Counsellee's experience of cancer genetic counselling with pedigrees that automatically incorporate genealogical and cancer database information

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Abstract

While pedigree drawing software is often utilised in genetic services, the use of genealogical databases in genetic counselling is unusual. This is mainly because of the the unavailability of such databases in most countries. Electronically generated pedigrees used for cancer genetic counselling in Iceland create a pedigree that automatically incorporates information from a large, comprehensive genealogy database and nation-wide cancer registry. The aim of this descriptive qualitative study was to explore counsellees' experiences of genetic services, including family history taking, using these electronically generated pedigrees. Four online focus groups with 19 participants were formed, using an asynchronous posting method. Participants were encouraged to discuss their responses to questions posted on the website by the researcher. The main themes arising were motivation, information and trust, impact of testing and emotional responses. Most

of the participants expressed trust in the method of using electronically generated pedigrees, although some voiced worries about information safety. Many experienced worry and anxiety while waiting for results of genetic testing, but limited survival guilt was noted. Family communication was either unchanged or improved following genetic counselling. The use of electronically generated pedigrees was well received by participants and they trusted the information obtained via the databases. Age did not seem to influence responses. These results may be indicative of the particular culture in Iceland, where genealogical information is well known and freely shared. Further studies are needed to determine whether use of similar approaches to genealogical information gathering may be acceptable elsewhere.

Keywords: Electronic pedigrees, genetic counselling, genealogy database, risk assessment, cancer genetics, patient satisfaction.

Introduction

Recording the family health history to gain insight into possible inheritance patterns for a specific disorder has been a major tool in medical genetics for many decades (Bennett 2009). This process includes obtaining accurate information about family members, preferably for at least three generations (Eccles 2004). The family history information includes number of individuals, current ages or ages of death of relatives, as well as relevant health information. The information gathering may be done face-to-face, by telephone or via a written questionnaire (Bennett 2012). In the course of taking a pedigree, medical information on relatives is often provided by a family member without the explicit consent of the person concerned.

Genetic counsellors increasingly use electronic recording methods in their work. In 2013, a study of how genetic counsellors use electronic family history tools found that over 70% had used such

tools to record family histories. The same study found that the majority of genetic counsellors felt that linking electronic medical records to a family history tool would be time saving (Widmer, DeShazo et al. 2013). This has been supported by other studies concluding that time and effort could be saved by enabling counsellees to record their family history by electronic methods (Guttmacher, Collins et al. 2004, Hulse, Ranade-Kharkar et al. 2011). One context in which accurate family history is particularly important is in the field of cancer genetics, where counselling can benefit both the counsellee and his or her family by identifying those at risk, providing options for surveillance and preventive treatment (Brewster, Fordyce et al. 2004, Stefansdottir, Arngrimsson et al. 2013, Nelson, Pappas et al. 2014).

While staff of many genetic services uses pedigree-drawing software, the electronically generated pedigrees used for genetic counselling in Iceland differ, as they automatically incorporate information from a large, comprehensive genealogy database and the nation-wide cancer registry. This ability to link cancer and genealogical registries to the pedigree can be used to generate relevant information for the family history, that in turn can be used in cancer genetic counselling (Stefansdottir, Johannsson et al. 2013). In order to do this, counsellees consent to have their family tree generated from the genealogy database of the Genetical Committee of the University of Iceland, which holds accurate genealogy information about Icelanders back to at least 1840 (Tulinius 2011). The history of cancer(s) is then added, using the population-based Cancer Registry operated by the Icelandic Cancer Society (http://www.krabbameinsskra.is). By this method, comprehensive, electronically generated pedigrees (EGP) are made, enabling very accurate family history for risk assessment and calculations (Stefansdottir, Arngrimsson et al. 2013, Lee, Cunningham et al. 2014). However, like other methods of family history taking, this may also help to identify individuals who may be at increased risk of having a mutation in either

the *BRCA1* or *BRCA2* gene, (hereafter referred to as *BRCA* mutations) or other inherited cancer predisposition. Specific founder mutations in BRCA genes are present in the Icelandic population. The *BRCA2 c.771_775del5* mutation *(*formerly known as *BRCA2:999del5),* is carried by appr. 0.6% of the Icelandic population (Thorlacius, Sigurdsson et al. 1997) while the *BRCA1:5193G->A,* is rare and the population frequency has not been determined. Although only 5-10% of breast cancers can be explained by inherited mutations in the BRCA genes, the risk of mutation carriers developing breast or ovarian cancer is considerably raised compared to women in the general population (Janavicius 2010) and early clinical surveillance is advised. Therefore, cancer genetic counselling can benefit both the counsellee and his or her family. Some family members may be aware of their risk, but for others this may occur without their knowledge. Counsellees are therefore not shown the family EGP to protect the privacy of other family members.

Using family history to assess risk of hereditary cancers is a widely used method and an important part of genetic health services. We were however, unable to indentify published literature on the counsellees' experience of having a family history taken, with or without the use of EGPs. The aim of this study was to gain insight into the counsellees' experience of cancer genetic counselling where EGP´s were used to document accurate family history and make a risk assessment. In this study, EGP´s were created using information from two databases; a comprehensive genealogy database and a nationwide population cancer registry.

Methods

A qualitative descriptive approach (Sandelowski 2000) was used to ascertain the views of the counsellees. Focus groups are generally used to collect data through exploration of a topic with a

number of participants, where one or more group facilitators moderate the focus group (Kevern and Webb 2001). Using online focus groups is an adaption of the conventional face-to-face group methods in qualitative research (Hansen Katharine 2006), where all text is available after replies have been made. On the other hand, online focus groups lack the human presence of face-to-face group sessions (Schneider, Kerwin et al. 2002). Some participants find it easier to participate if they do not have to travel or be at a specific place on a specific time (Chen and Hinton 1999). In addition, the visual anonymity of the Internet makes it sometimes easier to give personal information without being identified (Montoya- Weiss, Massey et al. 1998), thus giving participants the chance of revealing only what they want about themselves. Online focus group studies can be done in two different ways. We used an asynchronously method, where the participants log on in their own time to read contributions from others and then post contributions themselves (Tates, Zwaanswijk et al. 2009, Zwaanswijk and van Dulmen 2014). The other way is synchronously, where participants log on at the same time and exchange written sentences on the chosen forum (Fox, Morris et al. 2007). The choice of method must be made according to the topic and availability of participants.

Participants

The participants were individuals from families identified as having a *BRCA* mutation and had attended for cancer genetic counselling between 01.01.2007 and 31.12.2012. In all, there were 158 eligible females and 67 eligible males. All participants had been tested for one of the two known Icelandic founder mutations; the *BRCA1:5193G->A,* and the *BRCA2* c.771_775del5 mutation, the majority for the *BRCA2* mutation. An invitation letter was sent to eligible participants describing the study objectives and requesting participation. We aimed to recruit

between six and 10 individuals to each focus group, with variation in terms of age, gender and genetic status. We believed saturation was reached after the first two focus groups, but continued to recruit to test this. As all participants in the first three groups were women, a decision was made to purposively invite only males to the 4th group.

In all 26 returned consent forms by either email or post and 19 remained as participants, 17 females and two males. The average age of those invited (n=225) was 50.4 (range 23-86 years), while the average age of participants was 52.2 (range 33-69 years). Group one had three women, group two and three seven women in each and group four had two males. Eleven participants were mutation positive and eight were mutation negative.

The forum

The Phpbb forum https://www.phpbb.com/, a free flat-forum bulletin board was chosen for the study. The forum was hosted on the Icelandic Human Genetics Society site, www.mannis.is. To ensure privacy, the board was closed from others than the participants. The Internet Protocol (IP) numbers and email addresses of participants were concealed. Users chose their own user names and passwords when registering. When each group finished the board was taken down and all communications completely deleted.

The Questions

Group one and two received the same ten questions, posted on the board one at a time. A reminder was sent by email to the entire group each time a new question had been posted. In the reminder and on the board, participants were encouraged to post their own questions and comments. A second reminder was sent when a week had passed without replies. When the first two groups were completed, a decision was made to make small changes to the questions, as is

usual in qualitative studies where data collection is influenced by concurrent data analysis (Green et al, 2007). Therefore, groups three and four received 15 questions. Although the content had not been changed, complex questions were presented as several simpler questions. Based on the experience of limited response from the first two groups, for group three, the first seven questions were put on the board all at once, followed later by the last eight questions. These changes resulted in better return of replies. The questions are listed in a supplemental file.

Data analysis

We followed the approach used by Braun and Clarke (2000) for thematic analysis of data. All comments were initially independently coded, by two of the authors (VS and HS) by hand using descriptive coding. The codes were sorted into categories and themes and discussed by both researchers until a consensus was reached.

Results

Emerging themes arising from the results were motivation for testing, informational need for testing, impact of testing, emotional response to testing and EGP (Table 1). One main outcome was that participants did not oppose the use of, and most trusted, the information from the electronic databases from which the EGP´s were sourced. There was some concern about data privacy, however concern about other family members' attitudes to use information from databases was limited and family communication remained the same or increased.

Table 1. Main themes and categories, arising from analysis.

Themes	**Categories**
1. Motivation	Family history Experience of condition Family experience of mutation or testing Awareness Experience (self or family) of genetic counselling
2. Electronic pedigrees (EGP)	Knowledge • Prior knowledge of EGP´s or genetic counselling • No prior knowledge of EGP´s or genetic counselling Family attitude – no worries Requirement Positive attitude towards EGP´s Diverse attitude towards EGP´s
3. Information and trust	Information • Sufficient • Would have liked more later Trust • EGP´s • Genetic counselling • General Data protection/privacy issues/worries Insurance worries
4. Emotional response	Emotions • Negative • Relief • Positive • Verification
5. Impact of testing (not covered in the article)	Waiting time • Difficult or long Family communication • No change or positive • Little • Other Decision making about future Survival guilt • None • Little • Definitely

	Lifestyle changes

Theme 1: motivation

The strongest motivation for seeking genetic counselling was knowledge of the family history of breast cancer:

"I decided to ask for genetic counselling as my mother died at 49 because of breast cancer and my mother's sister at 69," (female, age 45, mutation negative).

Some had knowledge about the mutation in the family:

"I have an aunt who got breast cancer and she had been asked to relay the information to us that we might carry the gene," (female, age 57, mutation positive).

Few of the participants had information on genetic counselling from their relatives. For some, the idea of better cancer surveillance was one of the triggers:

"I had heard about genetic counselling and found it to be of interest mainly because of the surveillance available to BRCA carriers," (female age 55, mutation positive)

Theme 2: electronic pedigrees

While many knew about the use of pedigrees, some did not but were nevertheless impressed by the possibilities offered:

"I had no idea. This is a very cool tool both for families and professionals," (female age 36, mutation negative).

Some were already aware of the use of EGP´s:

"I knew about it. This gene is common in my family and my pedigree has been mapped," (female age 56. mutation positive).

Theme 3: information and trust

The majority trusted that the information from the databases was correct and that the professionals could be trusted with the information:

"Yes I trust the service as much as possible. Still, it is vital to ensure that insurance companies will not be able to access the information"(male age 46, mutation positive).

However, some voiced concern over the amount of data available and were worried that it – especially the mutation results – could be used against them or their descendants later on:

"I had not thought much about it, but knew that something like this had to exist. Of course it is fabulous that this can be mapped, but then it is a question of how long it will be so. Will my children or their children be able to buy life insurance or will it be like: "no, you belong to this family and therefore we will not insure you, and so on?" (female age 44, mutation positive).

Others shared this opinion:

"It is important that information like this is available for the individual himself, asking for them, but not for others. No such information used in research should be identifiable. On the other hand, the individuals themselves should be able to get them," (female age 56, mutation positive).

Information about data privacy was mentioned by participants:

"I have to agree with those who question what happens to the information (genetic). If doing this will result in institutes being able to access the information and use it against individuals or their family in any way, then I am not sure about how good this is. I think it will always be a question of information privacy," (female age 45, mutation negative).

Some of those who broadly supported the idea of using EGP´s were concerned about protection of privacy:

"Yes I knew that it was possible to make such pedigrees. I think it is good if privacy issues are taken care of," (female age 64, mutation negative).

Experience of genetic counselling

The last question was about the quality of the service. The majority of comments were positive. "*When we came before the testing and also when the results were ready, I found the information to be good,*" *(female age 34, mutation positive).*

”*I found the whole procedure nice. The interviews were of good quality. From the beginning I felt secure and that they were really good people"(female, age 55, mutation positive).*

However, some talked about the possibility of improving the follow up:

“*I got warm welcome and good information. However, when the results had been given, I felt that the overall management could have been better. But I know I should have asked for further counselling," (female age 45, mutation positive).*

This was in agreement with others:

"*If there was something, it might be that the whole procedure was sort of mapped beforehand so that you would always know what was happening," (female age 56, mutation positive).*

Discussion

It is estimated that over 96% of all Icelanders have access to the Internet (Iceland 2016). This was the basis of using an online focus group for the study instead of the more conventional face-to-face method. To our knowledge, this method has not been used before in similar studies in Iceland. The asynchronously method enables participants to connect and comment on their own time instead of logging on at specific times but at the same time does not encourage discussion between participants. As for the ratio of males to females, it reflects the clinical situation in breast and ovarian cancer genetic counselling clinics.

The family history taking is an important part of genetic counselling as risk assessment is based on the outcome. The outcome of genetic counselling in families with cancer history has been studied (Codori, Waldeck et al. 2005), but to our surprise we did not find previous studies on the counsellees' experiences of the procedure of family history taking itself.

Trust is an important part of health services. Our participants expressed trust in our method of using EGP´s although some mentioned data privacy in the context. This may have been confirmed by our policy not to share the EGP with the counsellee or other family members in case they hold sensitive information not known to others in the family. However, some participants voiced worries about the possibility that insurance companies or others might use the information "against" the participant or family. The nature of insurance companies demand that all relevant health information is provided, and as genetic testing is increasingly a part of health services, results from them may be included (Joly, Knoppers et al. 2003). However, few had any

concerns for the attitudes of other family members regarding giving and accessing genealogy and cancer information about the family. This may have to do with the general attitude towards genealogical information in Iceland, where genealogical and other personal information is freely exchanged and discussed.

Those who seek genetic counselling usually do so on the basis of family history or their own medical history. It is the job of professionals to evaluate the family history and make decisions about genetic testing based on the level of risk of the person having a particular condition or mutation. A family history of breast or ovarian cancer was the most common reason to seek genetic testing, followed by a family experience of genetic counselling and/or prior testing of other family members.

Good information and support enhance the counselle´s ability to communicate to family members about the testing and the outcome (Lafreniere, Bouchard et al. 2013). Family communication is an important way of disseminating information about genetic testing and many individuals share test results, at least with first-degree relatives (Finlay, Stopfer et al. 2008). One of the roles of a genetic counsellor is to help individuals and families understand complex genetic information and share it with the family (Alliance 2009). Those receiving additional information are more satisfied, especially if this leads to better understanding (Roshanai, Rosenquist et al. 2009). This may indicate that the opportunity for a follow up session to reinforce and expand on the information given could be valued and useful to counselees. As can be expected, when more family members learn about the family mutation, the number of people with some knowledge prior to genetic counselling grows. This can help when giving complicated information to the counsellee, as other family members may have already shared their knowledge

and experience. However, the counselee's prior knowledge does not mean that the counsellor should give less information or shorten the process.

Many of our participants found that family communication had either not changed or was positively affected during the process of genetic counselling. Having the mutation in common seemed to strengthen the bonds in some families. This has been supported elsewhere (Forrest, Burke et al. 2008) and genetic counsellors are well aware of the importance of addressing the family communication with counselees (Mendes A 2015).

To our knowledge, the use of EGP´s that utilise information from genealogy databases with linkage to disease databases, to assist the genetic professional are not used in genetic health services elsewhere than in Iceland, but should perhaps be promoted as means of easier, better and more accurate information for the genetic counsellor. It may be argued that in a country like Iceland where much genealogy information is already available and easily found, the attitude may be different from other countries. However, it has been suggested that national and regional databases hold valuable information and are an under-used and neglected source of information (Bain, Chalmers et al. 1997). In any genetic health service in the world, family history taking is an integral part as well as in many medical services. There, abundant information about families can be found - often without most of the family members being aware of it. This situation is therefore not unique and trust may be in part due to the knowledge that all health records should be confidential. Also, various large genealogy databases exist on the Internet where they can be easily accessed.

While EGP´s in this form are not used elsewhere we suggest that the experience from this study and others (Stefansdottir, Arngrimsson et al. 2013, Stefansdottir, Johannsson et al. 2013), can be

used to facilitate ways of using existing secure databases as means to improve risk assessment. This could especially apply where electronic databases are available, such as in the Nordic countries where comprehensive information is available in both national and cancer registries (Stefansdottir, Arngrimsson et al. 2013, Stefansdottir, Johannsson et al. 2013, Bauer 2014).

Strength and limitations of the study

All participants in our study had been counselled at the same place by members of the same genetics team. Over the period where participants had received genetic counselling, the service evolved and this may have had an impact on differences in experience. While one of the limitations of the study was a relatively low response rate, we felt that saturation of themes was achieved. It may reflect the lack of familiarity with use of an online forum or reluctance to revisit a difficult period in the life of the participant. With increasing use of social media, this is likely to change. The ratio of males to females reflects the clinical situation in breast and ovarian cancer clinics.

Conclusions and implication for practice

The use of EGP´s enables the genetic counsellor to make a faster and more comprehensive risk assessment. While our participants did not oppose the use of the EGP in genetic counselling and gave mainly positive feedback, further studies are needed to determine to what degree this can change clinical management. It is possible that some of our results were culture dependant, as the knowledge of genealogy is high in Iceland. However, similar to other nations, Icelanders are also concerned about data privacy. Our results indicate general patient support for wider use of electronic databases in genetic counselling, although we are aware of the importance of careful planning and evaluation to ensure that systems are fit for purpose and practice.

For genetic counselling practice more generally, it is crucial that counsellors appreciate client concerns about protection of data, as this has implications for the trust relationship between clients and counsellors, with an ultimate impact on the way in which clients view the information provided to them (Skirton 2001). Stringent systems of consent to access personal information, offering support for discussion with relatives and protection of confidential information are already key components of service in many genetic counselling settings (Committee on Health Research and the Privacy of Health Information 2009). However, while genetic health professionals may understand this, it is important that clients are also made aware of the arrangements to access and protect their data. This will enable maximum use of genealogical and disease history information for patient benefit, while enhancing patient confidence in the process.

Compliance with Ethical Standards:

Funding: This study was funded by institutional funding – i.e. no grant.

Conflict of Interest: Author Vigdis Stefansdottir declares that she has not conflict of interest.

Author Heather Skirton declares that she has not conflict of interest.

Author Oskar Thor Johannsson declares that he has not conflict of interest.

Author Jon Johannes Jonsson declares that he has not conflict of interest.

This study was approved by the National Bioethics Committee no 15-038: All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed consent: Informed consent was obtained from all participants included in the study.

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Supplements

Group 1 and 2 questions

1. Why did you go for genetic counselling and what were your ideas about it beforehand?

   I would like you to ponder about the EGP´s we use for the risk assessment, made with the information from the genealogy database and Cancer Registry. Did you know anything about them beforehand – if it was possible to make them? What were your thoughts about the capability to make such extensive pedigrees?

2. Did you have anything against using EGP´s in the genetic counselling and were you worried about other family member's issues or attitude regarding the extensive information in the pedigree?
3. Do you trust the information in the EGP? Any thoughts?
4. What did you think about also having to give your information for a handmade pedigree?
5. When you had to make decision about the genetic testing, how did you feel about your decision? Describe your feelings during the waiting period.
6. Can you describe your emotions right after and for the first few days after you got the results? What about later?
7. Has the family dynamics and communication changed after the genetic counselling. In what way?
8. Do you think that anyone in your family is suffering from survival guilt? (Survival guilt can be described as when one feels "guilty" when not having a mutation, when others in the family have it).
9. Question 10: This question is a bit complicated and long: What do you think has been well done during the genetic counselling? What do you think can be done better? A) Information before the testing, b) information after the testing, c) anything else? Also, is there anything else you want to share?

Group three and four questions

1. Why did you come for genetic counselling?
2. What ideas had you about the genetic counselling prior to your visit?
3. Did you know anything about EGP´s prior to genetic counselling?
4. Can you tell us about your thoughts when you knew that it was possible to get the extensive information from the electronic databases?
5. Were you against using EGP´s in the genetic counselling?
6. Did you worry about others in the family because of the use of the EGP and the information they contained?
7. What did you think about also having to give your information for a handmade pedigree? (This is only for those who had no relatives that have come before to genetic counselling).
8. Do you trust the information in the EGP? Any thoughts?

9. When you had to make decision about the genetic testing, how did you feel about your decision?
10. The waiting period for the results – can you say something about that?
11. Can you describe your emotions right after and for the first few days after you got the results? What about later?
12. Has the family dynamics and communication changed after the genetic counselling. In what way?
10. Do you think that anyone in your family is suffering from survival guilt? (Survival guilt can be described as when one feels "guilty" when not having a mutation, when others in the family have it).
13. What do you think the genetic counselling has done well?
14. What do you think we can do better?
    a) Information before the genetic testing
    b) Information after the genetic testing
    c) Anything else you want to add?