

**Title:** A GENETIC COUNSELING INTERVENTION TO FACILITATE FAMILY COMMUNICATION ABOUT INHERITED CONDITIONS

**Running head:** An intervention to facilitate family communication

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**Abstract** (180 words)

This paper describes the development and implementation of the first intervention to facilitate family communication of genetic information based on a genetic counseling model of practice. The intervention is telephone-based and therefore designed to complement face-to-face genetic counseling consultations. It was developed by firstly reviewing the literature and a model of genetic counseling practice, leading to definition of seven core principles underpinning the intervention. A counseling framework based on these principles was developed through iterative role playing and review, tested for consistency with good practice and piloted on ten study participants. It was found to be feasible to implement and consistent with good genetic counseling practice. Implementation included training of the genetic counselors who would deliver the intervention as part of a randomized controlled trial. Noteworthy deviations from good genetic counseling practice were observed, with unexpected additional insights into the 'black box' of genetic counseling that may have wider implications and would benefit from further investigation. The intervention is currently being evaluated in a randomized controlled trial, to assess its impact on the number of family members attending genetic services.

**Key words**

Family communication, genetic counseling, intervention, randomized controlled trial, model of practice.

## INTRODUCTION

There is an uneasy consensus amongst genetics practitioners that the person who is found to have a disease-causing mutation should be the one to alert biological relatives of their risk (Forrest et al., 2007; Godard et al., 2006). However, the intrinsic complexity of genetic conditions, genetic testing and family dynamics make informing relatives a potentially fraught task for the patient to undertake. The difficulties that arise are evident in patient accounts; even those who report no problems with informing their family do not communicate with all at-risk family members (Gaff et al., 2007; Wiseman et al., 2010). Consequently, people who are eligible for genetic testing may not be aware they have this option.

Guidelines suggest that genetics practitioners should encourage and support patients to communicate relevant information to relatives (Forrest et al., 2007) and this appears to be common practice. For example, in an international study genetic counselors reported that they consistently provide education and counseling to encourage family communication of genetic information, including providing letters to pass on to family members (Forrest et al., 2010). While these strategies appear to facilitate communication (Forrest et al., 2008a; Suthers et al., 2006), people with genetic conditions commonly see it as their own responsibility to inform relatives, sometimes with support from health professionals (Chivers Seymour et al., 2010; Plantinga et al., 2003).

Nonetheless, our experience indicates that genetic counselors remain concerned about those relatives who do not appear to have been informed, particularly when a genetic test is available to them which may substantially influence future health care. For at least the past decade, medical geneticists and genetic counselors have debated whether they should more actively intervene to ensure that patients' relatives are made aware of their genetic risk at sessions on family communication held at national and international professional conferences. Views are polarized. The discussion usually centers on

strategies of persuasion and finding solutions for the patient; strategies which are also evident in clinical practice in healthcare practitioners' responses when patients reveal that they do not intend to communicate with relatives (Gaff et al., 2010). Internationally, guidelines and position papers addressing communication of genetic information within families have a common focus on the uninformed relatives, and advocating directive approaches by practitioners to encourage family communication (Forrest et al., 2007).

In contrast, genetic counseling is a patient-centered process that aims to support patient autonomy and promote informed choices and adaptation to the medical, psychological and familial implications of a genetic condition or risk (Resta et al., 2006). A genetic counselor who actively encourages communication when a patient is reluctant or uses persuasion and solution-finding shifts attention away from the patient and does not promote patient autonomy (Hodgson & Gaff, 2013).

Despite the significance of family communication to genetics services, there has been little consideration and no evaluation published to date of what a patient-centered genetic counseling approach to facilitate family communication would entail. It was recently suggested that "rather than enhancing the content of the information given, genetic counseling strategies that assess personal and familial barriers to communication may provide more targeted support and may facilitate more open sharing within the family" (Montgomery et al., 2013 p.545). A previous study suggested that additional genetic counseling may increase the number of relatives informed (Forrest et al., 2008a) but little information was provided about the intervention used; furthermore the methodological approach of the study limited the conclusions that could be drawn.

In order to more rigorously determine if genetic counseling increases the number of relatives informed (as measured by attendance at genetic services), funding was obtained to conduct a randomized controlled trial. The purpose of this paper is to describe the development and implementation of the intervention, grounded in a genetic counseling model of practice, to facilitate family communication of genetic information. We describe unexpected counselor strategies identified during training in the intervention which may have broader relevance to genetic counseling practice. The impact of the intervention, as measured by the numbers of relatives attending genetic services, is currently being evaluated in a randomized controlled trial (Australia and New Zealand Clinical Trials Register # ANZCTR12608000642381) and the evaluation aims, methodology and results will be reported separately.

## **DEVELOPMENT OF THE INTERVENTION**

Development of the intervention was informed by a framework for complex interventions (Craig et al., 2008). This framework enables development of an intervention which meets the requirements of randomized controlled trials for being replicable and standardized, while also allowing adaption to individual patient needs congruent with genetic counseling practice. A model of genetic counseling and empirical evidence provided the foundation for the intervention and are described below. The complete process of development and implementation is described in Table 1.

The intervention is intended to be applicable to the practice of genetic counseling in any health system. In Australia, the majority of genetic counselors work in hospital-based genetic services which are funded by the relevant state government.

Institutional review was performed by the Human Research Ethics Committee of each hospital with a participating genetic service, meeting Australian requirements for the ethical conduct of research on

humans. Human Research Ethics Committee approval encompassed conduct of the randomized controlled trial and training of the genetic counselors delivering the intervention.

### **Model of practice**

The Reciprocal Engagement Model (REM) of Genetic Counseling (Figure 1) is unique in that it is focussed on genetic counseling practice and integrates both its educative and counseling aspects (Veach et al., 2007). As such, it is well placed to serve as the model of practice for a genetic counseling intervention. Although the goals and strategies of the model have not been fully elucidated, its five central tenets have been explored previously in relation to family communication (Hodgson & Gaff, 2013; Koerner et al., 2010). Tensions potentially arise between the model's tenet of supporting patient autonomy and an intervention which intends to increase the numbers of family members informed by the patient. We have previously suggested a resolution to these tensions, noting that autonomy is supported by a patient-centered, deliberative dialogue and suggesting that such a dialogue may promote self-awareness and self-efficacy (Hodgson & Gaff, 2013).

### **Evidence base**

The intervention was informed by the literature on family communication of genetics information that, at the time, predominantly identified the factors associated with communication and described the experiences of patients. At the time of development, one systematic review described the process and outcome of family communication (Gaff et al., 2007). Using this review, its cited studies and subsequent publications available at the time (as cited in results), we identified further requirements that needed to be met in design of the intervention. These are presented as 'intervention principles' in the following section.

## **Intervention principles**

Table 2 summarizes the requirements of the intervention identified from a systematic literature review of the process of communication (Gaff et al., 2007) and the Reciprocal Engagement Model of genetic counseling practice. The principles defined by the model of practice are from an earlier paper (Hodgson & Gaff, 2013), while those identified from the literature review are as follows<sup>1</sup>:

*Allow opportunity for the patient to communicate:* Communication has been described as a process, rather than an act (Forrest et al., 2003). Most close relatives are told of the diagnosis within a week after the genetics consultation. However, communication may occur over a longer time as the patient considers the effects of disclosure on relatives, what should be disclosed and the 'right time' to do this. Allowing a period of time for deliberation and communication means that the intervention can focus on the challenges that may have arisen in communication, for example, when the patient has not acted on their intention to inform a relative.

*Support adjustment of the patient to their own genetic status.* Evidence suggests that people need to first make sense of what a genetic test result means for themselves before being ready to talk to their family.

*Recognize and explore the diverse motivations for disclosure and non-disclosure.* There are many reasons why patients tell family members about a genetic condition (Wiseman et al., 2010), including for their personal support and for advice. The underlying motivation for communication is likely to influence the clarity of the messages received by relatives (Forrest et al., 2008b). In a similar vein,

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<sup>1</sup> For brevity, the information comes from the systematic review by Gaff et al (2007), unless stated otherwise.

professional responses to non-disclosure need to consider the distinct reasons for not sharing information. Forrest and colleagues (Forrest et al., 2003) suggested that non-disclosure can be broadly categorized into positive, neutral and negative. 'Positive' non-disclosure occurs when a person believes that non-communication will have a beneficial outcome, for example protecting a vulnerable person from bad news. This assessment of the situation by the patient could be accurate. Alternatively, it may be a defensive mechanism: an unconscious projection by the patient of their own unresolved fears and emotions on to relatives as during adjustment to their newly discovered genetic status. Neutral non-disclosure arises when the patient does not perceive need for the person to be informed. Again this may be a realistic evaluation of the circumstances, but this perception may also arise from assumptions held by the patient. Finally, negative non-disclosure occurs when the patient is unable to overcome obstacles such as lack of contact, family conflict or lack of opportunity. Such barriers may be taken at face value but the possibility of unconscious ambivalence also exists (Rollnick et al., 2008 p.34). A deliberative dialogue about family communication would include exploring and responding to conscious and unconscious barriers and is consistent with the REM tenet that patient emotions matter.

A further principle is that the intervention must be feasible to deliver in practice. This will allow rapid application of study findings supporting the use of this intervention in clinical practice. Any aspects which are not amenable to use in clinical practice are likely to become barriers to future adoption.

### **Development process**

The intervention, henceforth referred to as the Genetic Information in Families (GIF) intervention, was developed through an iterative and reflective process by the authors: a genetic counselor with extensive clinical experience and a research interest in family communication (CG) and an academic genetic counselor (JH). Scenarios which required the counselor to consider and respond to the motivations for

disclosure and non-disclosure described in the previous section above were developed and used in a series of audiotaped role plays of genetic counseling. After each role play, we reflected on the interactions and simulated patient reactions to identify a series of common dialogical 'elements' which were grouped as 'domains' within a counseling framework. These were recognized as having commonalities with aspects of Egan's Skilled Helper Model(Egan, 2002), a problem-management approach used in genetic counseling training, and to Motivational Interviewing, a counseling approach to behavioral change(Rollnick et al., 2008 ). The structure of the Skilled Helper model and language of both frameworks was useful in conveying central aspects of the GIF intervention.

## **THE GIF INTERVENTION**

The intervention requisites were considered in both the delivery of the intervention and the counseling framework. Table 3 shows the key aspects of delivery of the GIF intervention, mapped against the requisites described in section 3.1 above.

The counseling framework focuses on three domains. As all genetic counselors are trained in counseling and communication skills, it is expected that a genetic counselor will have the necessary skills and judgment to apply those skills appropriately in each domain. The three domains are: getting the picture, forming an intention, and planning to act. Although these domains appear to be a logical linear sequence, in fact they are interwoven. Aspects of maintaining and enhancing the patient's capacity to communicate, for example, are tightly linked to exploration of the patient's situation and addressing any misconceptions. Furthermore, because there may be many at-risk family members and therefore numerous communication events, 'cycles' may occur as the genetic counselor explores the patient's communication with each relative within a single call telephone call. The extent to which each domain

is addressed in any individual telephone call is at the genetic counselor's discretion, influenced by the patient's personal situation and the point at which the genetic counselor is talking with the patient. For example a first telephone call with a person who has not encountered any challenges in communicating is likely to be different to the third call with a person who has encountered negative family reactions.

### Getting the picture

The genetic counselor aims to gain insight into the patient's current situation, both the events as described by the patient and the potential underlying factors affecting communication. Specifically, the genetic counselor explores with the patient which relatives have been informed, as well as the patient's motivations, experiences and assumptions. For example, the patient may assume relatives understand they are at risk, even though the patient predominantly sought support and did not specifically mention inheritance or risks. The assumption that relatives who have been informed will pass the information on to their offspring may also be erroneous. The patient may also hold misconceptions about the condition or its inheritance, which should be addressed at this time.

*Getting the picture* includes identifying those relatives that have not been informed to date and eliciting the reasons for this given by the patient. Throughout, the genetic counselor keeps in mind the 'positive', 'negative' and 'neutral' reasons for not talking about genetics with relatives and the possibility that the patient is ambivalent or projecting difficult emotions. Accordingly, a fundamental part of the picture is the patient's own response to learning their genetic status at the consultation and their ongoing adjustment.

### Forming an intention

This domain relates to the patient's commitment to act. It includes maintaining or enhancing the patient's capacity to communicate, their intentions and commitment. The genetic counselor does this by providing an opportunity for the patient to reflect on their communication and develop perspective and insight into family communication. The genetic counselor recognizes positive attributes and actions, promotes adjustment (where necessary) and treats communication as a decision to be made by the patient. Over time, the genetic counselor works with the patient to resolve ambivalence. This may result in an intention to communicate. Of course, the patient may instead decide that he or she is not yet ready or willing to communicate with one or more relatives. In this case, the genetic counselor accepts this decision and explores whether the patient does not want the relatives to know at all or is willing for them to learn the information in other ways.

#### Planning to act

If the patient has stated a commitment to communicate to specific relatives, then a plan for communication is developed. The genetic counselor elicits the patient's options to act and encourages the patient to consider details, for example when, where and how they will communicate with specific family members and whether other people will be involved. The counselor may explore the patient's expectations and potential responses of relatives to the news, as a way of preparing the patient for communication.

### **ECOLOGICAL VALIDITY AND PILOTING**

Ecological validity is the extent to which an intervention is appropriate for "real world" settings (Schmuckler, 2001). To ascertain ecological validity, the GIF intervention was presented for feedback at a local forum of genetic counselors and at a national conference of genetic counselors. It was also discussed with two experts, who were key in the development of the REM (Prof Veach, Prof LeRoy). All

concluded that the GIF intervention is consistent with good genetic counseling practice. The intervention was approved for piloting by the RCT steering committee, which included medical geneticists.

The intervention was then piloted. For consistency and familiarity with the intervention, it was delivered during the piloting phase by one genetic counselor only (author JH). Piloting was performed on the first 10 participants recruited to the study to test whether it could be applied to these participants' situations and their receptivity to the process. The only difference between piloting and application of the intervention in the subsequent randomized control trial was the delivery by a single counselor who had not seen the participants in clinic. These telephone calls were recorded and transcribed verbatim. The applicability and relevance of the counseling framework was assessed by the authors through post-phone call debriefing, reflection (JH), and review of transcripts of the phone calls (CG).

We found that the intervention could be applied in each situation encountered. No limitations of the framework were encountered: it allowed the genetic counselor to tailor her responses to reflect the patient's situation, as well as respond to all the issues that arose. Participants were willing to discuss their experiences and were not reluctant to engage in the process.

## **TRAINING**

The genetic counselors who had agreed to deliver the intervention were trained as part of the establishment of study procedures prior to the randomised control trial. Training was conducted once intervention design had been confirmed, that is after the pilot study was completed .

All genetic counselors (n=12) from the four participating genetic services who were either certified or had a minimum of two years genetic counseling experience were trained in the intervention. The average years of experience at the time of training was 7.9 years (range: 2-19 years) and nine were female. Seven were working in general genetics, two were practicing exclusively in cancer genetics and one genetic counselor was working in each of cardiac genetics, neurogenetics and pediatric genetics.

The purpose of training was to ensure consistency of approach and address any pre-existing practices which were not consistent with the intervention. After a group introduction to the intervention, each genetic counselor was trained individually in a one hour session. These sessions comprised a discussion about the genetic counselor's response to the intervention principles, detailed explanation of the counseling framework and delivery.

After the individual training session, each genetic counselor participated in at least one standard mock telephone call to evaluate if they were able to deliver the intervention satisfactorily. Scenarios were developed based on experience with piloting of the interventions (to reflect situations likely to be encountered), and included issues such as reticence to communicate with a family member. Conditions were chosen which would be familiar to all genetic counselors (regardless of specialisation). An example of one scenario is given in Box 1. The trainer played the part of the study participant.

Immediately after the mock telephone call, the genetic counselor was encouraged to reflect on it with the trainer and feedback was provided immediately. The call and feedback to the counselor were recorded and transcribed. Both of these were reviewed to identify divergence in counseling from the principles of the intervention as evident in the transcript or trainer feedback. Similar instances were grouped together as a category. Any concerns about the intervention expressed by the genetic

counselors to the trainer were also documented. Eleven of the genetic counselors required only one training session in the intervention. As one recording of the mock telephone call failed, eleven were available for analysis.

Three types of divergence from the intervention principles were evident in the mock telephone calls.

Two genetic counselors did not challenge patient assumptions about relatives' understanding and intentions. Five attempted to problem-solve or find solutions, for instance<sup>2</sup>:

GC: Right... Do you think that might be more helpful to actually give your mum the letter to pass on so that people are not having to tell people about it and they have a contact number that they can ring up and find out for themselves?

One of the counselors who did not use problem-solving commented in the post-training feedback that it was a conscious effort not to do so. At some point in the mock interview, five genetic counselors were not client centered: interestingly this was manifested by three genetic counselors when they responded to a description of an unwelcome reaction from a relative by explaining normalizing the *relative's* reaction. The opportunity to explore the patient's own reaction and feelings was missed:

P: ..... but mum's a bit more distant about it

[8 conversational turns exploring who has been told]

GC: For grandparents it can be really tough to know that their grandson has got something that they might have unwittingly passed down

P: Yeah

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<sup>2</sup> Key to transcription. Audiotapes were transcribed verbatim. C=genetic counselor, P = participant. For ease of reading some quotes have been truncated without changing the meaning. .... denotes where this has occurred.

GC: It's pretty common for grandparents to feel a tremendous sense of loss and also maybe guilt that they're the ones who passed on this gene change and it's not only affecting their grandson but also their daughter too

Each genetic counselor's individual concerns were also elicited immediately after training, with two areas raised by several genetic counselors. Firstly, there was concern about covering "all that was required". In fact the intervention is deliberately structured to allow exploration over a series of phone calls, in contrast to usual practice where family communication may be discussed as part of a broader agenda in only a single consultation. Secondly, the genetic counselors did not want to be restricted in what they were allowed to say, and were particularly concerned that they would not be able to discuss inheritance or genetics if they thought this was needed. This misunderstanding was rectified by making the Intervention Manual more explicit about addressing misconceptions about inheritance and genetics.

## **DISCUSSION**

The GIF intervention presented in this paper offers an approach which incorporates genetic counseling strategies and targeted support, building on preliminary findings (Forrest et al., 2008a) and our earlier consideration of ethical issues (Hodgson & Gaff, 2013). . Post-consultation, the patient's own genetic counselor explores the patient's feelings and adjustment over (up to) three deliberative, patient-centered phone discussions, identifying the conscious and unconscious causes of any gaps between the patient's intention to talk to specific relatives and his or her communication to date. By first giving the patient time to adjust to their status and to inform relatives, it complements discussion in the consultation (Chivers Seymour et al., 2010; Daly et al., 2001) and provision of written information (Suthers et al., 2006), allowing the issues that have actually arisen to be addressed. The GIF intervention aims to maintain or even strengthen the patient's sense of capacity to communicate.

Communication is treated as a decision to be made by the patient and the intervention aims to empower patients to make an informed and considered choice.

The structure and content of the intervention has similarities to Egan's Skilled Helper Model (Egan, 2002), which is perhaps unsurprising given that this text is used in some genetic counseling training programs and that the three domains we describe are generally consistent with other counseling models. However, a fundamental point of difference is that the underlying 'problem' to be addressed with this intervention is predominantly a problem for the genetic service and at-risk relatives, not necessarily for the patient. There is some evidence that patients feel satisfied with their communication with relatives, notwithstanding adverse responses (Gaff et al., 2005). Patients can feel that they have discharged their responsibility to relatives even though they remain uninformed, at-risk relatives (Gaff et al., 2005). A second point of difference is the centrality of inheritance and genetic information. Genetic counseling has an educative component where information is provided about genetics and health options (Kessler, 1997). Having an accurate understanding of relevant information is considered central to a person's ability to make an autonomous, informed decision. The importance of this aspect was reflected by some genetic counselors during training, who were concerned that a 'counseling intervention' would not permit them to address patient misconceptions about genetics and inheritance or provide additional information if necessary.

Promoting autonomy and self-efficacy are fundamental to this intervention. Recent studies have found an association between self-efficacy and the number of relatives informed (De Geus et al., 2012; Montgomery et al., 2013). We hypothesize that patients will inform a greater number of relatives as a result of both participating in a deliberative dialogue and giving more consideration to any barriers to communication they have encountered. However, the goal is not to ensure that all patients

communicate. After deliberation, the patient may make an informed decision not to disclose to one or more relatives. This is likely to discomfort those genetics practitioners who advocate for practitioner-driven communication. However, it is worth noting that people are usually willing to share genetic information, even with those relatives with whom they would not feel comfortable discussing other general matters (Koehly et al., 2003). If a patient remains reluctant, it is conceivable that existing tensions and conflicts would play out during any discussion about genetics and, as a result, communication would not be effective or the information well received.

### **Practice implications and limitations**

The GIF intervention is firmly grounded in good genetic counseling practice and designed to be feasible for genetic counselors to implement. By following up after the patient has had time to communicate with relatives, it supplements discussion held in the clinical consultation and enables exploration of 'passive' non-disclosure, i.e. when the patient intended to communicate with a relative but has not (yet) done so. Counseling follow-up also complements more mechanistic approaches to facilitating communication, such as letters for relatives, which aim to ensure that patients have the physical resources to pass on accurate information. Finally, it can be applied with families affected by any genetic condition and particularly those for whom a genetic test may be available to relatives.

The GIF intervention describes a counseling approach to family communication about genetics and assumes that genetic counselors have the requisite communication and counseling skills. Although evidence suggests that the goals of genetic counseling are universal, there appears to be considerable variability between genetic counselors in the way they counsel (Veach et al., 2007). The GIF intervention permits the genetic counselor to tailor counseling to meet the patient's needs, however variability in the extent to which the counselor adheres to the guiding principles could impact on the

outcome of the randomized clinical trial. Even though the majority of genetic counselors trained in the GIF intervention understood the issues arising in communication and intent, during training almost half resorted to problem-solving or solution-finding when faced with a patient reluctant to communicate. Our hypothesis suggests that this would reduce the effect of the intervention measured in the randomized controlled trial, although it is also possible that some patients may respond positively to a more directive approach. It is not clear if the use of solution-finding is limited to issues of family communication (reflecting ingrained practices intended to ensure that a relative is aware of their risk), or if the genetic counselors use this strategy in other situations. The process of genetic counseling has previously been described as a 'black box' (Biesecker & Peters, 2001) and the extent to which the Reciprocal Engagement Model reflects the actual practice of genetic counseling has not been tested. Little is known about the techniques genetic counselors use to achieve patient-centered genetic counseling and few process studies have been conducted. As a result, concordance between the goals espoused by the profession and its practice has not yet been demonstrated.

In a similar vein, to our knowledge, this is the first description in the literature of genetic counselors normalizing relatives' responses, rather than those of the patient. Our impression is that the genetic counselors were seeking to promote understanding and consequently minimize any distress experienced by the patient. However, it is possible that a distressed patient may experience this as a justification of the relative's response and lacking understanding or empathy for their own. Further research is needed to more fully understand the use of strategies such as solution-finding and rationalising relative's responses by genetic counselors, as well as their impact. In addition, these findings cannot be generalised to the wider profession and observational studies of genetic counseling practice are needed to ascertain the extent to which these potentially problematic techniques are applied. In the context of this study, telephone calls will be audiotaped to allow adherence to the

counseling framework to be ascertained. This may also provide material for studies looking at variations between genetic counselors within this framework.

## **SUMMARY AND NEXT STEPS**

We have designed an intervention to facilitate family communication which is grounded in the genetic counseling precepts of fostering autonomy and self-efficacy. It is designed to be delivered by genetic counselors, regardless of experience or disease specialization, and supplements existing genetic counseling practices. We have identified the use of strategies, such as problem solving and normalizing relatives' responses, which could impact on the extent to which family communication is enhanced by use of the intervention. However, in order to evaluate whether or not it warrants application in practice, a randomized controlled trial is necessary.

It has been suggested that randomized controlled trials lie on a continuum between 'explanatory', which are performed in ideal circumstances, and 'pragmatic', which are designed to test an intervention under the usual conditions in which an intervention will be applied (Thorpe et al., 2009). The characteristics of the GIF intervention outlined above, and the degree of flexibility that genetic counselors have to tailor it to the needs of a patient, make it suitable for randomized trials that are strongly pragmatic in nature. The results of the randomized controlled trial currently underway will determine the impact of the GIF intervention on uptake of clinical services by relatives. Given the fine line between providing a patient with welcome support and the patient perceiving pressure to communicate, it will also be necessary to evaluate patients' perceptions of the value of the intervention.

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**Conflict of interest**

The authors declare they have no conflict of interest.

## REFERENCES

- Biesecker, B. B., & Peters, K. F. (2001). Process studies in genetic counseling: peering into the black box. *Am J Med Genet*, *106*(3), 191-198.
- Chivers Seymour, K., Addington-Hall, J., Lucassen, A. M., & Foster, C. L. (2010). What facilitates or impedes family communication following genetic testing for cancer risk? A systematic review and meta-synthesis of primary qualitative research. *J Genet Couns*, *19*(4), 330-342.
- Craig, P., Dieppe, P., Macintyre, S., Mitchie, S., Nazareth, I., & Petticrew, M. (2008). Developing and evaluating complex interventions: the new Medical Research Council guidance. *BMJ: British Medical Journal (International Edition)*, *337*(7676), 979-983.
- Daly, M., Barsevick, A., Miller, S., Buckman, R., Costalas, J., Montgomery, S., & Bingler, R. (2001). Communicating genetic test results to the family: a six step, skills building strategy. *Family and Community Health*, *24*(3), 13-26.
- De Geus, E., Aalfs, C. M., Menko, F. H., Sijmons, R. H., de Haes, J. C. J. M., & Smets, E. M. A. (2012). Sharing genetic cancer information with relatives: Development of an instrument to assess counselees' knowledge, motivation and self-efficacy. *European Journal of Human Genetics*, *20*(1), 430.
- Egan, G. (2002). *The skilled helper* (7th ed.). Pacific Grove: Brookes/Cole.
- Forrest, K., Simpson, S. A., Wilson, B. J., van Teijlingen, E. R., McKee, L., Haites, N., & Matthews, E. (2003). To tell or not to tell: barriers and facilitators in family communication about genetic risk. *Clinical Genetics*, *64*(4), 317-326.
- Forrest, L. E., Burke, J., Bacic, S., & Amor, D. J. (2008a). Increased genetic counseling support improves communication of genetic information in families. *Genet Med*, *10*(3), 167-172.

Forrest, L. E., Curnow, L., Delatycki, M. B., Skene, L., & Aitken, M. (2008b). Health first, genetics second: exploring families' experiences of communicating genetic information. *Eur J Hum Genet*, *16*(11), 1329-1335.

Forrest, L. E., Delatycki, M. B., Curnow, L., Skene, L., & Aitken, M. (2010). Genetic health professionals and the communication of genetic information in families: Practice during and after a genetic consultation. *Am J Med Genet A*, *152A*(6), 1458-1466.

Forrest, L. E., Delatycki, M. B., Skene, L., & Aitken, M. (2007). Communicating genetic information in families--a review of guidelines and position papers. *Eur J Hum Genet*, *15*(6), 612-618.

Gaff, C. L., Clarke, A. J., Atkinson, P., Sivell, S., Elwyn, G., Iredale, R., . . . Edwards, A. (2007). Process and outcome in communication of genetic information within families: a systematic review. *Eur J Hum Genet*, *15*(10), 999-1011.

Gaff, C. L., Collins, V., Symes, T., & Halliday, J. (2005). Facilitating family communication about predictive genetic testing: probands' perceptions. *J Genet Couns*, *14*(2), 133-140.

Gaff, C. L., Galvin, K. M., & Bylund, C. L. (2010). Facilitating family communication about genetics in practice. In C. L. Gaff & C. L. Bylund (Eds.), *Family communication about genetics: theory and practice* (pp. 243-272). NY: Oxford University Press

Godard, B., Hurlimann, T., Letendre, M., & Egalite, N. (2006). Guidelines for disclosing genetic information to family members: from development to use. *Fam Cancer*, *5*(1), 103-116.

Hodgson, J., & Gaff, C. (2013). Enhancing family communication about genetics: ethical and professional dilemmas. *J Genet Couns*, *22*(1), 16-21.

- Kessler, S. (1997). Psychological Aspects of Genetic Counseling. IX. Teaching and Counseling *J. Genetic Counseling*, 6(3), 287-295.
- Koehly, L. M., Peterson, S. K., Watts, B. G., Kempf, K. K., Vernon, S. W., & Gritz, E. R. (2003). A social network analysis of communication about hereditary nonpolyposis colorectal cancer genetic testing and family functioning. *Cancer Epidemiol Biomarkers Prev*, 12(4), 304-313.
- Koerner, A. F., LeRoy, B., & McCarthy Veach, P. (2010). Family communication patterns. In C. L. Gaff & C. L. Bylund (Eds.), *Family communication about genetics: theory and practice* (pp. 184-198). New York: Oxford University Press
- Montgomery, S. V., Barsevick, A. M., Egleston, B. L., Bingler, R., Ruth, K., Miller, S. M., . . . Daly, M. B. (2013). Preparing individuals to communicate genetic test results to their relatives: report of a randomized control trial. *Fam Cancer*, 12(3), 537-546.
- Plantinga, L., Natowicz, M. R., Kass, N. E., Hull, S. C., Gostin, L. O., & Faden, R. R. (2003). Disclosure, confidentiality, and families: experiences and attitudes of those with genetic versus nongenetic medical conditions. *Am J Med Genet*, 119C(1), 51-59.
- Resta, R., Biesecker, B. B., Bennett, R. L., Blum, S., Hahn, S. E., Strecker, M. N., & Williams, J. L. (2006). A new definition of Genetic Counseling: National Society of Genetic Counselors' Task Force report. *J Genet Couns*, 15(2), 77-83.
- Rollnick, S., Miller, W. R., & Butler, C. (2008 ). *Motivational interviewing in health care*. New York: The Guilford Press.
- Schmuckler, M. A. (2001). What is ecological validity? A dimensional analysis. *Infancy*, 2(4), 419-436.

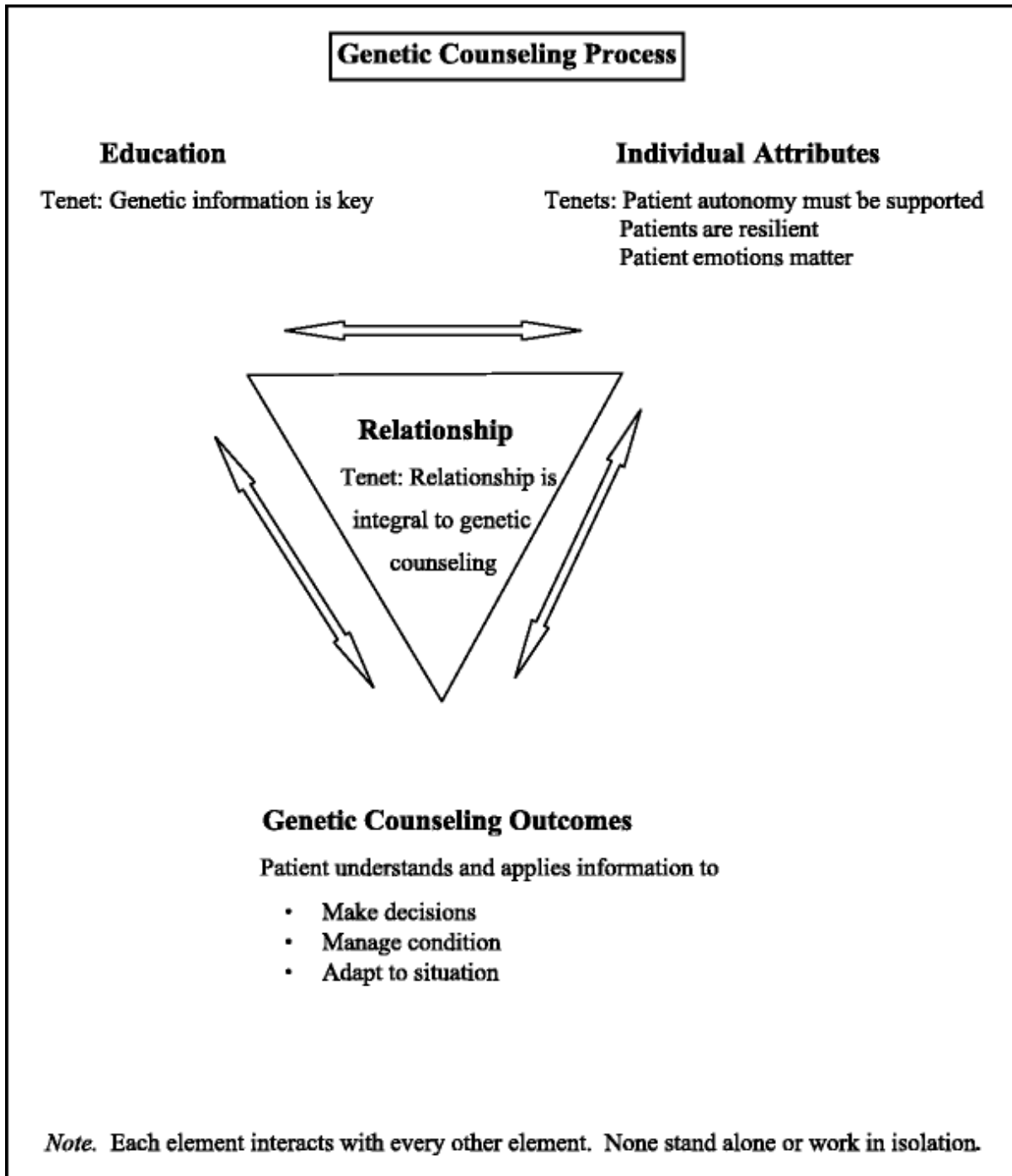
Suthers, G. K., Armstrong, J., McCormack, J., & Trott, D. (2006). Letting the family know: balancing ethics and effectiveness when notifying relatives about genetic testing for a familial disorder. *J Med Genet*, 43(8), 665-670.

Thorpe, K. E., Zwarenstein, M., Oxman, A. D., Treweek, S., Furberg, C. D., Altman, D. G., . . . Chalkidou, K. (2009). A pragmatic--explanatory continuum indicator summary (PRECIS): a tool to help trial designers. *CMAJ: Canadian Medical Association Journal*, 180(10), E47-E57.

Veach, P. M., Bartels, D. M., & Leroy, B. S. (2007). Coming full circle: a reciprocal-engagement model of genetic counseling practice. *J Genet Couns*, 16(6), 713-728.

Wiseman, M., Dancyger, C., & Michie, S. (2010). Communicating genetic risk information within families: a review. *Fam Cancer*, 9(4), 691-703.

**Figure 1** The Reciprocal Engagement Model of Genetic Counseling *Reproduced from Veach et al (Veach et al., 2007)*



**Box 1** Example of a scenario used in training

*Information given to the genetic counselor:*

Adam (4 yrs old) has recently been diagnosed with Fragile X syndrome. His mother Fiona (35 yrs old) had never heard about Fragile X before Adam's diagnosis. You are phoning Fiona two weeks after the clinic appointment where this appointment was made.

Fiona has 2 sisters - Jenny (28yrs old) who has no children and Linda (30yrs old) who has 2 sons (7yrs and 2 yrs). Linda lives 1200km away.

*Information available only to the trainer:*

Fiona says that she has told "all of her close family" and discloses that her husband (James) has found it difficult to talk about Adam's diagnosis.

It becomes apparent that Fiona has only told her mother and sister Jenny, motivated by a desire a support for after Adam's diagnosis. She has not told Linda as they are not close and she assumes that her mother will pass the information on to Linda.

**Table 1:** Process of development and implementation of the GIF intervention

<b>Objective</b>	<b>Methodology</b>	<b>Conducted by</b>
Define the principles underpinning the intervention	<ul style="list-style-type: none"><li>• Review of literature</li><li>• Identified model of practice</li></ul>	Authors
Develop the counseling framework	<ul style="list-style-type: none"><li>• Role plays</li></ul>	Authors
Establish consistency with good genetic counseling practice ('face validity')	<ul style="list-style-type: none"><li>• Consultation with experts and practitioners</li></ul>	Authors
Pilot the intervention	<ul style="list-style-type: none"><li>• Delivery of the intervention to the first ten study participants</li><li>• Reflection and review of the interactions</li></ul>	Author (JH)  Authors
Train the genetic counselors delivering the intervention	<ul style="list-style-type: none"><li>• Group introduction and training session (intervention principles and counseling framework)</li><li>• Individual training session with mock phone call and immediate feedback</li><li>• Review of transcripts of mock phone calls</li></ul>	Authors  Authors, trainer  Authors

**Table 2:** GIF Intervention Requisites and the corresponding REM tenets

<b>Intervention requisite</b>	<b>Tenet</b>
Explore the conscious and unconscious barriers to communication	Patient emotions matter
Empower patients to make informed and considered choices about family communication	Patient autonomy must be supported
Maintain or enhance patients' sense of their capacity to communicate	Patients are resilient
Respect and support the relationship between counselor and patient	Relationship is integral to genetic counseling
Allow opportunity for the patient to communicate	Patients are resilient
Support adjustment of the patient to their own genetic status	Patients are resilient
Recognize and explore the diverse motivations for disclosure and non-disclosure	Patient emotions matter

**TABLE 3:** Key delivery attributes

<b>Key attribute</b>	<b>Rationale</b>	<b>Requirements fulfilled</b>
Timing: post-consultation	Post-consultation delivery allows time for the patient to communicate ‘as usual’. The intervention can then focus on the adjustment of the patient after the consultation and on exploration of the barriers actually encountered.	<ul style="list-style-type: none"> <li>• Allow opportunity for the patient to communicate as usual</li> <li>• Support adjustment of the patient to their own genetic status</li> <li>• Explore conscious and unconscious barriers to communication</li> </ul>
Mode: telephone	A second face-to-face consultation to explore family communication would be onerous for the patient (who is likely to be satisfied with their communication) and difficult to implement in genetic services with waiting lists for appointments.	<ul style="list-style-type: none"> <li>• Feasible to deliver in clinical practice</li> </ul>
Frequency: multiple time points (3,6 and 9 months)	The first call at three months allows some time to adjust to the news, particularly for parents with a newborn affected child. Calls are made at multiple time points as communication with relatives can occur over an extended period of time. This schedule allows patients the opportunity to communicate between calls and receive	<ul style="list-style-type: none"> <li>• Support adjustment of the patient to their own genetic status</li> <li>• Allow opportunity for the patient to communicate</li> </ul>

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support.

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Agent:	Follow up by the genetic counselor	• Respect and support the relationship
Genetic counselor who saw the patient at the consultation	reflects the 'real world' situation and maintains the relationship established by the counselor in the consultation.	between counselor and patient • Feasible to implement in practice

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