A family genetic risk communication framework: guiding tool development in genetics health services

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Abstract Family communication of genetic risk information is a complex process. Currently, there are no evidence-based interventions to help genetics professionals facilitate the process of disclosure within families. This study was designed to create a framework to assist in the development of tools to support patients in communicating genetic risk information to family members. A systematic review identified the factors relevant in communicating genetic risk information in families. A guiding theory for the proposed framework was selected and populated with the factors identified from the review. The review identified 112 factors of relevance. The theory of planned behaviour was selected to guide framework development, organising the framework in terms of the patient's attitudes about disclosure, perceived pressure to disclose and

perceived control over disclosure. Attitudes about disclosure are influenced by a desire to protect oneself or family members, and the patient's perceptions of relevance of the information for family members, responsibility to disclose, family members' rights to information and the usefulness of communicating. Perceived pressure to disclose information is shaped by genetic professionals, family members and society. Perceived control over disclosure is affected by family relationships/dynamics, personal communication skills, the ability of the patient and family to understand the information and coping skills of the patient and family member. The family genetic risk communication framework presents a concise synthesis of the evidence on family communication of genetic information; it may be useful in creating and evaluating tools to help genetic counselors and patients with communication issues.

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Introduction

Genetic testing can provide information about health risks to family members beyond the patient being tested. Passing on information about genetic tests to other family members can provide them with useful information for their own health decision making but may be complicated because not everyone welcomes such information. From the clinical perspective, genetic counsellors sometimes face tensions between their perceptions that other family members should be informed about potential genetic risks, patients' rights to confidentiality and family members' rights to receive (or decline) this information (Godard et al. 2006). Research suggests that, in general, most patients do not actively resist passing on genetic information to their relatives when it would be of

benefit, but they face multiple issues in working out who needs to know, what to tell them, when, and how (Forrest et al. 2003; Wilson et al. 2004). Several literature reviews have illustrated that family communication is a complex behaviour (Gaff et al. 2007; Wilson et al. 2004; Wilson and Etchegary 2010; Wiseman et al. 2010). However, little attention seems to have been given to developing evidence-based interventions to support professionals in their efforts to facilitate familial communication of genetic risk information (Suthers et al. 2006; van der Roest et al. 2009).

Noting that family communication is a complex issue, we based our approach on the Medical Research Council Complex Interventions Framework (Campbell et al. 2000, 2007). This suggests that trials of complex health care interventions should be preceded by careful development work, preferably informed by an explicit theoretical understanding of both the problem(s) an intervention is designed to address and the rationale for why it might be effective.

Our starting point for this work is that individual patient autonomy must be respected; therefore the general outcome of interest, from a clinical perspective, is that genetic risk information is shared *according to a patient's wishes*—this implies an informed decision to disclose risk information. Some patients wish to share risk information but face problems in doing so (Forrest et al. 2003; Chivers Seymour et al. 2010).

The aim of the project reported here was to develop a framework which would organise the factors apparently relevant to family communication at an individual patient level, from the perspective of a health care provider, in order to clarify the nature of issues a patient might face in making a decision about disclosure to relatives and in the process of communication itself. Such a framework would help genetic counsellors promote informed decision making, ‘diagnose’ disclosure or communication problems in their patients and identify the most appropriate interventions to assist them in their communication decisions. The framework would also assist research by highlighting intervention research to meet specific types of communication issues and by focusing attention on specific indicators by which they could be evaluated.

Methods

We conducted the study in two parts: a systematic review to identify the important factors involved in family communication of genetic risk information and synthesis/organisation of this information into a family communication-specific framework.

Systematic review

The purpose of the systematic review was to assemble comprehensive evidence regarding family communication of genetic risk information as a first step to populating a

framework. Our starting point was a published systematic review (Wilson et al. 2004) which we expanded and updated to capture all relevant reports from 1900 to 2007. The detailed systematic review methodology, including search strategy, can be found in the Electronic supplementary material (ESM 1). Two people independently screened titles and abstracts for relevance. Studies were retained if: they included participants who had undergone genetic testing for any disorder or were known to be at increased risk for a genetic disorder because of a family history; they aimed to explore, examine or assess any aspect of communication among family members about genetic risk; and used explicitly described qualitative or quantitative methods to address a clearly defined research question. We excluded narratives, commentaries, book chapters and editorials. Because of the wide range of eligible study designs, quantitative synthesis was not considered.

Framework development

Selection of a guiding theory

Anticipating a wide diversity of factors relevant to family communication of genetic risk, we sought an existing general theory or framework as a starting point to organise the results of the systematic review. To do this, we developed five criteria to assist in theory/framework selection: (1) relevance to family communication of genetic risk information; (2) ability to incorporate a wide range of factors which might underlie communication of genetic risk information; (3) ability to delineate or categorise target behaviours potentially amenable to intervention (i.e. modifiable factors) and orientation towards clinical and patient decision making/interventions; (4) ability to predict outcome of interest (whether communication would occur); and (5) evidence of validity. We reviewed the published literature and consulted with experts to identify a range of potential theories and reviewed them against these criteria.

Populating the guiding theory

For each study identified in the systematic review, we recorded the reported factors associated (positively or negatively) with family communication. We organised these into a parsimonious list of individual, distinct, mutually exclusive factors (e.g. we conflated ‘not having access to the contact information for a family member’ and ‘not knowing what country they lived in’ into ‘not having information to contact the family member’). We noted the frequency with which the same factor was identified across different studies. Next, we mapped each distinct factor on to the appropriate construct of the pre-selected general theory, categorising the distinct factors into natural sub-themes within each construct as appropriate. Tables which describe the specific process for each factor included in the framework are included in the ESM 2.

As a first check on the completeness of the framework, we conducted a final additional literature search, applying the previous search strategy to identify eligible reports published in Medline to November 2012. We mapped the findings against the framework and sought to identify all issues or observations in this last set of literature that it could not accommodate.

Results

Systematic review

The original review (Wilson et al. 2004) identified 31 relevant reports. The electronic search update resulted in 6,231 citations (1,575 duplicates), with an additional 211 manuscripts identified by independent review of abstracts and titles and retrieved for further examination. After exclusions we included 38 new

reports from Nov 2003 to Oct 2007 to give a total of 69 individual studies. A flow diagram documenting inclusion and exclusion can be found in the ESM 1.

The research identified was mostly qualitative or descriptive in nature, consisting of surveys (27), interviews (43), evaluations of clinical tools (2), focus groups (1) and case reports and series (2); we identified no controlled trials. Reports addressed aspects of communication between adults and between adults and children. The reports focused mainly on late-onset, autosomal dominant disorders, particularly hereditary breast and ovarian cancer (35 studies), Huntington's disease (11 studies) and hereditary colorectal cancer (8 studies). The large group of other conditions comprised cystic fibrosis, sickle cell anemia, Duchenne muscular dystrophy, chromosome translocations/balanced translocations, haemophilia, hereditary melanoma, inherited high cholesterol and hearing loss (Table 1). Detailed information on included studies and a summary of their main findings can be obtained from the corresponding author.

Table 1 Genetic conditions identified in the systematic review

Disorder	Inheritance	Citations from systematic review
Late onset		
Hereditary breast and ovarian cancer (BRCA1/2)	Autosomal dominant	Adelsward and Sachs (2003), Blandy et al. (2003), Bowen et al. (2004), Bradbury et al. (2007), Claes et al. (2003), Clarke et al. (2005), d'Agincourt-Canning (2001), Daly et al. (2003), Farkas Patenaude et al. (2006), Forrest et al. (2003), Forrest Keenan et al. (2005), Foster et al. (2004), Gadzicki et al. (2006), Green et al. (1997), Hallowell et al. (2003, 2005) Hamilton et al. (2005), Hughes et al. (2002), Julian-Reynier et al. (1996, 2000), Kenen et al. (2004a, b), Landsbergen et al. (2005), Lerman et al. (1998), Lim et al. (2004), MacDonald et al. (2007), McGivern et al. (2004), McKinnon et al. (2007), Mellon et al. (2006, 2007), Miesfeldt et al. (2003), Segal et al. (2004), Smith et al. (2002), van Oostrom et al. (2007) and Wagner Costalas et al. (2003)
Huntington's disease	Autosomal dominant	Bruce et al. (2003), Clarke et al. (2005), Cox (1999), Forrest et al. (2003), Forrest Keenan et al. (2005), Hamilton et al. (2005), Holt (2006), Klitzman et al. (2007), Shakespeare (1993), Skirton (1998) and Taylor (2005)
Hereditary non-polyposis colon cancer (colorectal adenoma)	Autosomal dominant	Adelsward and Sachs (2003), Berth et al. (2006), Gaff et al. (2005), Koehly et al. (2003), Kohut et al. (2007), Mesters et al. (2005), Peterson et al. (2003) and Schroy et al. (2005)
Hereditary melanoma	Autosomal dominant	Kasparian et al. (2006) and Riedijk et al. (2005)
Inherited high cholesterol (LDLR, APOB, PCSK9 and LDLRAP1)	Autosomal dominant or recessive	van den Nieuwenhoff et al. (2006, 2007)
Childhood onset		
Cystic fibrosis	Autosomal recessive	Denayer et al. (1992a, b), Duster (1999), Fanos and Johnson (1995), Henneman et al. (2002) and Ormond et al. (2003)
Chromosome translocation/balanced translocations	Complex inheritance pattern	Ayme et al. (1993), Clarke et al. (2005), Suslak et al. (1985) and Wolff et al. (1989)
Hemophilia	x-linked recessive	Gallo et al. (2005), Gregory et al. (2007), Sorenson et al. (2003) and Varekamp et al. (1992)
Duchenne muscular dystrophy	x-linked recessive	Fitzpatrick and Barry (1990)
Sickle cell anemia	Autosomal recessive	Duster (1999)
Hearing loss (GJB2 and GJB6)	Autosomal dominant	Blase et al. (2007)

Framework development

Selection of a guiding theory

We identified nine candidate theories which we rated against five criteria: (1) relevance to family communication of genetic risk information ('relevance'); (2) ability to incorporate a wide range of factors which underlie communication of genetic risk information ('range of factors'); (3) ability to delineate or categorise target behaviours amenable to intervention ('amenable behaviours'); (4) ability to predict whether communication will occur in varying circumstances ('predictive ability'); and (5) evidence of validity ('validity'). The theories are summarised in Table 2.

Table 2 shows that two theories met all criteria, the social cognitive theory (Bandura 1986) and the theory of planned behaviour (TPB) (Ajzen 1991). We selected the latter for the purposes of our study because it has a greater amount of empirical evidence supporting its validity, it is the most widely used model of the psychosocial determinants of behaviour, and because it has been used across a wide variety of healthcare settings and behaviours (Armitage and Conner 2001; Godin and Kok 1996; Hardeman et al. 2002). Empirical analyses suggest that, on average, its constructs predict about 40 % of the variance in intention and about 20–30 % of the variance in behaviour (Armitage and Conner 2001; Godin and Kok 1996; Hardeman et al. 2002; Sheeran 2002). The TPB has also been used extensively in the health professional and patient setting examining decision making and behaviour (Allaire et al. 2012; Kasper et al. 2012; Simms et al. 2012; Trinh et al. 2012).

The TPB suggests that behaviour (e.g. disclosing risk information to a target relative) is predicted by the strength of an individual's intention to perform it (see Fig. 1). Intention, in turn, is determined by three constructs, i.e. an individual's attitude towards the behaviour (box d), perceived social pressure to perform the behaviour (box e) and perceived behavioural control over the behaviour (box f) (Ajzen 1991).

Attitudes are formed from beliefs about the outcomes of the behaviour (*behavioural beliefs*), weighted by positive or negative evaluations of that outcome (box a). Depending on whether the behavioural beliefs are positively or negatively evaluated, the result will be a favorable or unfavorable attitude toward the behaviour. Subjective norm (SN) is a function of an individual's beliefs about the expectations of other important individuals or groups (*normative beliefs*) and the person's motivation to comply with these groups (*motivation to comply*; box b). Perceived behavioural control (PBC) is predicted by beliefs about factors likely to facilitate or inhibit the behaviour (*control beliefs*) and an evaluation of the power that each factor has to affect behaviour (*power*; box c). Demographic and personality factors are assumed to affect behaviour indirectly through their influence on attitude, norms and control beliefs (Ajzen 1991, 2007).

Populating the guiding theory

A total of 112 discrete factors relating to family communication of genetic risk information were identified through the systematic review. Table 3 summarises those most frequently cited, describes the subsequent theme each factor represents in the framework and the construct of the TPB within which the factor and theme fall. The factors formed 12 themes, 5 within the attitude construct, three within the SN construct and 4 within the PBC construct.

Figure 2 depicts the final family genetic risk communication framework, populated by the findings of the systematic review. It organises the 112 factors into their relevant domains and highlights the way in which they might interact in promoting or preventing 'intention to communicate genetic risk information'.

The factors within each TPB construct should act in a positive or negative way to form an individual's attitude, PBC and SN, which in turn, should predict their intention to communicate. Factors influential in forming any three of the

Table 2 Theories considered for use in developing the family communication of genetic risk framework and ratings against the five key criteria

Theory	Relevance	Range of factors	Amenable behaviours	Predictive ability	Validity
Circumplex model of marital and family systems (Olson et al. 1989)	No	Yes	No	No	Yes
Double ABCX model of adjustment and adaptation (McCubbin and Patterson 1983)	No	Yes	No	No	Yes
The Beaver's systems model (Beaver and Hampson 2003)	No	Yes	No	No	Yes
The McMaster model of family functioning (Epstein et al. 2003)	No	Yes	No	No	Yes
Family communication patterns model (Ritchie and Fitzpatrick 1990)	No	Yes	No	No	Yes
Health belief model (Rosenstock 1974)	No	Yes	Yes	No	No
Protection motivation theory (Rogers 1983)	No	No	Yes	Yes	Yes
Social cognitive theory (Bandura 1986)	Yes	Yes	Yes	Yes	Yes
Theory of planned behaviour (Ajzen 1991)	Yes	Yes	Yes	Yes	Yes

Fig. 1 Theory of planned behaviour (Ajzen 1991)

constructs (their attitude, PBC and SN) with regard to disclosure may be in conflict, may all be positive or may all be negative. Some, all, or none of those particular factors might apply in the formation of each construct for any given individual. In keeping with the TPB, ‘intention’ is construed as the function of all three constructs (Ajzen 1988, 2007). The weight each construct carries for each individual may be different because attitudes, PBC and SN and the factors which influence their formation, will vary between people and situations. Despite these differences, however, the theory defines communication behaviour as a function of these three main constructs. It provides a parsimonious account of a complex behaviour.

In the case of disclosure of genetic test results to at-risk family members, the framework predicts that an individual’s attitude towards disclosure of genetic risk information to a relative is potentially formed by one or more of the following

factors: a desire to protect the relative in question and/or oneself from harmful news, perception of the relevance of the genetic risk information for the relative, perception of disclosure responsibilities, assessment of the relative’s right to the genetic information and perception of how useful the information will be to that relative.

Four themes emerged which affect perceptions of behavioural control: the family dynamics and relationships within the family, the individual’s communication skills, whether the individual and family member has the ability to understand complex genetic information and whether the individual and family member has skills to cope with his or her own bad news as well as the reactions of others.

In terms of the pressure that an individual feels to perform (or not perform) the behaviour (i.e. communicate or not), the literature identified three sources of pressure: other family members, for example, a relative who wants to

Table 3 The most frequently cited factors perceived as affecting communication of genetic risk information among family members according to the number of times cited in the literature

Communication factor	TPB construct	Theoretical framework theme	No. of citations
Desire to protect oneself and family from potentially harmful information	Attitude	Desire to protect	21
Age and maturity level of recipient	Attitude	Desire to protect and perception of relevance	15
Existence of unhealthy family relationships	PBC	Family dynamics and relationships	14
Lack of contact with the recipient	PBC	Family dynamics and relationships	13
Feelings of responsibility to inform the recipient	Attitude	Perception of responsibility	13
Lack of emotional closeness between the recipient and teller	PBC	Family dynamics and relationships	10
Perception that the information is not relevant to the recipient	Attitude	Perception of relevance	8
Belief that the recipient has a right to the information	Attitude	Perceptions of ‘right to know’	7
Feelings of guilt	Attitude	Desire to protect	7
Viewing communication as a means to social support	Attitude	Desire to protect	7
Situational factors of the recipient (e.g. drug abuse, illness, pregnancy and marriage plans)	Attitude	Desire to protect	7

Female sex was the most commonly cited factor (22 citations); however, according to TPB theory, demographic factors of the communicator fall within the external factors influencing the TBP constructs

Fig. 2 TPB-based family genetic risk communication framework

control family information or a receiver of information who does not want to know, genetic professionals or other health professionals and society as a whole in the form of the 'appropriateness' of discussing such topics.

Overall, the strength of an individual's attitudes, perception of control over the behaviour and the pressure to tell or not tell will determine one's intention to disclose genetic risk information. The systematic review identified immutable factors such as culture, past experience, personality, disease severity and gender that are also thought to influence communication of genetic risk. In the model, their effect on communication intention and behaviour are assumed to be mediated through attitude, SN and PBC.

In our check of the framework, our search of MedLine identified 53 additional articles published between 2007 and 2012 which met the original eligibility criteria. All communication-related factors reported could be accommodated in the framework without revision or adaptation.

Discussion

There is a wealth of information available on the topic of family communication of genetic risk information. The wide and varied literature, however, makes it difficult to translate research findings to the development of interventions for patients and their families. Published models created to explain the process of disclosure were developed with the intention to explain a very complex process (Barsevick et al. 2008). For example, the application of the social-ecological model to communication in families with inherited hyperlipidaemia (van den Nieuwenhoff et al. 2007) or Klitzman et al.'s (2007) analysis of the factors considered in relation to Huntington disease risk disclosure. In both cases, the model development was secondary to the exploration of the issue and not the original purpose of these studies. It is reassuring to note that the content of both models is consistent with our proposed framework, suggesting that regardless of the lens

used to examine the issue, similar barriers and facilitators are identified. Family-centric models have also been developed which examine the issues faced by individuals and families arising during genetic testing in general (Rolland and Williams 2005; Peterson 2005). These more general models have highlighted family communication as an important factor and are both consistent with and complement the family genetic risk communication framework.

The value of an organising framework lies in its insights from diverse disciplines and disorders and in offering a parsimonious explanation of family communication factors to act as a bridge to alterations in clinical practice. We suggest that family genetic risk communication framework may facilitate the diagnosis of problems faced by patients, support informed decision making on whether and how to disclose information and the conceptualisation and development of interventions explicitly targeting particular genetic risk communication barriers. However, at this stage of its development, we do not propose that it is a fully validated guide for genetic risk communication intervention development. Nevertheless, given that the framework is derived from an existing theory which itself has been validated, the component constructs may be more amenable to change by carefully targeted interventions than those which come from a ‘black box’ approach (Campbell et al. 2000, 2007; Michie 2008). As a minimum, we hope it will stimulate discussion, and it may find use in its current form as an educational tool for genetic counselors or as an aide memoire in the clinic.

Supporting patients in working through their disclosure or communication issues is unlikely to be a simple task in clinical practice. Tools designed to support clinicians will probably fall under the concept of ‘complex intervention’ (Campbell et al. 2000, 2007), that is, their effectiveness will be affected by multiple factors such as the setting in which they are used, the skills and experience of the practitioner using them, the level of training available, the time available to implement them in practice, how rigidly they have to be applied and other contextual variables. Emery (2005) is an example in genetics of how recognising intervention complexity is useful in tool development. Over a series of studies, Emery et al. illustrated the development of the GRAIDS decision support tool for genetics referral decisions by family physicians, from exploratory research (Emery et al. 1999b), to a simple prototype tool (Emery et al. 1999a, 2000) through to a computerised system supported by specific education and training (Emery et al. 2007). The study reported here is the first step in applying a similar process to developing clinical practice tools to support decisions and actions around family communication of genetic risk. It promotes clarity about the different kinds of underlying disclosure or communication challenges and indicates the socio-cognitive constructs that may be the most amenable targets for specific intervention development.

The study reported here has limitations. Firstly, the systematic review, while comprehensive, was qualitative in nature. Therefore, while the themes identified are probably a realistic representation of the issues, we cannot claim that they represent the frequency of issues encountered by typical populations of patients attending genetics clinics. In addition, there are many other base frameworks which could have been selected as a starting point. Within the genetics literature, for example, the three themes identified by Dancyger et al. (2011) might be a useful starting point: responsibility to tell, perceived emotional and developmental readiness of the information recipient and communication within the existing family culture. Looking outside genetics, the emergence of the theoretical domains framework (Francis et al. 2012; Cane et al. 2012) also offers a more comprehensive approach which draws together the overlapping constructs represented in multiple individual behavioural theories. The framework is generic, and its applicability to the special issues of communication with (non-adult) children may be under-developed. This is an area of special sensitivity with many ethical concerns, and a detailed exploration of this matter was not within the scope of the current research.

The application of the framework presents the possibility of patient- and situation-specific interventions, focused on particular constructs such as attitude to communicating risk information or perceptions of control over communication. Ultimately, we envisage a toolbox of interventions from which the genetics professional may match the appropriate interventions for each client. Finally, the framework could accommodate intervention design for family members other than the index patient.

Conflict of interest statement The authors declare that they have no conflict of interest.

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