Patient Education and Counseling xxx (2017) xxx–xxx

Contents lists available at ScienceDirect

Patient Education and Counseling

journal homepage: www.elsevier.com/locate/pateducou

Investigating barriers to genetic counseling and germline mutation testing in women with suspected hereditary breast and ovarian cancer syndrome and Lynch syndrome

Josephine Shaw[a], Caroline Bulsara[b], Paul A. Cohen[a,b,c,*], Madeleine Gryta[d], Cassandra B. Nichols[e,f], Lyn Schofield[e], Sarah O'Sullivan[e,g], Nicholas Pachter[e,h,i], Sarah J. Hardcastle[j]

[a] St. John of God Subiaco Hospital, Subiaco, Western Australia, Australia
[b] Institute for Health Research, University of Notre Dame Australia, Fremantle, Western Australia, Australia
[c] Division of Women's and Infants' Health, School of Medicine, University of Western Australia, Crawley, Western Australia, Australia
[d] School of Medicine, University of Notre Dame Australia, Fremantle, Western Australia, Australia
[e] Genetic Services of Western Australia, Subiaco, Western Australia, Australia
[f] Inherited Cancer Connect Partnership (ICCon)
[g] WOMEN Centre, West Leederville, Western Australia, Australia
[h] School of Paediatrics and Child Health, University of Western Australia, Australia
[i] School of Medicine and Pharmacology, University of Western Australia, Australia
[j] Health Psychology and Behavioural Medicine Research Group, School of Psychology and Speech Pathology, Faculty of Health Sciences, Curtin University, Perth, WA, Australia

ARTICLE INFO

Article history:
Received 17 August 2017
Received in revised form 1 December 2017
Accepted 11 December 2017

Keywords:
Genetic counseling
BRCA1/2 testing
Lynch syndrome testing
Barriers
Gynecologic cancers

ABSTRACT

Objective: The aim of the current study was to explore barriers to genetic counseling and testing in women with gynecological cancers deemed at significant risk of carrying a germline mutation.
Methods: A qualitative study using semi-structured interviews and inductively analysed thematically. Eight patients with ovarian or endometrial cancer participated in individual semi-structured telephone interviews that assessed motivation for genetic counseling and testing, perceived benefits and barriers, timing of the approach, perceptions of the referral process to genetic services and locus of control in relation to cancer and health.
Results: Analysis of the interview transcripts revealed five themes relating to perceptions of genetic counseling and testing: *Lack of importance; Level of information received; Timing of referral processes; Fear and anxiety; Resistance to and perceptions of counseling.*
Conclusions: Participants had a limited understanding of hereditary cancer syndromes and did not appreciate the benefits of genetic testing. A consistent approach at the time of referral to genetic services is needed to ensure that the level and format of information is appropriate for patients.
Practice implications: The rationale for genetic testing needs to be better explained to patients and the timing of referral should be based both on treatment priorities and patient preferences.

1. Introduction

Genetic predisposition is a risk factor for ovarian and endometrial cancer. It is estimated that 13% of ovarian cancers are caused by germline mutations in genes such as BRCA1, BRCA2 and the mismatch repair genes associated with Lynch syndrome [1]. Approximately 2% of endometrial cancers are associated with hereditary dispositions such as Lynch syndrome [2,3]. It is an imperative to identify women with germline mutations because there are no effective screening tests for ovarian and endometrial cancers and risk-reducing strategies are available for women at high risk [4]. Furthermore, poly-ADP ribose polymerase (PARP) inhibitors improve progression-free survival and have recently been approved for use in germline BRCA mutation carriers with

* Corresponding author at: St. John of God Subiaco Hospital, Subiaco, Western Australia, Australia.
E-mail address: paul.cohen@sjog.org.au (P.A. Cohen).

https://doi.org/10.1016/j.pec.2017.12.011
0738-3991/

recurrent, platinum-sensitive high-grade serous tubo-ovarian carcinoma [5].

Genetic counseling in conjunction with germline mutation testing enhances patient knowledge of cancer genetics and understanding of risk, thereby helping to inform the decision-making processes regarding treatment and management. Despite this, some patients choose to decline referral to genetic services [6]. Few studies have investigated barriers to genetic counseling and testing in women with gynecological cancer diagnoses and these have been survey-based or retrospective analyses [2,7–12]. To our knowledge, no study has used a qualitative approach. We aimed to explore barriers to genetic counseling and testing in gynecological cancer patients considered to be at significant risk of carrying a germline mutation, using qualitative methodology.

2. Methods

2.1. Participant recruitment

Following Human Research Ethics Committee approval (St John of God Healthcare Human Research Ethics Committee, #1034, 21.09.2016 and the Women and Newborn Health Service Human Research Ethics Committee, #2016098EW, 30.08.2016), purposive sampling [13] was used to identify eligible participants from the records of Genetic Services of Western Australia (GSWA), a statewide clinical genetic service in Western Australia. Inclusion criteria were: women with an ovarian or endometrial cancer diagnosis, referred to GSWA between January 2012 and December 2016 who either declined all genetic counseling appointments; attended for counseling but declined testing; agreed to undergo testing but did not proceed with testing; or initially declined testing but underwent testing at a later time. This yielded a total sampling frame of 31 patients. One patient was excluded from the study due to having an intellectual disability. An invitation letter was mailed to 30 eligible patients along with the Participant Information Sheet and Consent Form. Those who did not contact the researchers to opt out of the study were contacted by phone and invited to participate. If the patient agreed to participate, they were asked to sign the consent form and mail it back to the research team. Once the study consent form was received, the participant was contacted to arrange a suitable interview time.

2.2. Data collection

Interviews were conducted by the lead investigator (JS), a research psychologist, who invited participants to share their experiences through a series of open-ended questions (interview schedule) that were adapted from Appendix B of Schlich-Bakker et al., 2007 [14], which assesses motivation for genetic counseling/testing, perceived benefits and barriers, as well as timing of the approach. Participants' perceptions of the referral process to genetic services were also explored. Questions were developed by the research team and based on previous protocols [2,8]. Interviews were conducted by telephone in accordance with participant preference and were audio recorded and transcribed verbatim. Demographic data were collected prior to the commencement of the interview. No incentives for participation were offered.

2.3. Data analysis

Interviews were analysed using inductive thematic analysis [15] with the assistance of QSR NVivo Version 11.0. The analysis comprised several steps that were based on previous methods as described by Hardcastle et al. [16]. The first step involved *immersion* whereby the interview transcripts were read several times to gain an overall sense of participants' perceptions. The second step involved assigning codes to key concepts. The third step involved deciding whether codes could be amalgamated to form overarching themes. The final step involved identifying and revising themes and checking for overlap. During these steps, inductive analysis was used to detect themes that arose from the data in relation to perceptions of genetic counseling and testing. This is in comparison to a deductive approach whereby themes are decided in advance and used as a basis for interpreting the data. To broaden data interpretation, three interview transcripts were analysed independently by three authors (CB, SJH & JS) who met to share their coding results and to reach consensus regarding identification of themes and their labels. This enabled reflection upon, and exploration of, alternative explanations and interpretations of the data. Pseudonyms were created to protect the identity of individual participants.

3. Results

Of the 30 eligible patients, eight patients participated in the study, twenty-one patients declined study involvement or could not be contacted and one patient died after the invitation letter had been mailed. Table 1 shows the participants' characteristics. Of note, none of the participants were of childbearing age and half had no offspring. Five participants had received a diagnosis of endometrial cancer and were referred to GSWA after they had completed treatment.

Non-responders consisted of twenty-one patients who either declined to participate ($N = 14$) or failed to respond ($N = 7$). Response bias analyses indicated that there was no significant difference in age (t [27] = 0.59, $p = 0.563$), location (x^2 (1) = 0.00. $p = 1.00$) and time of diagnosis (x^2 (1) = 0.21. $p = 0.671$) for participants compared to non-responders. Participants were more likely to have a diagnosis of endometrial cancer relative to non-participants who were more likely to have a diagnosis of ovarian cancer (x^2 (1) = 5.83. $p = 0.009$). The duration of interviews ranged from 20 to 30 min.

Data analysis identified five themes relating to perceptions of genetic counseling and testing: *Lack of importance; Level of information received; Timing of referral processes; Fear and anxiety; Resistance to and perceptions of counseling*. Table 2 provides an overview of the themes with additional illustrative quotes (Table 3).

3.1. Lack of importance

Some participants believed that genetic testing was not important or relevant for several reasons including no perceived benefits; no offspring; or not relevant at their stage in life. Several participants did not envisage any benefits from undertaking genetic testing: "*I couldn't see any benefit to anybody or to me or my family or anybody else*" (Ann, aged 83). Other participants perceived that genetic testing would only be relevant to them if they had children: "*My main reason is because I don't have any offspring*" (Kay, aged 61). A few participants also felt that genetic testing was not relevant to them at their stage in life: "*I'm at the age now I don't care about you know what happens to me now . . . besides I'm 70 odd, time's running out . . . at my time of life what does it matter?*" (Jill, aged 79 years).

The perceived lack of importance attributed towards genetic testing also precluded any discussion with family members concerning a potential gene mutation. In some cases this was related to the family members' genders: "*I'd say if I had daughters maybe I would give it more thought*" (Jan, aged 55). Other participants were happy to discuss genetics with their family but did not feel it necessary to convey the implications or risks to family members:

Please cite this article in press as: J. Shaw, et al., Investigating barriers to genetic counseling and germline mutation testing in women with suspected hereditary breast and ovarian cancer syndrome and Lynch syndrome, Patient Educ Couns (2017), https://doi.org/10.1016/j.pec.2017.12.011