I said "Mum, I'm not paying $5000 to go and have a test"'

Sarah, 50, NoK, daughter

Another said she had wanted to contact the FCC after receiving her notification letter, but life events had intervened. She regretted not contacting the clinic and mistakenly believed the opportunity was no longer available.

> 'I should have followed it up…That was very wrong of me…the last 12 months seem to have gone so quickly…,'
>
> Margaret, 82, AOCS

Active intervention by genetic counsellor psychosocial researchers. Five participants, who encountered barriers to accessing an FCC or misunderstood the availability of genetic counselling, were actively referred by the genetic counsellor psychosocial researchers. This referral involved the researchers contacting the FCC staff to explain AOCS and the research results and advocating on behalf of the participants. A number of other participants, while not actively referred by the researchers, had aspects of the process of genetic counselling and genetic information clarified during the interview. For others, recruitment to and participation in an interview motivated them to contact an FCC. For one participant, the researcher conducting the interview sought information about the availability of genetic counselling at a hospital geographically closer to home. For these participants, the uptake of genetic counselling in response to a notification letter increased owing to their participation in the psychosocial interview.

DISCUSSION

This psychosocial study examined the factors that influenced research participants' access to genetic services after receiving a notification letter from the AOCS population-based research study. Less than half of the participants contacted an FCC and made a genetic counselling appointment. Although the majority had not made an appointment, many of these individuals encountered barriers to contacting an FCC such as a lack of knowledge about genetic services or logistical barriers (long waiting times and/or the required referral pathway was perceived to be complicated). For many, the psychosocial interview acted as an intervention, as it provided an opportunity to share frustrations or was an opportunity for information and support relating to these results to be provided. This, in turn, facilitated their access to an FCC for genetic counselling.

Barriers to accessing an FCC

A number of participants encountered significant barriers to accessing an FCC after receiving notification that included FCC staff being unaware of the study, long waiting times for an appointment and the need to go through multistep referral processes to access the genetic service. The FCC staffs' lack of knowledge at the point of initial contact dissuaded some participants from further FCC contact despite their intention to follow up the genetic research results. This occurred despite the involvement of stakeholders when planning the disclosure process as recommended by Keogh *et al*,[8] where all Australian FCCs were informed about the study, the research findings and process of notification. This information was possibly not communicated with all staff, including those responsible for the first contact with new patients. This missed opportunity to access an FCC and clarify these participants' genetic status could potentially be inhibiting the early detection of cancer and subsequent improved prognosis for the participant and their family members.[6]

Those participants who may not have a strong family history of cancer or come through traditional clinical referral pathways are unlikely to have a previous 'genetic awareness', such as patients with a strong family history of cancer. In fact, Alsop *et al*[16] found 44% of AOCS participants with *BRCA1/2* germline mutations had no reported family history of breast or ovarian cancer.[16] The barriers experienced by the AOCS research participants in accessing an FCC are likely to be compounded by the dissimilarities to the 'standard' patient population traditionally cared for by cancer genetic services. Participants who may not have personal or family experience of cancer, have not experienced family discussions about diseases 'coming down the line'[17] and who receive a letter unexpectedly informing them of the availability of genetic information may not be able to articulate or personally advocate to genetic staff about why they need an appointment. They are unlikely to be able to tell the frontline staff at an FCC or a genetic health professional that a pathogenic mutation has already been identified in their family through a research study, and therefore, there exists a clinical imperative for confirmatory genetic testing in an expedient manner. Hence, the participant or NoK may not have been able to articulate their reason for requiring a genetic counselling appointment or may not have understood the referral pathways.

The increasing use of new genomic technologies in medical and genetic research may see a significant increase in the number of participants being offered clinically significant genetic research results and subsequently contacting genetic services in response to receiving a notification letter. It is therefore imperative that clinical genetic staff respond to a participants' initial approach in a facilitative manner so as to not unduly negatively influence participants' decision to follow up the notification letter.

Participants in this study did not actively decide not to contact genetic services to clarify their genetic status; rather, they experienced barriers that prevented their access to genetic services. This contrasts with previous studies that have identified specific reasons, such as insurance implications, where research participants have actively declined to clarify their genetic status, which in turn has impacted the uptake of genetic services after receipt of a notification letter.[12,13] However, these identified barriers may provide some explanation of the differences between Australian and International uptake rates of genetic testing after returning research results. The participating centres in the US and Canada of the Colon Cancer Family Register study employed a study genetic counsellor who provided genetic counselling in person or via telephone.[8] The presence of the genetic counsellor is likely to have made it easier for the US and Canadian participants to access information and support in comparison to the Australian participants who were required to actively seek a referral and contact a genetic service not involved with the research study.

Participation in the psychosocial research interview acted as an unintended intervention

The invitation to participate in the AOCS psychosocial research interview prompted a number of participants to follow up the notification letter and a further five participants were actively referred to an FCC by the psychosocial researchers. Although not intended and not usually part of a research interview, this information and support was provided by the psychosocial researchers, who were incidentally experienced cancer genetic counsellors. This provision of genetic counselling through the unintended intervention may be similar to the study genetic counsellors employed internationally and therefore resulted in a similar outcome regarding uptake of genetic testing.[8]

The intensive notification process implemented by kConFab did not increase the numbers of participants who contacted an FCC.[10]

This contrasts with the increased uptake of genetic services by participants after involvement in this current psychosocial study. However, a research nurse rather than genetic counsellor conducted the calls, and the rationale was to confirm receipt rather than discuss the letters' content.

Participation in a psychosocial interview inadvertently facilitated some participants' follow up of their notification letter as the researchers were able to address practical and logistical issues, as well as participants' needs for more support and information. A single notification letter returning research results to participants may therefore not be adequate for some participants to make a decision and contact an FCC to clarify their genetic cancer risk.

Moving forward

These results highlight the need for further consideration of research participants' ability to make a decision about contacting and accessing genetic services based solely on the receipt of a research notification letter. Given the barriers experienced by participants and NoK illustrated in these findings, deficiencies exist in the translation of research results to clinical outcomes. Hence, it may be timely to rethink the method of returning research results to research participants. Although it is accepted that researchers have an ethical responsibility to return some clinically significant genetic testing results to individual participants, it is unclear to what extent researchers should ensure the results are received and understood. How far should the responsibility extend to the researchers? Have researchers got an ethical duty to do more than send a letter? Regardless, it is important to be mindful of the difficulties in fulfilling these potential obligations as the practicalities may impose burdens on researchers and existing logistics and infrastructure.[18,19] This study illustrated how skilled genetic counsellors unintentionally addressed participants' unmet needs through the psychosocial interview. Therefore, could the responsibility to disclose be further undertaken by more clinically experienced services?

From the perspective of the clinical genetics services, a lack of awareness of the existence of research participants and their needs meant that in some instances appropriate and timely health services were not extended to these individuals. More intensive communication between researchers and clinical genetics staff may partially address the logistical access barriers experienced by participants through an increased awareness of their existence and that their needs may be distinct from those who come via a clinical route. However, it should be noted that the AOCS researchers were proactive in their communication to Australian FCCs about the study and the possibility of participants making contact. Additionally, FCC staff could consider the possibility for participants to bypass some of the routine clinic processes that can be onerous for patients (for example, completing a family history questionnaire or verifying cancer diagnoses) as it is likely this data will have already been gathered on recruitment to the original study.[16] These suggestions may go some way to facilitating access to genetic services for participants who wish to act upon the information contained in their notification letter.

Hallowell *et al*[14] recommended that results should be fed back to participants in a multi-step way, which could include web-based interactive information technology, a more family, rather than individual-centred, approach and the option of having genetic counselling input as required at every step in the process.[14] Further strategies that may provide support to participants include the employment of genetic counsellors to explain results to participants in the first instance.[20] Telephone consultations may be adequate if accompanied by comprehensive supporting materials.[20] Given that some participants indicated they required further information or support in their decision making, we speculate a telephone conversation with a genetic counsellor about the notification content may have allowed for an initial explanation of the study results and enabled questions/concerns to be answered with an appropriately trained professional before participants committed to an FCC appointment. Enhancing current practice may be another possibility where participants receive a telephone call from a genetic counsellor to enquire if further information/support or assistance with referral to an FCC is required after receiving their generic notification letter. Alternatively, it may be the case that a notification letter that includes access to a telephone hotline staffed by genetic counsellors may meet the information and support needs of the majority of participants as well as being more cost effective.

There is clear need for future research to investigate the feasibility of different practices of returning research results, particularly given the issues identified in access to information from this study and the costs of face to face disclosure.[21] Such practices would need to ensure a balanced approach so that the conduct of research is not crippled, undue distress to participants is avoided and appropriate follow-up occurs. Furthermore, while the onus regarding participants' low uptake rate of contacting an FCC in Australia has often focussed on participants actions/inactions, this study has highlighted important systemic problems that also impact uptake.

Limitations of study design

Individuals were interviewed at different time points following receipt of the notification letter, and while no consistent differences emerged in their responses, some accounts may have been influenced by hindsight. We were only able to access individuals who wanted to discuss receiving the notification letter. Thus our results may not represent the views of others who did not wish to engage with the study or FCC in any capacity.

CONCLUSION

This study demonstrates that there are a number of complex barriers to research participants accessing clinical genetic services following receipt of a notification letter. The process of gaining access to an FCC, clinic processes and patient informational and support needs emerged as important influences on genetic counselling and testing uptake.

Our study suggests that FCC staff need to be better informed about research projects and the potential impact this may have on their clinical services. Improved communication between researchers and clinical genetics staff may result in increased awareness that participants who have not come through the usual clinical referral pathways are a diverse group, may be less certain about the reason for their contact and therefore require additional information/support before making a decision regarding attending an FCC.

The data offers useful insights to enable researchers and clinicians alike to consider alternative strategies for returning clinically significant research results and may assist the planning of future research projects. An approach that addresses participants' support and informational needs and is inclusive of advocacy and open communication between research and clinical teams may facilitate participants to make informed decisions about following up research results and successfully initiate contact with an FCC.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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