

REVIEW

Process and outcome in communication of genetic information within families: a systematic review

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The communication of risk is a central activity in clinical genetics, with genetic health professionals encouraging the dissemination of relevant information by individuals to their at-risk family members. To understand the process by which communication occurs as well as its outcomes, a systematic review of actual communication in families about genetic risk was conducted. Findings from 29 papers meeting the inclusion criteria were summarised and are presented narratively. Family communication about genetic risk is described as a deliberative process, in which: sense is made of personal risk; the vulnerability and receptivity of the family member is assessed; decisions are made about what will be conveyed; and the right time to disclose is selected. The communication strategy adopted will depend on these factors and varies within families as well as between families. Inherent in these processes are conflicting senses of responsibility: to provide potentially valuable information and to prevent harm that may arise from this knowledge. However, the research 'outcomes' of communication have been professionally determined (number of relatives reported as informed, uptake of testing, knowledge of the recipient) and are typically unrelated to the concerns of the family member. The impact of communication on the individual, family members, and family relationships is of concern to the individual conveying the information, but this is largely self-reported. Currently, there is insufficient information to inform the development of theoretically and empirically based practice to foster 'good' communication. The implications for future research are discussed.

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Introduction

The ethos of clinical genetics is to consider the risks of a genetic condition not only for the consultant attending the genetics clinic but also to relatives of that person.¹ The

extent to which the genetic specialist should shoulder the responsibility for ensuring that family members are aware of their risk is a perennial debate in genetics. In contrast to infectious diseases, communication of genetic risk to those considered at risk is 'ethically dubious' in nature, with a lack of clarity about what individuals should reasonably be expected to do, and how professionals should respond when they are aware that communication within a family has failed or is blocked. This will depend in part on the nature of the information available (risk information only or genetic test results) and implications of the condition.

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For instance, a greater imperative may be felt when preventive treatment options or reproductive choices are being restricted. In general, there seems to be an uneasy consensus that genetic services should rely predominantly on the consultand to convey information, except in exceptional circumstances.²

The consultand, however, does not always convey risk information to their at-risk family members. First-degree relatives are consistently given genetic risk information directly by the proband more frequently than the second- and the third-degree relatives. Wilson *et al*³ have comprehensively reviewed the complex factors influencing whether information about genetic risk is shared with family members, describing individual and family characteristics, disease, and sociocultural factors that can affect this process. It has been suggested that disclosure of genetic information is best described as a process rather than an act,⁴ implying a collection of actions rather than a single event. If genetics professionals are to continue to rely on family members to convey information – or, conversely, if they seek greater participation in this process – it is important to extend beyond identifying the factors that influence who in the family will be informed and gain an understanding of the process of communication of genetic information within families and its consequences. We conducted a systematic review of papers to illuminate these aspects of communication, with the intention of informing future practice in the facilitation of communication within families by health professionals.

Methods

Search strategy

Six electronic databases (Medline, CINAHL, Cochrane Library, EMBASE, PsycInfo, and National Research Register) were searched from 1985 to January 2006. The search strategy was broad (high recall and low precision), as this review was part of a wider review of risk communication in genetics.⁵ Searches focussed on risk and communication; genetics, predisposition, screening, or counselling; and service delivery, health services, and organisation. The strategy used for Medline is shown in the Supplementary Material. Follow-up searches included both electronic (Citation Indices) and manual searches (key authors encountered most frequently in the field and reference lists of included papers).

Assessment for inclusion of studies

The inclusion criteria were peer-reviewed publications on the process and outcome of communication of genetic risk to family members. Communication related only to actual discourse, dissemination, or behaviours with case reports, hypothetical, or anticipated communication excluded. Genetic risk included information about the condition, numeric risk, or genetic test results. All papers meeting

these criteria and utilising qualitative methods were included; quantitative data were only included if it specifically provided information on outcomes of communication. Two reviewers assessed all titles and abstracts and examined full-copy papers of potentially relevant references. Decisions were made independently about inclusion or exclusion, with reference to a third reviewer if there was disagreement between the first two.

Data extraction and analysis

Papers identified as containing information about communication of risk were read in detail by two investigators (CL Gaff and AJ Clarke). An aggregative synthesis approach, focussing on summarising data, was adopted.⁶ Relevant findings were coded on data extraction templates in an Access database and are presented in narrative form. Multiple papers from a single study have been extracted and presented as a single study.

Results

Twenty-nine papers from 26 studies were subjected to data extraction. Figure 1 illustrates the search results and a summary of relevant findings is presented in the Supplementary Material. While this review focusses on the communication of risk information, none of the papers included describe the content communication in these terms. Nineteen studies discussed transmission of test results, while the remainder were about the diagnosis or genetic condition or were not specific about what was communicated. The majority of papers ($n = 21$) investigate families with hereditary cancer syndromes, with very few on more 'traditional' Mendelian genetic conditions such as Huntington disease (HD) ($n = 3$) or cystic fibrosis (CF) ($n = 3$) or chromosome anomalies ($n = 1$).

Three themes relating to the review topic emerged from analysis of the data: deliberation before communication, communication strategies, and outcomes of communication (Table 1).

Deliberation before communication

If an individual does not 'immediately' communicate information about genetic risk to their relatives, it appears that they undergo a period of deliberation in which decisions about disclosure are made. Hamilton *et al*⁷ describe these deliberations as encompassing (1) consideration of the effects of disclosure, (2) selection of what information to disclose, and (3) planning the timing of disclosure. Forrest *et al*⁴ describe an additional phase of 'making sense of personal risk' before being able to deal with deciding who to tell and what to say, with those who continue to perceive their risk as uncertain or ambiguous most likely to have problems knowing what message to give their relatives. However, the ways in which individuals construct and make sense of their risk is beyond the scope

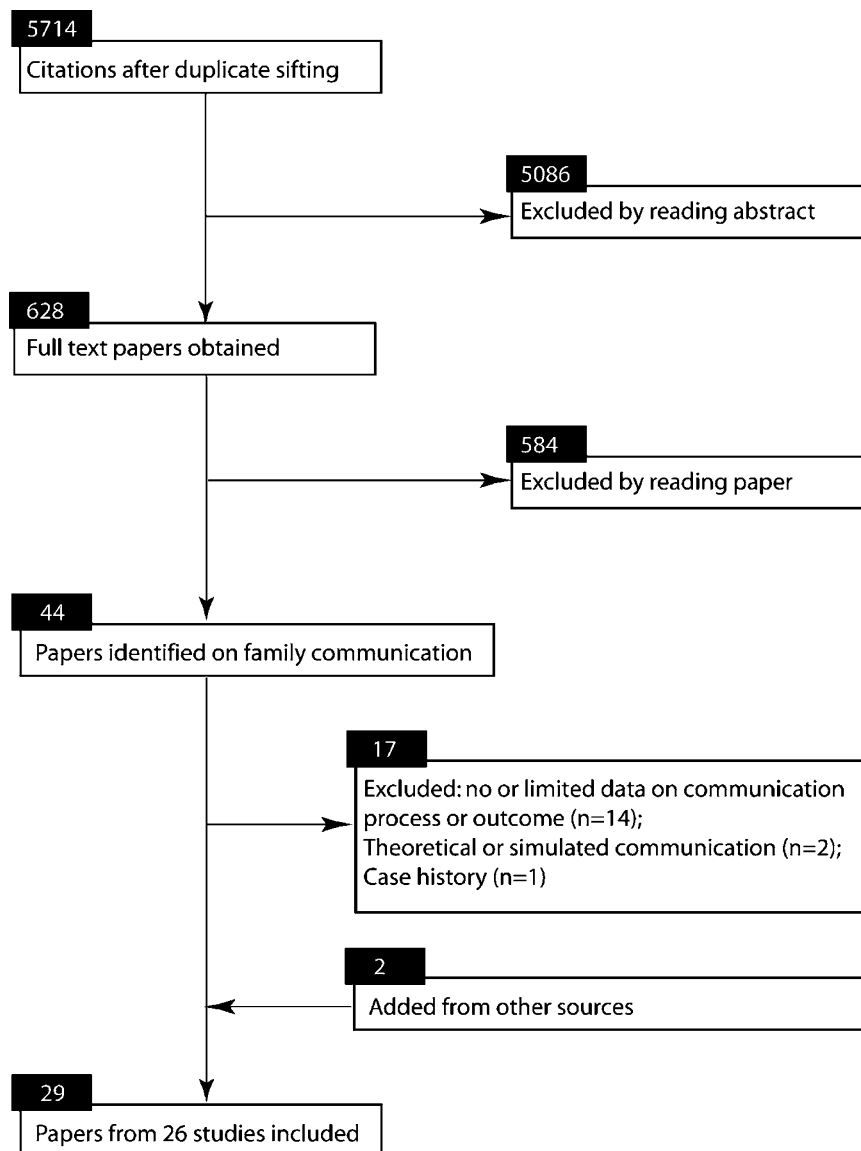


Figure 1 Search results.

of this paper and are presented elsewhere.⁵ The remaining three aspects are discussed here in more detail.

Considering the effects of disclosure Underlying accounts of the decision whether or not to disclose risk information to relatives is a dilemma: the desire to protect relatives from potential harm is weighed against the wish to provide them with information that may have important health consequences.^{4,8,9} In weighing these conflicting responsibilities, the individual assesses their family members' vulnerability to the information as well as their receptivity.⁷ Vulnerability is assessed by considering life circumstances and mental and/or physical condition, as well as the potential harm that may be caused by news that

may be perceived as bad or harmful and able to cause upset or worry.^{4,8,10–16} Receptivity is assessed by predicting the response of the family member to the information (see Outcomes of communication).

Selecting what to disclose Individuals can be selective about what information they disclose. In one study, individuals disclosing their HD test results only provided more information if asked to do so, while those from families with hereditary breast ovarian cancer (HBOC) were more forthcoming.⁷ In contrast, some men undergoing testing for HBOC mutations disclosed only limited information about testing and its implications to their children.¹⁶ Sometimes, selectively conveying information

may delineate what constitutes 'family information' (ie news of a mutation in the family) and what is 'personal' (individual predictive test results).¹⁷

Timing of disclosure Disclosing at the 'right time' is of concern to informants.^{4,7,14,18,19} Generally, communication with close relatives happens within a week and late disclosure is less common.^{12,17,19–22} Forrest *et al*⁴ distinguish pragmatism, where an active approach (such as arranging an appointment or mailing letters) is adopted and prevarication, where the right opportunity is sought. The right 'time' appears to refer to both life stage, for example, availability of surveillance for cancer, forthcoming marriage, or children, and to the right opportunity, for example during normal social contact.⁴ It may also depend on the perceived emotional readiness of the recipient.⁷ It should be noted that communication of results in families with hereditary bowel cancer occurred as part of normal social interaction, but this appeared to be due to a more matter-of-fact approach to hereditary nonpolyposis colorectal cancer (HNPCC) rather than prevarication.^{17,20}

Communication strategies

In some cases, the outcome of the deliberative process will be to decide that the information is not disclosed. Information may be deliberately withheld or kept totally secret.²³ Alternatively, there may be a 'passive' failure to inform due to poor or nonexistent communication pathways and a low sense of responsibility to that individual.^{20,24} Even when communication does occur, it may not be completely open. Hallowell *et al*¹⁸ describes the different strategies observed in men undergoing testing for HBOC: complete openness, where children were informed about testing at the outset and were one of the first to hear of their father's results; limited disclosure, a cautious approach in which involvement in testing was deliberately downplayed; and total secrecy. Kenen *et al*²³ focus on the interaction between informant and recipient. As before, open and supportive communication is observed. Blocked communication was reported with recipients refusing to receive the information directly (eg hanging up telephone) or indirectly (apparent discomfort, lack of responsiveness, or failure to disclose information themselves). Aspects of selective disclosure were described in this study as 'self-censoring', with the informant not pursuing difficult conversations or seeking to protect other family members by only giving them reassuring information. Finally, some women used intermediaries if they felt uncomfortable contacting a particular person. These different patterns were observed within families as well as between families.

The use of intermediaries is observed particularly in relation to parents seen as having the responsibility to inform their children (generational responsibility or vertical transmission).^{4,17,20,25} Thus, an individual may directly inform their siblings but would (perhaps

implicitly) expect those siblings to inform their own offspring. A cascading of responsibility is apparent, with responsibility for informing others transmitted along with the information conveyed. The assumption that at-risk family members have been informed by other relatives^{11,15} is not necessarily borne out, parents do not always inform adult children^{21,25,26} and children under 18 years of age are less likely to be told.^{10,18,21,27} Intermediaries are recruited to inform other relatives,^{9,14,24,28} particularly by men.^{14,20} This appears to be a different concept to that of a 'pivotal' person who perceives him/herself to take responsibility for passing on the information and encourages relatives to consider testing.^{17,29,30}

Outcomes of communication

An obvious outcome of communication is the reported number of family members informed, but others are also apparent in the literature, namely: uptake of genetic testing, knowledge of the recipient, and family responses to the informant.

Uptake of genetic testing Seven studies investigated uptake of genetic testing by informed relatives.^{11,15,17,25,28,31,32} Most of these studies determined uptake in relatives informed about testing: the percentage of those informed who underwent testing ranged from 13%³² to 57–64%.¹⁷ Peterson *et al*¹⁷ found somewhat lower uptake of predictive testing for HNPCC (46%) in a family where disclosure had been delayed compared to families where there had not been a delay (57–64% uptake). Blandy *et al*²⁸ found that second- and third-degree relatives, when aware of the mutation, are less likely to proceed with testing than first-degree relatives and also suggested associations between uptake of testing in first-degree relatives and greater family support of the index case and knowledge of risk of transmission. Landsbergen *et al*¹⁵ compared the characteristics of index cases of families with a low uptake of predictive mutation tests to those of other families, finding significant relationships between low uptake and, not disclosing in person, less involvement with psychosocial workers during testing, and other variables relating to family dynamics. The authors suggest that less attention to psychosocial issues in counselling sessions may have had an inhibiting effect on the process of dissemination.

Knowledge of the recipient Little is known about the knowledge of the family member who has been informed by the proband. Denayer *et al*²² assessed knowledge of CF among siblings of parents of a child with CF, finding that most had only a superficial knowledge about the inheritance of this condition. While 80% reported that the family was a source of information about CF, mentioning CF parents as information source was not significantly associated with the genetic knowledge score. Binedell

*et al*³³ found that those presenting for HD predictive testing were able to give a risk figure of 50% but had misconceptions about inheritance. Hallowell *et al*¹⁶ found uncertainty about risk in women whose father had tested negative for a BRCA1/2 mutation. This is consistent with data showing that the person communicating the information may themselves have poor knowledge.¹¹

Impact on individuals, relatives, and family relationships Study design has precluded direct assessment of the responses of family members to the risk information given by consultands, with almost all accounts provided by the consultand.^{10,18,20,21,23,24,27,28,34} In only one study did some of the recipients of the information directly give their reactions to researchers, indicating their dissatisfaction with the limited disclosure or secrecy of their parents and demonstrating misconceptions about their risk.¹⁸

Anticipating or actually communicating with relatives can be experienced as burdensome or difficult by the informant,^{4,8,10,13,14,26,28} with consultands reporting adverse reactions by some family members, often typical of responses to bad news, or a high emotional temperature.^{18,20,23,24,28} A list of these is provided in Table 2. Even where there were no difficulties in communication, respondents expressed concern about the quality of their relationship with their relative or spouse in the future.¹⁰ There are reports that relationships had been strengthened after disclosure.^{9,36}

The impact on children and young adults of family communication might be expected to be different from that of adults. Carriers of BRCA1/2 mutations have reported concerns expressed by their children about their mother's future health.²¹ Although disclosure of BRCA1/2 results to children aged under 18 years did not relieve parental distress,³⁴ neither did it impact on parent-child functioning.²⁷ Indeed, some carrier mothers in another study perceived their relationship as closer after testing.²¹

Discussion

This review has summarised literature on process and outcome of family communication. As might be expected, communication is not uniform between or within families and the nature of interaction between informants and recipients differs. For example, a person may convey information openly and easily to some family members, but in others conversations may need to negotiate the direct refusal of the information by the family member or subtle cues that the information is unwelcome. It is therefore unsurprising that communication is, in some situations, immediate, while in others there is a delay and deliberation. This process may not be completely conscious on the part of the individual who undertakes, or is given, the task of communicating, but is evident in accounts of disclosure and nondisclosure.

It appears that communication occurs when a sense of responsibility to provide the family member with potentially important information outweighs concern about harming the individual by imparting 'bad' or potentially unwelcome news. This could be termed a 'calculus of responsibility' and demonstrates one side of the exchange of mutual obligations of trust among kin. Taking responsibility for transmitting information implies acting responsibly towards fellow family members. To act responsibly is to assess the potential consequence of sharing or withholding information and is embedded in a moral economy consisting of (mutual) assessments of competence and maturity, as exemplified by the deliberative communication strategy. Communication of genetic risk information is the result of intricate, existing ties of differing strengths.

Arguably, to genetic professionals, the potential psychosocial harm to family members is a relatively abstract concept compared to the health implications of nondisclosure. Consequently, there may be an inclination to prioritise the sharing of information over a desire to protect; reports of the experiences of genetics professionals tend to focus on overcoming problems that arise in family communication,³⁸⁻⁴⁰ despite recognition that these are the exception rather than the rule.³⁸ An active approach to communication of genetic information by health professionals has been actively debated, particularly for familial hypercholesterolaemia.⁴¹ However, to date it has been rare that genetic professionals intervene directly and other means to foster the client's capacity or willingness to communicate are preferred.^{39,40} There are hints that this may be possible: the provision of psychosocial support is associated with greater family communication and, where emotional problems blocked the flow of information, the desire for more support was expressed retrospectively.¹⁵ While families believe that the responsibility lies with them to inform family members, they also want health professionals available in a supporting role.^{4,21} This seems to go beyond the simple provision of written materials. Suggestions about the way professionals may enhance family communication have been made,^{2,15,16,42} but this systematic review did not identify any papers evaluating in practice the ways to foster the capacity or willingness to communicate in families. Caution should be exercised when moving beyond the descriptive studies identified in this review to those testing an intervention. Rather than considering all failure to communicate as the same and testing a 'one-size-fits-all' solution, it may be more fruitful and ethical to recognise the diverse nature and causes of poor communication and address each with a distinct approach. The categorisation of Forrest *et al*⁴ could be particularly useful here. They suggest that declining to inform may be perceived as (1) positive – acting in order to produce benefit (such as preventing harm), (2) negative – failing to act or overcome barriers, or (3) neutral – perceiving that nothing is needed. Each of these categories

Table 1 Summary of results

<i>Reference</i>	<i>Primary study focus</i>	<i>Data collection method</i>	<i>Condition information communicated</i>	<i>Findings</i>
Adelsward <i>et al</i> ⁸	Obtaining family information	Qualitative: observation of 31 consultations	Familial cancer Family history	Communication with family was not seen as problematic in half the consultations Evidence of difficulty speaking about cancer, being responsible for transmitting information to those who do not want to be informed and to those who are unsuspecting, as well as resisting the doctors' claims for information
Ayme <i>et al</i> ³¹	Uptake of test	Quantitative: audit	Balanced translocation Karyotype result	806 (44.4%) of 1816 at-risk relatives had been karyotyped. In 52 families (18.4%) no one had undergone testing
Binedell <i>et al</i> ³³	Experiences of at-risk individuals	Qualitative and quantitative data from 54 individuals	HD	Test requesters are more likely than nonrequesters to have been informed about testing by their relatives.
Blandy <i>et al</i> ²⁸	Family communication and its outcomes	Quantitative and Qualitative (questionnaire) data from 30 women	Condition BOC Test results	15% of all relatives informed were tested Men were reported as incredulous, difficult to inform and difficult to motivate to have testing. Strategies to inform relative included the use of intermediaries and letters.
Bonadona <i>et al</i> ¹⁰	Response to test results	Qualitative: interview with 23 individuals	Familial cancer Test results	All had told at least one close family member within a month. Participants were concerned about the potentially harmful nature of the information and conveying it accurately, with nine seeing it as a heavy responsibility. None reported family conflicts as a result of disclosure. But five expressed concerns about future relationship with their partners and other relatives.
Claes <i>et al</i> ¹¹	Family communication	Qualitative and quantitative: interviews of 56 women with structured questions and psychometric testing	BOC Test results	Most did not want someone else to inform their family. Of the participants with mutations who informed relatives, 40% did not have a single relative to undertake predictive testing. There was no association between informing relatives and uptake, but there was an association between informing distant relatives (all vs some/none) and uptake of testing by distant relatives ($P < 0.05$) Only a minority tried to inform distant relatives systematically. When close relatives and distant relatives were not informed, it was often because the woman assumed that they would have been informed by other relatives
Costalas <i>et al</i> ²⁶	Family communication	Quantitative: telephone survey of 162 individuals	BOC Test results	Some of those with nonconclusive results conveyed inaccurate interpretation of their results Siblings were reported as having difficulty in understanding results compared to adult children ($P = 0.001$) Mutation positive probands were more likely to report upset relatives than those who conveyed negative and indeterminate results ($P < 0.001$). There was no significant difference between reported distress of siblings and children hearing the results.
D'Agincourt-Canning ¹⁴	Family communication	Qualitative: interviews with 31 women, 5 men	BOC	Probands who were carriers or had indeterminate results reported being upset when explaining their results to some relatives. Feelings of responsibilities and obligations are evident. Although some women made efforts to inform all family members by letter, no obligation

Table 1 (Continued)

Reference	Primary study focus	Data collection method	Condition information communicated	Findings
Denayer <i>et al</i> ²²	To determine relatives' awareness of CF risk and how it is discussed.	Quantitative data from questionnaires of 183 aunts and uncles of CF patients questionnaire	Genetic information and test results	was felt to inform relatives they did not know and some relied on certain family members to act as the liaison for more distant but known family. Burdens of disclosing included dilemmas about who, when, and how to tell; concern about causing anxiety; feelings of responsibility for putting descendants at risk and fear of being blamed; family members not wanting to know
			CF	Knowledge about the risk of being a carrier or of having a child with CF was poor, with a minority able to provide this information
Forrest <i>et al</i> ⁴	Family communication	Qualitative data from 37 interviews	Genetic transmission	In all, 80% were informed of the hereditary nature by family, 10% had no information at all. Media was important when the family was not a source
			HD	Two styles of telling are described: pragmatism (active practical terms) and prevarication (looking for opportunities within the normal processes of family life, trying to find the right moment). The right time is often when the first key life decision affected by the disease needs to be made (eg serious relationship, marriage) or when perceived as old enough to understand.
Keenan <i>et al</i> ²⁹			BOC	Health professionals needed at times in supportive role
			Genetic information and/or test results	Both groups needed to make sense of their own risk (whether technically accurate or not) before dealing with whether and what to tell relatives.
Foster <i>et al</i> ⁹	Family communication	Qualitative data from 15 interviews	BOC	Parents are seen as having the primary responsibility to hand on information to offspring. Responsibility may extend vertically (ie grandmother has more authority than an aunt, even if she is not at risk and the aunt is) 'Pivotal people' may take the responsibility for informing relatives and encouraging testing
			Genetic testing	Participants experienced dilemmas balancing social role expectations and talking to relatives about genetic testing. The relative's carrier status had an impact on how and whether the women had difficulty in disclosing own results Most women experienced barriers to communication that restricted information flow regardless of test result and some did not talk a great deal about it They were likely to tell in person but delay informing if they were a noncarrier. This was also true if people had encouraged an affected relative to have a test and then been found not to have the mutation themselves. Disclosure could bring relatives closer but there was concern that estranged relatives will use the information against them The motivation for informing was usually to raise awareness of choices and predictive testing seen as good news; there was no evidence of dilemma of disclosure
Gaff <i>et al</i> ²⁰	Family communication	Qualitative data from 12 telephone interviews	HNPCC	Responsibility to second- or third-degree relatives was seen as discharged by telling parents of relatives. Men sought women's assistance with disclosure. Most would not change the way they communicated

Table 1 (Continued)

Reference	Primary study focus	Data collection method	Condition information communicated	Findings
Green <i>et al</i> ³⁵	Family communication	Predominantly Qualitative data with 46 preclinic and 40 postclinic interviews	Availability of testing	Family responses were reported in practical terms and ranged from interest to lack of interest in testing
			HBOC	The perceived usefulness of discussion about who to inform was variable and gender dependent, with men seeing it as more useful
Hallowell <i>et al</i> ¹⁶	Family communication	Qualitative data from 17 interviews with men, 8 spouses and 4 adult children	Genetic information	Generational responsibility was apparent in the collection of information before attendance, with mothers being the key informant and approaching other relatives on the proband's behalf, as was the desire to protect from distress
			HBOC	Approaches to family members to give information were often in the context of regular social contact, in order to reduce the alarm of being solely contacted about the possible cancer risk
Hamilton <i>et al</i> ⁷	Family communication	Qualitative data from 29 interviews (in person, by e-mail or telephone)	Testing process and test results	The majority of mothers and fathers saw communication to children as shared responsibility and both did in fact share telling of intention to undergo testing, but ongoing discussions were more with mother and children.
			HD	Parents tried to balance children's rights to information about their risks with parental duty not to cause them any (potentially) needless anxiety, resulting in different communication strategies, even within one family, in response to the perceived vulnerability of the daughters
Kenen <i>et al</i> ^{19,23}	Communication, including family communication	Qualitative data from 21 interviews	Test results	All men deliberately managed the timing of disclosure within immediate family, with it usually occurring once a decision about testing had been made or results were available, not during decision-making period.
			BOC	Three communication strategies were identified (see text). Some regret at communication pattern expressed by individuals
Landsbergen <i>et al</i> ¹⁵	Family communication and outcomes	Predominantly quantitative data from 50 questionnaires	Test results	Seven of 24 told family immediately after receiving their test results and relayed the information
			BOC	Remainder communicated test results in three phases (see text), Related to readiness of teller and receiver. Teller needed to prepare themselves to be emotionally ready. Closest relatives were told first and then prepared to tell others
Landsbergen <i>et al</i> ¹⁵	Family communication and outcomes	Predominantly quantitative data from 50 questionnaires	Family history	Differences between conditions observed with respect to content and attempts to influence testing decisions of relatives
			Test results	Mothers were protective of their children and can find it difficult to decide what and when to disclose
Landsbergen <i>et al</i> ¹⁵	Family communication and outcomes	Predominantly quantitative data from 50 questionnaires	BOC	Mothers were only concerned about daughters and granddaughters. There was variability in the openness of relationship with sisters
			Test results	Five communication strategies were identified (see text)
Landsbergen <i>et al</i> ¹⁵	Family communication and outcomes	Predominantly quantitative data from 50 questionnaires	BOC	The mean percentage of first- and second-degree relatives per family not having testing was 64% (7–100%).
			Test results	Families were classified into low uptake (below 20%) and 'others'. Significant differences ($P \leq 0.05$) in low uptake families include less presence of psychosocial workers during pretest counselling; retrospective desire for more support in the communication with family members; parents

Table 1 (Continued)

Reference	Primary study focus	Data collection method	Condition information communicated	Findings
Liede <i>et al</i> ³⁶	Needs evaluation	Quantitative data from 59 men	BOC Test results	informed less often after disclosure and less personally; and emotional problems blocking dissemination Five men would have liked assistance in communication. 10 said family relationships had changed as a result of the test results, 7 reporting positive changes.
McGivern <i>et al</i> ²⁵	Family communication	Predominantly quantitative data from 38 women	BOC Test results	Parents of at-risk people who were seen as having the responsibility to inform The content of communication included family history, reasons for testing, risk of mutation, feelings, insurance, cost, and surgery On average, 2.1 women and 0.6 men were reported as tested by each respondent. Gender differences were observed
Mesters <i>et al</i> ²⁴	Family communication	Qualitative: Semi-structured interview with 30 members of a registry	HNPCC Condition	Disappointing contact early in dissemination seemed to increase the chance that the index person would give up. If they had a few good conversations, then they were more able to handle a disappointing one Strategies included informing relatives alone, with partner, with another family member, cascade with responsibility being passed on to person told (ie via relative), or no plan. 'No plan' was least common and the remainder were equally employed Where there was no plan, then who was informed was based on assumptions rather than knowledge Some people found communication difficult due to family responses such as shock, anger, shoot the messenger; but people got used to the situation overtime and learnt to cope with feelings. If there was openness in family, then it was a confirmation of an assumption and news spread quickly and easily. Relatives were more likely then to be interested in the news and sympathise with messenger
Ormond <i>et al</i> ³²	Family communication	Quantitative data from questionnaires of 30 parents of a CF child and 18 carriers of CF	CF Test results	First-degree relatives of those with an affected child were usually informed as soon as possible The gender of carrier or recipient did not impact on communication patterns or uptake of screening for any group or category of relatives Few siblings of the respondents had undergone genetic testing (11.5% of all siblings) Communication was often imparted as secondary to the child's diagnosis
Parsons <i>et al</i> ¹³	Carrier identification	Qualitative: interviews with 19 parents	CF Test results	Communication with family members was discussed in terms of obligation Discussion with family members was seen as the most problematic consequence of newborn screening Difficulties arose because of pregnancy, number of people, having to contact ex-partners, not enough information, and not knowing how to tell
Peterson <i>et al</i> ¹⁷	Family communication	Predominantly qualitative data from 29 family members and 10 spouses from 5 families	HNPCC	Probands wanted family to have genetic testing and believed it was important, at-risk relatives saw testing as an opportunity to gain additional information related to HNPCC than a necessity

Table 1 (Continued)

Reference	Primary study focus	Data collection method	Condition information communicated	Findings
Koehly <i>et al</i> ³⁰			Test results	<p>Probands and mutation carriers took the lead role in communication. Most shared with at least one first-degree relative within 2 weeks. The same pattern of communication was used as for other nonurgent family news: spouse, children and siblings first. Noncarriers perceived communication to others as less relevant and were less involved</p> <p>It was considered inappropriate to discuss with nieces/nephews directly unless their parents present; the nuclear family seemed to serve as a boundary for responsibility. Dilemmas of disclosure were reported</p> <p>Uptake of testing rate of 57–64% in four families within 12–14 months after disclosure</p> <p>The importance of persuasion in encouraging testing was highlighted, with active and consistent persuasion needed. Family leaders in other areas did not necessarily take the lead in persuading to undergo genetic testing</p>
Segal <i>et al</i> ²¹	Communication to offspring	Quantitative data from 31 questionnaires	BOC	<p>The majority (72.2%) of women disclosed to children without the involvement of a spouse</p>
			Test results	<p>A quarter said there is a difference between disclosing to sons and to daughters, mostly due to the differences in implications for each sex. Slightly less (22.6%) reported no difference (but two of these had only sons).</p> <p>Two-thirds of carriers ($n = 12$) said their children had expressed concerns about their mother's health and their own testing. Older children tended to be more logical, want more facts, and show more concern for mother after disclosure, while younger children expressed more worries and a stronger desire for testing and prevention</p> <p>None reported negative effects on relationships. Further support would have been appreciated by those who disclosed and those who did not</p>
Tercyak <i>et al</i> ³⁴	Communication to children	Quantitative data from 133 adults	HBOC	<p>Greater general distress was found in those who used more active or avoidant coping strategies. The authors suggest that parents' psychological distress may have played a key role in determining their communication behaviour, with those experiencing more general distress being more likely to share</p>
			Test results	<p>The act of sharing with a child did not ameliorate parental distress and few changes in distress were seen. Lazarus and Folkman's transactional theory of coping drawn on to explain findings</p>
Tercyak <i>et al</i> ²⁷	Maternal communication to children (8–17 years old)	Qualitative and quantitative data from 42 mothers: questionnaire with some open-ended questions	BOC	<p>Mothers informed their child because of a belief they had a right to know and/or they felt a strong sense of responsibility (50%)</p>
			Test results	<p>Parents who did or did not disclose reported similar levels of parent–child functioning at follow-up</p>

BOC, breast ovarian cancer; CF, cystic fibrosis; HBOC, hereditary breast ovarian cancer; HD, Huntington disease; HNPCC, hereditary nonpolyposis colorectal cancer.

Table 2 Summary of communication difficulties

<i>Difficulties in communication reported</i>
Information not believed/testing rejected ^{8,20,28}
Failure or difficulty in understanding ²⁶
Failure to convey results ^{4,9,13,14,20}
Speaking about cancer ⁸
Timing ^{4,8,13,14,16,19}
Informing the unsuspecting ⁸
Nonspecific ^{25,26}
Content or knowledge ^{4,13,14,19,28}
Emotional reactions ^{24,37}

is likely to require a different approach but distinct strategies for these different types of communication blocks were not evident in the literature.

Even with improved communication, it seems likely that there will remain relatives with whom the individual is reluctant or unable to communicate. Unwritten rules of family communication dictate who is the most appropriate person to inform certain family members.^{4,17,29,30} Further, family rifts are a common cause of failing to communicate. To attempt to alter existing family dynamics in an endeavour to meet the ends of the genetic services seems to be a dubious undertaking and, in the context of short-term genetic counselling rather than family therapy, possibly unachievable. Rather than valuing the outcomes of communication identified here (eg number of family members informed, knowledge of the recipient), attention may be better focussed on the professional's exploration of existing family dynamics and communication patterns: that is, assessing the process of counselling about family communication rather than these more quantitative or focused results.

Strengths and weaknesses

The strengths and weaknesses of this review arise in two areas: the protocols used to conduct the systematic review and the nature of the literature identified. The review meets most of the AMSTAR criteria,⁴³ although meta-analytic combination of study findings was not possible due to the heterogeneous nature of results, but study characteristics and quality are reported (summarised here and fully available in Edwards *et al*⁵). The search criteria for the overall review focussed on risk communication and also included designated-specific conditions, consequently some relevant papers may have not been identified. However, we sought to overcome this by including papers cited by those studies that were identified.

With respect to the literature reviewed, studies were predominantly on communication about hereditary cancer syndromes and, more specifically, hereditary breast cancer. Differences in communication between families with HD and those with HBOC have been described,^{4,7} and it may be hypothesised that family communication is influenced

by the implications of the condition: the perceived treatability, perceived preventability, and reproductive implications. Similarly, communication about conditions with autosomal-dominant inheritance may not reflect communication about recessive or X-linked conditions. Therefore, conclusions drawn from data on families with familial cancer should only be extrapolated to other genetic conditions with caution.

The studies reviewed may be considered small studies and criticised for lack of representativeness. It is possible, perhaps even likely, that the results represent the views of those at the extremes of communication (very difficult–very easy). Finally, although the review intended to address 'risk communication', none of the studies framed the content of communication as solely 'risk'. It seems reasonable to cautiously assume this relationship, as one of the strongest motivations to communicate test results to family members is the desire to inform them of their risk or options,^{12,25} but this is certainly not the only one and communication may be used as a strategy to cope with bad news or obtain family support.^{25,32,44}

Further research

There is a noticeable lack of examination of cultural, genetic, and gender diversity in these studies. As the implicit rules governing family communication of genetic information are likely to be dependent to an extent on cultural context,⁴⁵ there is a need for studies within other cultural groups. A similar comment may be made about gender, which has been recognised as a factor affecting disclosure and nondisclosure.³ While there are hints in the literature reviewed that gender may affect the process and outcome of communication about genetic information, wider conclusions are hampered by the gender-associated nature of the conditions usually studied (ie HBOC). It would be useful to explore in more depth the experiences of men communicating about conditions such as HNPCC, HD, and CF, as different strategies may be more effective in assisting men to discuss genetic risk. Similarities and differences in the process of communication for genetic conditions with differing implications and inheritance patterns also need to be explored. There would also be value in considering the family communication processes that lead to an understanding of risk, which may occur over a long period of time rather than in the context of conveying a piece of information. The findings of such investigations may be relevant to communication about common complex conditions. We also propose that greater attention needs to be paid to the use of theoretical models of communication to complement empirical studies. The study of communication about genetic information needs to move beyond the simple sender–receiver model of information transfer assumed by most of the studies reviewed, which usually focus on the sender of the information. Rather, understanding needs to be developed

about the meaning of such information to both the individuals concerned, the dynamics between them and also the influences on their relationship. Theories from the family communication literature specifically addressing information sharing within families may prove useful to inform such studies.

Conclusion

The objective of genetic services, implied by the content of research studies on family communication and guidelines in this area,⁴⁶ is to maximise the flow of important information to relevant family members. But the myriad of factors that affect communication means that obligations to family members flow along channels that are not always identical with those through which genes are passed between generations: a sense of responsibility does not always map onto current biomedical models of inheritance.

Family communication is a complex process and the discussions between professional and client about dissemination of information need to recognise and respect this complexity. However, the nature of interactions about genetic information remains poorly understood. Greater attention needs to be paid to the process of communication with family members. This would inform professional discussion and possibly also the development of strategies that assist – but do not compel – clients to communicate with at-risk family members. Concurrently, desirable outcomes (beyond a one-dimensional assessment of the number of relatives informed) should be defined and include those valued by both the person communicating the information and the recipient.

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