Enhancing family communication about genetics: ethical and professional dilemmas

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Abstract

When a new genetic condition is diagnosed within a family, genetic counselors often describe a sense of responsibility towards other at risk family members to be appropriately informed about their status. Successful communication of genetic information in families is contingent on many factors. While a small number of probands directly state their intention not to inform their relatives, many who do intend to communicate this information appear to be unsuccessful for a wide range of reasons and may benefit from follow up support from a genetic counselor. Drawing on the reciprocal-engagement model (REM) of genetic counseling practice we explore how enhancing family communication about genetics raises a number of ethical and professional challenges for counselors - and describe how we resolved these. A subsequent manuscript will describe the counseling framework we have developed to enhance family communication about genetics.

Introduction

Knowledge of the presence of an inherited condition in the family or the availability of genetic testing can enhance informed decision making about health management and/or reproductive options (Bower et al., 2002) . Genetic counselors describe a feeling of responsibility towards the family of their patients, wishing to ensure that they are informed and aware of the implications of a new genetic diagnosis or test results for themselves (Andorno, 2004; Dugan et al., 2003; Lucassen & Parker, 2004) . However, a combination of practical, ethical and medico-legal factors mean that family members are not usually contacted directly by genetic counselors, who commonly rely on the patient to convey the information (Forrest et al., 2010). Although genetics consultations usually include a discussion about informing family members (Forrest et al., 2007) , a significant proportion of those who are at risk appear to remain uninformed e.g (Costalas et al., 2003). In the literature, ‘non-disclosure’ is usually discussed in relation to a person who directly refuses to

convey information to another family member, e.g (Leung, 2000). However, a prospective study has shown that genetic counselors are rarely told directly by a patient that they do not intend to pass genetic information on to another family member (Clarke et al., 2005). More commonly, patients seem to state an intention to communicate with ‘all’ family members, but in reality don’t do so (Kerzin-Storarr et al., 2002; Landsbergen et al., 2005; Suthers et al., 2006). This has been termed ‘passive non-disclosure’ to distinguish it from the former situation of active refusal (‘active non-disclosure’) (Gaff et al., 2005). By definition, passive non-disclosure will not be evident at a consultation where communicating with family members is first discussed and must therefore be addressed in follow up contact between the patient and genetic counselor. Professional guidelines demonstrate that promoting communication about genetics within families is an appropriate role for genetic counselors and other health professionals (Forrest, et al., 2007). However, addressing non-disclosure of genetic information by patients brings into conflict several aspects of a genetic counselor’s professional practice and ethical code, making it difficult to resolve these tensions in a manner that is both consistent with the ethos of genetic counseling and feasible in practice. These tensions were highlighted for us when we were asked to develop a ‘genetic counseling intervention to enhance family communication’ for a funded randomised control trial study. Funding had been provided on the basis that the intervention would increase the number of family members informed. A goal of increasing the number of informed family members and, in particular, *intervening* to achieve this goal seemed to us to be inconsistent with the non-directive stance of genetic counseling. It was not possible to resolve this to our satisfaction by simply putting together a protocol of counseling skills and strategies, instead we found ourselves returning to and re-examining the fundamental principles of genetic counseling practice. Despite the extensive discourse on non-directiveness in the literature, it was the tenets of the reciprocal-engagement model (REM) of genetic counseling practice

(Veach et al., 2007) that proved most useful in assisting us to identify and articulate the problems we were experiencing.

In this paper, we draw on the REM to describe the conflicts we faced and the way we were able to reconcile these to develop a genetic counseling model that, in the words used in a randomised controlled trial, would serve as an 'intervention'.

Reciprocal Engagement Model

The 'reciprocal-engagement model' of genetic counseling practice arose from a workshop of North-American genetic counseling program directors. Five tenets of practice were defined:

1. Genetic information is key
2. Relationship is integral to genetic counseling
3. Patient autonomy must be supported
4. Patients are resilient
5. Patient emotions make a difference

While all of these tenets are interdependent and influenced by each other, the genetic counselor-patient relationship is of paramount importance. The counselor and patient are co-informants, with the counselor providing expertise in genetics and counseling and the patient contributing his or her life story. They work together to determine what is in the patient's best interests, with the counselors' central beliefs including that patient autonomy must be supported, patients are resilient and that patient emotions matter. The fifth tenet, 'patient emotions make a difference', recognises the importance of the counselor exploring and attending to patient emotions so that both the patient and the counselor gain insight into the patient's individual concerns as well as the familial context. The ultimate goal is that the patient will use their understanding to make informed decisions, adapt to their situation and cope with their condition. Counselor reflection is an integral part of the REM (Veach, et al., 2007)

The influence of family communication patterns on genetic counseling practice has been previously examined through the lens of the REM (Fox et al., 2007). Although the extent to which the REM reflects actual practice remains to be determined, it does present a model of how genetic counselors can “communicate and assist patients in decision making” (Fox, et al., 2007 p.729). In considering the tenets of the REM, it became clear that genetic counseling around family communication – or rather, lack of family communication – brings the tenet of *Genetic information is key* (being informed is better than being uninformed) into conflict with that of *Patient autonomy must be supported*.

Genetic information is key

Genetic information is fundamental to genetic counseling, with education about “inheritance, testing, management, prevention, resources and research” a core element of this process (Resta et al., 2006 p.77). In particular, the process of informed decision-making relies on the decision-maker understanding the implications of an inherited genetic condition and considering all relevant information about the benefits, risks and options: without such necessary knowledge a person cannot make the choice that is optimal for them (Hodgson & Spriggs, 2005; van den Berg et al., 2006). The centrality of genetic information is reflected in the NSGC definition of genetic counseling, that is “helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease” which includes ‘education about inheritance, testing, management, prevention, resources and research’ and “counseling to promote informed choices and adaptation to the risk or condition” (Resta, et al., 2006 p.77). Consequently, “being informed is better than being uninformed” has been identified as a common stance of genetic counselors (Veach, et al., 2007 p. 719).

Genetic knowledge is no less important for the family member than the patient: family members also have risk(s) arising from their genetic inheritance and consequently options

which could be exercised. In fact genetic information can be seen as co-owned by biological family members, with models of co-ownership of genetic information suggesting that family members should have access to a patient's information when it also has implications for them (Davey et al., 2006; Leonard & Newson, 2010). Genetic counseling practices such as discussion at the consultation, letters to those at risk and follow-up contact (Forrest, et al., 2007) reflect an impetus to ensure all possible efforts are made to inform the relevant family members of their risks and options. In fact, the Australian genetics community successfully lobbied for changes to privacy laws to allow practitioners to directly inform family members of their risk of a genetic condition, although this is an unusual practice and possible only in specified circumstances (Forrest, et al., 2010; Skene & Forrest, 2010). The desire to ensure family members are informed is more evident when there is clearer evidence of medically beneficial intervention. Arguably, promoting dissemination of genetic information is consistent with the philosophy of socialised medicine systems, which aim to maximise the health of an entire community through the allocation of limited health care resources.

The importance of genetic information for family members is commonly recognised by people attending genetics services, many of whom are motivated to attend not only for their own benefit but for that of their biological family members, and who see the benefit to the future health of family members and descendants as an important outcome of genetic testing (McAllister et al., 2011). 'Genetic information is key' is relevant not only to the patient but also to their family members. However, when a genetic counselor learns that a patient has not told (at-risk) family members about the diagnosis of a genetic condition or availability of a genetic test, the 'genetic information is key' tenet of genetic counseling is challenged.

Family members are not fully aware of their situation and options and, in this framework, are therefore unable to make fully informed decisions. Looking with the perspective of only this tenet, a genetic counselor could perceive the patient as failing to respect the shared nature and

centrality of genetic information in providing choices and allowing informed decision making.

Patient autonomy must be supported

The core of this tenet is that patients should make self-directed decisions because ‘the patient knows best’. The concept of patient autonomy within the REM framework has not yet been elaborated in depth. Autonomy as a concept has a “different meaning and a different role in different settings” (Hodgson & Spriggs, 2005 p.92). At a minimum, autonomous choices are those made freely by competent persons and informed by correct information (Beauchamp & Childress, 2001). This does not necessarily entail a deliberative process by the person who has made the decision. Simplistically, supporting autonomy could mean accepting without further exploration a patient’s decision not to pass on genetic information and justified by invoking the non-directive practice of genetic counseling. However, non-disclosure may result in the patient later experiencing regret, guilt or damage to family relationships. Furthermore, any non-disclosure of genetic information to a family member will impact on that family member’s own autonomy. Not knowing this information reduces the family member’s capacity to make an informed – or possibly any - choice in relation to the genetic condition present in the family. Interestingly, an international review of guidelines and position papers on communication of genetic information in families found that many recommended directive counseling to encourage patients to communicate genetic information to their family members (Forrest, et al., 2007). This stance, which prioritises the autonomy of other family members above that of the patient, conflicts with the REM tenets by suggesting that the patient may not be able to make the ‘right’ decision about whether or not to communicate. As patients mostly appear willing to inform other family members, the autonomy of other family members does not come into conflict with that of the patient. However, the challenge for genetic counselors in responding to non-disclosure is to resolve

the tension between supporting the autonomy of their patient, while believing the genetic information to be critical for other family members to make informed decisions about their own health.

Resolving the conflict

In considering these tenets within the context of the goals of a randomised controlled trial, it was clear that a genetic counseling ‘intervention’ must actively support patient autonomy whilst simultaneously maximising the number of family members to whom information is conveyed. Both of these cannot be achieved if supporting patient autonomy is perceived as a passive process or, as White suggests, “non-interference” with patient choices (White, 1998 p.7). Seymour Kessler was the first to describe nondirective genetic counseling as an active counseling process (Kessler, 1997). White expanded upon this, explaining that ‘supporting patient autonomy’ does not mean passive acceptance of choices made by patients, but instead refers to an active process whereby genetic counselors empower patients by engaging them in a supportive and “deliberative” dialogue (White, 1997p.297). A supportive dialogue empowers individuals by recognising their existing resilience and competence. In the context of family communication, a deliberative dialogue can assist people to recognise the way their communication preferences and practices are enacted in response to both conscious and unconscious decision-making processes (Baumeister et al., 2011). It remains patient-centred; by addressing the patients *own* fears, concerns and motivations and considering their influence on any communication and its possible outcomes. In contrast, encouraging the patient to consider the needs of a family member or to put themselves in that person’s shoes is not patient-centred as it shifts the focus away from the patient (Gaff et al., 2010). A person who engages in a patient-centred deliberative process makes an ‘examined’ choice. This may mean a decision not to communicate, thus the genetic counselor is not striving to ensure *all* family members are informed. Rather the genetic counselor is seeking, gradually

to increase patients' adjustment to their situation and their self-efficacy in relation to family communication. We hypothesize that this will *more often* result in patient's communicating with family members, as those who may be initially reluctant to communicate are able to explore and resolve their conscious and unconscious fears, assumptions and projections regarding the genetic information. We suggest that while this kind of deliberative process is unlikely to reduce the frequency of communication it may, on occasion, result in a better informed decision not to communicate/share information.

Application in practice

Utilising a case study, we consider now how such an approach is relevant in responding to both active and passive non-disclosure.

Active non-disclosure

When a genetic counselor is faced with a patient who actively refuses to communicate to a family member, the counselor is well-placed to address this directly in the consultation (see Case Study, Appendix 1). In our experience, it is common for practitioners to wish to persuade their patient to communicate with their family member, see (Adelsward & Sachs, 2003) for an example of persuasive strategy. As indicated earlier, the 'focus' of the consultation may then move from the patient to the family member. This is illustrated in Option A of the Case study. Learning that Mrs Taylor no longer plans to tell her daughters about her *BRCA1* test result, the genetic counselor, Emma, is disconcerted by the apparent 'back flip' and feels concerned that Mrs Taylor's daughters might not be allowed to make a decision for themselves about testing. While tacitly acknowledging Mrs Taylor's feelings – that such communication would be difficult – and less tacitly that Mrs Taylor has control over whether her daughters are told or not, Emma's gaze then turns towards the daughters and she follows a path of exploring the consequences for *them* of non-disclosure. This could

be experienced by Mrs Taylor as both persuasion and a failure by Emma to recognise the nature of her concerns.

In contrast, in Option B, Emma's focus remains on the patient. She sees Mrs Taylor's 'back flip' as reflecting her state of mind in response to learning about the *BRCA1* mutation and remains focussed on Mrs Taylor's emotions rather than her intentions. Emma recognises that, as Mrs Taylor adjusts over time, her intentions may change and Emma creates opportunities to further facilitate the process of adjustment.

Passive non-disclosure

Passive non-disclosure is more subtle than active non-disclosure in that there is no overt refusal to disclose but nonetheless the information is not conveyed to relatives. While active non-disclosure can often be identified during a genetic counseling session, passive non-disclosure only becomes apparent once the patient has had an opportunity to disclose the news to family members as intended and not done so. This gap between intention to communicate and action may be due to an inability to overcome perceived barriers, a belief that the information is not relevant to some family members or reduced motivation to disclose after adverse communication experiences with other family members. It may also reflect a degree of ambivalence about informing relatives. Unless genetic counselors have follow-up contact with the patient, they are likely to remain unaware of this happening and even then only if they specifically explore the patient's experiences when communicating with the family. There is some evidence to suggest that follow-up, with explicit discussion of family communication, by a genetic counselor increases the contact of other family members with genetic services (Forrest et al., 2008). At least some barriers to communication may therefore be resolved through genetic counseling. Patient centred follow-up, remaining focussed on patient's emotions, is likely to assist with adaptation over time thereby enhancing conscious decision-making. As genetic counseling in this context supports the patient's autonomy,

passive non-disclosure does not seem to raise the same tensions between the tenets of genetic counseling as active non-disclosure.

Conclusion

People who feel satisfied that they have fully communicated genetic information to their family may feel that they have discharged their responsibility to all of their family members, even when this is clearly not the case (Gaff, et al., 2005). This discrepancy is more clearly a problem for the genetic health professionals who believe that it is important for ‘all’ at risk family members to know their status. However, encouraging further communication with family members can be seen as coercive by the patient and potentially damaging for their relationship with the service: the counselor is effectively placing greater value on family members’ access to information about their risks than the patient’s autonomy or sense of empowerment. However, to do ‘nothing’ does not sit easily with genetic professionals either, as it deprives family members of potentially valuable information.

We have explored these tensions by returning to the core principles or tenets of genetic counseling as described by the REM , and resolved them by drawing on the Rogerian principle of ‘patient centred counseling’ as it applies to genetic counseling.

Our reflection on these matters leads us to propose that a *genetic counseling* intervention to enhance family communication needs to:

1. Empower patients to make informed and well considered choices about disclosing genetic information.
2. Maintain or enhance patients’ sense of their capacity to communicate, by recognising the patient’s intrinsic strengths, rather than focussing solely on what has not been done, and exploring the patient’s own feelings about and reactions to communicating, both at a conscious and unconscious level.

3. Recognise that harm could result from persuading a patient to communicate when he or she feels uncomfortable
4. Accept that all at-risk family members may not be informed, at least in the first instance.

This type of intervention would enable genetic counselors to respond to active or passive non-disclosure in a manner that remains supportive of the autonomy of their patient while maximising the number of family members who will be informed.

The actual impact of an intervention based on these principles is, of course, unknown. This paper is the first of a series that outlines the development and testing of an intervention to enhance family communication. Here we have described the principles upon which the intervention is founded, elucidated by returning to the tenets of genetic counseling.

Subsequent papers will describe the intervention and its iterative development, pilot testing and outcomes of the randomised control trial.

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Appendix 1

Case Study

Emma, a genetic counselor with two years experience post-certification, is seeing a woman with a personal history of breast and ovarian cancer. Earlier in the consultation, Emma told Mrs Taylor that a causative mutation had been found in *BRCA1*. Mrs Taylor had anticipated this result, but is more distressed than she expected herself to be. While exploring the implications of this result for other family members, Mrs Taylor unexpectedly said she had decided not to tell her daughters, both in their 20s. As one of the reasons Mrs Taylor had wanted testing was to help her daughters, Emma was surprised.

Option A

Emma commented that Mrs Taylor seemed to have changed her mind about telling her daughters. In response, Mrs Taylor was emphatic that testing wouldn't be any use to them and they would not want to know. Acknowledging how hard it would be to discuss with them, Emma asked Mrs Taylor if her daughters were expecting to be told the result. Mrs Taylor repeated that she did not intend to tell them and elaborated that she felt that this was best for them. Emma reminded her that she had previously thought they would wish to know and asked how they might feel if they later learned the result had been withheld from them.

Option B

Emma commented that the result must have had a pretty big impact on Mrs Taylor to change her mind about telling her daughters like that. After a period of extended silence, while Mrs Taylor composed herself, she agreed and said she really hadn't thought it would hit her this hard. She didn't want to put her daughters through this, especially as one of them was looking forward to getting married soon. After giving Mrs Taylor the opportunity to 'sit' with her result and to talk more about her feelings, Emma suggested that Mrs Taylor might find it helpful if they met another time or talked further by phone as she adjusted to the news and its implications.

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