

REVIEW

How communication of genetic information within the family is addressed in genetic counselling: a systematic review of research evidence

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Supporting consultands to communicate risk information with their relatives is key to obtaining the full benefits of genetic health care. To understand how health-care professionals address this issue in clinical practice and what interventions are used specifically to assist consultands in their communication of genetic information to appropriate relatives, we conducted a systematic review. Four electronic databases and four subject-specific journals were searched for papers published, in English, between January 1997 and May 2014. Of 2926 papers identified initially, 14 papers met the inclusion criteria for the review and were heterogeneous in design, setting and methods. Thematic data analysis has shown that dissemination of information within families is actively encouraged and supported by professionals. Three overarching themes emerged: (1) direct contact from genetic services: sending letters to relatives of mutation carriers; (2) professionals' encouragement of initially reluctant consultands to share relevant information with at-risk relatives and (3) assisting consultands in communicating genetic information to their at-risk relatives, which included as subthemes (i) psychoeducational guidance and (ii) written information aids. Findings suggest that professionals' practice and interventions are predicated on the need to proactively encourage family communication. We discuss this in the context of what guidance of consultands by professionals might be appropriate, as best practices to facilitate family communication, and of the limits to non-directiveness in genetic counselling.

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INTRODUCTION

Supporting consultands of genetic services to effectively communicate information about genetic risks with their relatives is key to obtaining the full benefits of genetic health care.¹ Genetic health practitioners typically rely on the proband (or, for child probands, the parents) to inform relatives about their potential at-risk status. Passing on such information can be problematic, which may prevent relatives from making informed choices regarding risk management of the disease and life planning decisions.² Individual characteristics and patterns of family behaviour and relationships, disease characteristics and cultural factors may withhold or delay disclosure of genetic information to at-risk relatives, even when consultands see this as their personal responsibility.^{3–7}

Although guidelines recommend that professionals should not contact family members directly, that also state that professionals should actively encourage consultands to transmit relevant risk information to relatives and support them throughout the communication process; however, there is lack of clarity regarding how this should be done.^{8,9} There has been some discussion on how to cascade information about genetic health risks to the relatives of patients with familial hyper-cholesterolaemia, including the active contacting of relatives directly by professionals, although this depends entirely upon information provided by the proband.¹⁰ With genetic diseases increasingly treatable and preventable, some recommend a more proactive role of genetic professionals.^{11,12}

A potential obstacle to professional encouragement of family communication may be too great a respect for the principle of non-directiveness, when understood as the rather unhelpful notion of simply having to give the patient what they ask for.¹³ An adequate notion of non-directiveness puts emphasis on the need sometimes to challenge the statements, attitudes and beliefs of the consultand or patient.^{14,15} Thus, too great an emphasis on respect for the patient's wish not to disclose information to relatives or the relatives' wish not to know may become a barrier to disclosure. This raises ethical issues for health professionals and services, as their degree of responsibility for ensuring relatives' awareness of their risks – and the extent to which they should be proactive in this task – have long been debated in genetic health care.^{16,17}

Previous reviews have been published on the process and outcomes of communication of genetic information within families,^{18–20} including the communication between children and their parents,²¹ the factors influencing patterns of intrafamilial communication and awareness,^{22,23} and on the analysis of the communication between genetic specialists and patients.²⁴ However, to our knowledge there has been no systematic review of studies that have analysed how family communication of information about genetic risks is addressed in genetic counselling practice. This study aims to address that gap.

To inform discussion on the facilitation of family communication about genetics, it is relevant to examine how genetic health-care professionals deal with this issue in clinical practice. This systematic

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review aims to identify and critically reflect on the research evidence available on this topic with the objective of answering the following questions: (1) How and by what means is family communication about genetics approached in genetic counselling practice? (2) What are the actions taken by the professionals involved? (3) What are the characteristics and contents of the interventions specifically assisting consultants in their communication of genetic information to their relatives?.

MATERIALS AND METHODS

This systematic review follows the process developed by the PRISMA statement,²⁵ including the definition of the relevant search terms, selection of studies based on clear inclusion and exclusion criteria and quality appraisal of papers.

Search strategy

The electronic databases PubMed, Academic Search Complete-EBSCO, Scopus, Web of Science and Google Scholar were searched focussing on articles published in English between January 1997 (after direct mutation detection became available and pre-symptomatic testing protocols were generally adopted)²⁶ and May 2014. The search was limited to peer-reviewed journal articles and was performed between January and May 2014. A preliminary search was performed to determine the appropriate search terms; after experimentation, the following terms were defined and searched: *genetic counsel(l)ing OR genetic services AND health professional OR genetic counsel(l) or OR geneticist AND family OR relatives OR kin OR offspring AND communicat* OR disclos* OR transmi* OR shar**.

In addition, the indexes of four relevant journals (Journal of Genetic Counseling, American Journal of Medical Genetics, Clinical Genetics and European Journal of Human Genetics), as well as reference lists and key-author searches were hand-searched to identify additional relevant articles missed by this search strategy. Studies were selected and reviewed based on the inclusion and exclusion criteria (Table 1).

Data evaluation

A total of 2926 papers were initially identified as potential papers for inclusion. Of these, 372 were duplicates, leaving 2554 papers for examination. One additional paper was identified in the hand-search. Based on the titles and abstracts of the remaining papers, 2521 were excluded, leaving a total of 34 papers. The full papers of these studies were obtained. After complete reading by the first and second authors, 20 papers were excluded from the analysis (Figure 1). A total of 14 remaining papers met the inclusion criteria. All these remaining papers were eligible for quality appraisal.

Table 1 Inclusion and exclusion criteria

Inclusion criteria

- Papers documenting qualitative, quantitative or mixed-methods research, including randomised controlled trials;
- Papers including data on how genetic health-care professionals address the issue of family communication of genetic information in genetic counselling practice or other specialist clinical genetics settings (where the paper reported data about genetic counselling practice more broadly, they were included if data related to how family communication about genetics could be extracted from the overall data);
- Papers reporting the implementation of an intervention, support or tool focussed on the facilitation of the communication of information about genetic information in families (where the paper reported data about interventions non-specifically addressing family communication about genetics, they were included if data related to how family communication about genetics could be extracted from the overall data).

Exclusion criteria

- Papers describing clinical or professional guidelines, recommendations for practice, position papers on ethical or legal issues, conference abstracts, editorials or theory-based or commentary papers;
- Papers that do not report analysis of qualitative and/or quantitative data;
- Papers solely documenting patient perceptions on the genetic counselling process or service delivery.

All titles and abstracts of the identified studies were independently assessed by two authors (AM and MP) for inclusion or exclusion in the review. Papers were excluded when both reviewers agreed that inclusion criteria were not met. Disagreements were resolved by discussion. Eligible papers were assessed for their quality using the tool suggested by Kmet *et al.*²⁷ This tool is suitable for evaluation of both quantitative and qualitative studies, using two types of checklists for the studies' appraisal. For each paper, a score between 0 and 2 was assigned against each question ('yes'=2, 'partial'=1, 'no'=0). For studies using mixed-methods design, we decided to score the papers using both qualitative and quantitative checklists. Authors (AM and MP) independently assessed each paper and then met to discuss scores; areas of disagreement were discussed until consensus was reached. As the tool does not specify a cutoff score to discard poor quality papers, we decided to adopt the criteria already followed by Skirton *et al.*,²⁸ where a cutoff point of 60% was defined. The reported score is the mean score of the assessments made by the authors.

Data analysis

Relevant data were extracted from the 14 studies included and were compiled in Table 2 (see Supplementary Material). A meta-analysis of the data would not be feasible given the variability among studies regarding study design, interventions and populations. A thematic analysis was conducted to synthesize the data qualitatively, aiming to identify overarching themes through an iterative process of examining data displayed in the studies to identify themes or patterns.²⁹ Data are presented in narrative form. The analytic process was enabled through ongoing discussion between the authors (AM and MP). Three overarching themes emerged from the analysis of the data: (1) direct contact from genetic services: letters sent to relatives of mutation carriers; (2) professionals' encouragement of initially reluctant consultants to share relevant information with at-risk relatives and (3) assisting consultants in the communication of information to at-risk relatives, which includes subthemes (i) psychoeducational guidance and (ii) written information aids.

RESULTS

Description of the data

Data from the studies included are presented in Table 2. The countries of origin of the 14 studies included in the review are: 6 – United States, 2 – The Netherlands, 2 – Australia, 1 – Sweden, 1 – United Kingdom and 1 – Finland; 1 multisite study, involving both the United Kingdom and Australia. The focus of the studies regarding type of disease was: 6 – hereditary cancers; 5 – combination of adult-onset inherited conditions; 1 – combination of paediatric single gene conditions; 1 – inherited cardiac disease, and 1 – inherited hereditary cholesterol. Regarding methods, studies used: 8 – quantitative methods (5 – survey-based and 3 – clinical intervention studies); 3 – qualitative

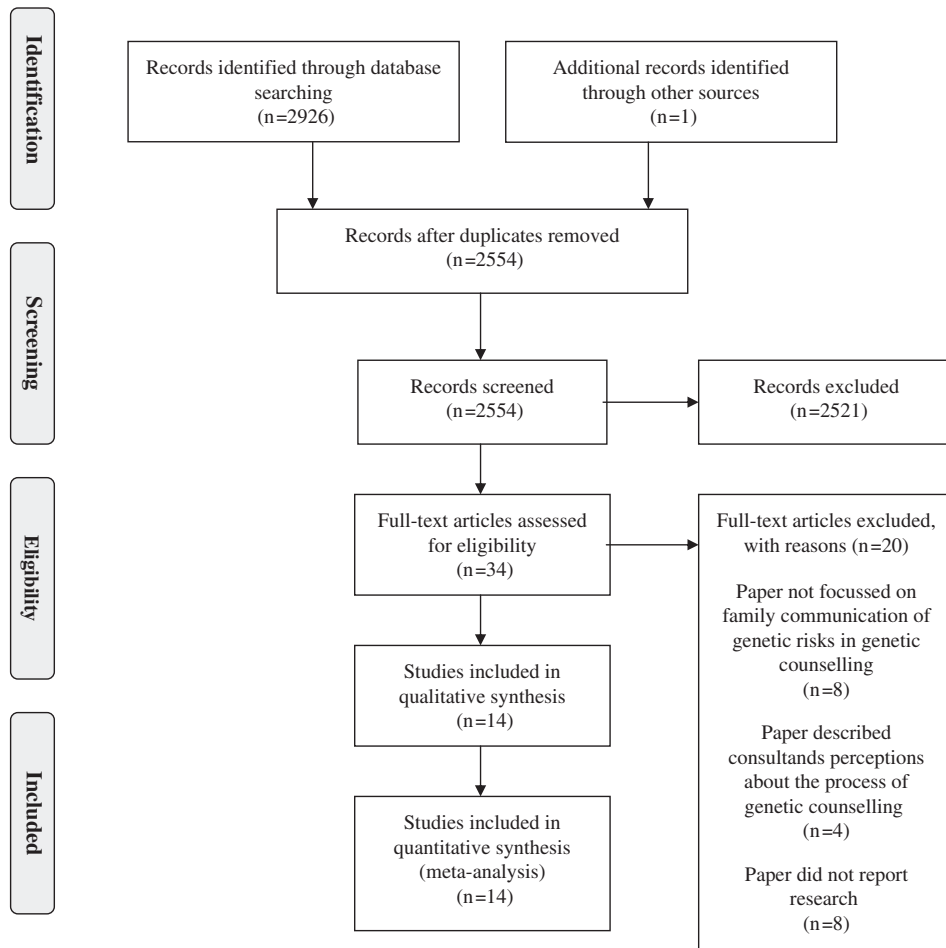


Figure 1 PRISMA 2009 flow diagram.

methods; 2 – mixed-methods; 1 – randomised controlled trial. Sample/participant sizes ranged from 16 to 626 participants. 9 studies were published between 2000 and 2010, and 5 since 2011.

Themes that emerged from the studies

Direct contact from genetic services: letters sent to relatives of mutation carriers. This theme comprises two papers^{30,31} presenting studies in which services/professionals directly contact the family members potentially at-risk. The two papers explore the acceptability and feasibility of direct contact by genetic services to high-risk relatives informing them about the availability of genetic counselling and testing. Direct contact consisted in sending a letter to relatives of mutation-positive consultants. In both studies, the wording of the letters was in general terms and neither identified the proband nor provided details about the disease; the time of sending this letter was decided with the proband. No privacy or autonomy concerns were reported by participants (the recipients of the letter). For those relatives who contacted the genetic services seeking further information, genetic counselling comprised the exploration of the pedigree and discussions about advantages and disadvantages of genetic testing, including possible psychological reactions and employment and insurance issues. Post-test counselling was arranged and mutation carriers were referred for surveillance measures. Results of these studies show high levels of acceptability for genetic services to notify high-risk relatives directly. This was corroborated by the absence of

adverse psychosocial and legal reactions, and the higher uptake of genetic services to clarify their status by at-risk relatives contacted directly than by those informed by family members.

Professionals' encouragement of initially reluctant consultants to share relevant information with at-risk relatives. This theme arose in three papers with studies focussing on active nondisclosure, that is, situations where consultants explicitly refuse to pass relevant risk information to their relatives.^{32–34} Professionals' practice included encouragement of consultants about the appropriateness of sharing relevant risk information with at-risk relatives, but without coercion. In cases of explicit nondisclosure, professionals' tried to persuade consultants to disclose relevant information to relatives, including:³² further discussion with the consultant (to reinforce the importance of disclosure and to clarify the consultants' reasons for nondisclosure), involving more experienced colleagues in the discussions with the consultant, and using written reminders as efforts to encourage disclosure; in no circumstance was coercion reported. Professionals also reported having reflected upon the possibility of notifying at-risk relatives without the consultants' consent,^{33,34} for which colleagues' advice, formal case discussion at a conference presentation and using legal consultants and institutional review boards were the most frequently utilised resources to inform their decision. The actual disclosure of information to relatives without the consultant's

Table 2 Overview of the studies included in the review

Reference	Aims of the study	Design and method	Genetic condition(s)	Key findings	Quality assessment score and quality issues
Aktan-Collan <i>et al.</i> ³⁰	To examine the attitudes of high-risk family members to direct contact by the genetics clinic and the psychosocial responses to genetic risk communication	Cross-sectional survey study. Participants were 286 adults at 50% risk for Lynch syndrome aged 27–90 years, contacted by letter with an enclosed reply letter with a consent form. Of these, 112 participated in counselling and PT in a central hospital in Finland. Descriptive statistics and measures of variability were used for data analysis	Lynch syndrome	- 51% of participants consented to the study; of these, 92% approved the genetic service seeking further information - Genetic counselling for relatives contacting genetic services included: exploration of the pedigree according to the participant's knowledge; discussion of the pros and cons of the uptake of PT, including psychological reactions and employment and insurance issues. Post-test counselling was arranged and mutation carriers were referred for surveillance examinations After identification of pathogenic mutation, the proband gave contact details of adult close relatives. The genetic service sent a letter to each close relative (the timing being decided by the proband). The wording of this letter was in general terms and without identifying the proband or providing details about the cancer risk. If the proband was unwilling to provide contact details but agreed to contact the relative, the letter was given to the proband to pass to the relative. The average proportion of relatives in each family whose genetic status was clarified was 23% in the baseline cohort and 40% in the intervention cohort. 41% of the relatives contacted the genetic service (34% wanting information on genetic testing and 7% wishing no further information about familial cancer risk)	87% (input variables only partially described; conclusions only partially supported by the results)
Suthers <i>et al.</i> ³¹	To compare two methods of informing at-risk relatives about the availability of genetic testing for a familial disorder (1) through the proband and (2) through direct contact of the genetic service	Comparative, quantitative clinical study. Participants were 74 kindreds with pathogenic mutations of familial cancers. Counsellors of a Australian familial cancer clinic were asked to inform their relatives about the availability of genetic testing (baseline cohort); the clinical service directly sent letters to at-risk relatives informing that genetic testing was available (intervention cohort). Outcome measures were the number of relatives whose genetic status was defined. Data analysis included Qui-squared test or two-tailed <i>t</i> -test	HBOC, HNPCC and Cowden syndrome	- Professional actions included taking 'further steps to persuade the consultant to make a disclosure', namely: further discussion with the consultant (to reinforce the importance of disclosure and to clarify the consultants' reasons for nondisclosure), involving more experienced colleagues in the discussions with the consultant, and using written reminders as efforts to encourage disclosure. There was no disclosure of information to relatives without the consultant's consent - Active offers to facilitate disclosure were made, including: giving copies of the summary letter or open letters to consultants, suggesting these to be passed to relatives - 65 Cases of nondisclosure were reported, representing <1% of the clinical workload in the participant institutions - 94% of the respondents 'always' or 'often' encouraged patients to inform their at-risk relatives - Of 119 respondents (46%) having had patients refusing to notify at-risk relatives, 24 (21%) reported that they seriously considered warning those relatives without patient's consent; of these, 1 (4.2%) actually did disclose - Advice from colleagues' and legal consultants and formal case discussion at a conference presentation were used as resources while reflecting upon notifying at-risk relatives	75% (Study's objectives and subjects characteristics not sufficiently described, no random allocation of the subjects, blinding of the subjects and of the investigators not reported)
Clarke <i>et al.</i> ³²	To investigate 'the frequency with which genetic professionals became concerned about the refusal of clients to disclose important genetic information to their relatives, and the actions taken by professionals'	Mixed-methods. Prospective analysis of clinical records. Participants were 12 Regional Genetic Services in the United Kingdom and 2 in Australia. Data were collected during 12 months. Data analysis involved: descriptive statistics and frequencies and thematic analysis	Wide range of conditions, but mainly HD, chromosome translocations and HC syndromes	- 85% (Variance of main results not reported; sampling strategy not described; reflexivity of the account was absent)	
Dugan <i>et al.</i> ³³	To investigate genetic counsellors' clinical experience in warning at-risk relatives and identify the key aspects of their decision-making process	Cross-sectional online survey study. Participants were 259 genetic counsellors (made available to an estimated 1000 members of the US National Society of Genetic Counsellors). Data analysis was not indicated, although descriptive statistics have been used	Familial translocations, HC syndromes, fragile-X, HD and specific genetic syndromes	- 85% (Estimate of variance of the main results not reported; methods and data analysis only partially described)	

Table 2 (Continued)

Reference	Aims of the study	Design and method	Genetic condition(s)	Key findings	Quality assessment score and quality issues
Falk <i>et al.</i> ³⁴	To investigate medical geneticists' clinical experience in warning at-risk relatives and identify the key aspects of their decision-making process	Cross-sectional survey study. Participants were 206 medical geneticists (obtained from 800 members of the American Society of Medical Genetics member lists). Data analysis involved descriptive statistics; group comparisons were made using χ^2 analysis or Fisher's exact test. Statistical analysis was done using SASv8.1 (SAS Institute, Carey, NC, USA)	Familial translocations, HC syndromes, fragile-X, HD, genetic syndromes and late-onset, X-linked, metabolic and cardiac disorders	- 97% of the respondents 'always' or 'often' encouraged patients to inform their at-risk relatives - Of the 123 respondents (60%) having had patients refusing to notify at-risk relatives, 31 (25%) reported that they seriously considered warning those relatives without patient consent; of these, 4 (12.9%) actually did warn relatives without patients' consent - Encounters with colleagues and with legal consultants, as well as seeking expert opinion from an institutional ethics committee were used as resources while reflecting upon notifying at-risk relatives. - In clinics with genetic counselors, the testing implications to other family members were explored. Guidance on sharing information about the condition with children and others was provided, including tailoring content to the children's age and stage of development. - The parent's opinion about how this information should be shared was also taken into account. Also, the need to share information with others (extended family, caregivers, school personnel) was explored, including potentially at-risk family members - Clinical geneticists stated that they instruct consultants to inform their relatives instead of making a direct contact themselves - Reasons given for this practice lie in the so-called 'mores' of clinical genetics, that is, the need to follow a autonomy-based and nondirective approach; the right of the relatives not to know; the lack of a universal wish among relatives to be informed; legal restrictions imposed to geneticists regarding direct contact to relatives; the responsibility of the proband and the welfare of the family and lack of resources	83% (Subject characteristics and input variables only partially described; estimate of variance of the main results not reported)
Gallo <i>et al.</i> ³⁵	To examine health-care professionals' views and strategies for individualizing information sharing when working with families who have a child with a genetic condition	Qualitative descriptive study. Participants were 37 health-care professionals from 3 clinical sites in the United States, representing 10 physicians (3 geneticists), 8 registered nurses, 8 genetic counselors, 5 nutritionists, 4 social workers and 2 advanced practice nurses. Thematic analysis was applied to analyse the data from semistructured interviews	Phenylketonuria, sickle cell disease, cystic fibrosis, neurofibromatosis, haemophilia, thalassaemia, Marfan syndrome and von Willebrand disease	- The parent's opinion about how this information should be shared was also taken into account. Also, the need to share information with others (extended family, caregivers, school personnel) was explored, including potentially at-risk family members - Clinical geneticists stated that they instruct consultants to inform their relatives instead of making a direct contact themselves - Reasons given for this practice lie in the so-called 'mores' of clinical genetics, that is, the need to follow a autonomy-based and nondirective approach; the right of the relatives not to know; the lack of a universal wish among relatives to be informed; legal restrictions imposed to geneticists regarding direct contact to relatives; the responsibility of the proband and the welfare of the family and lack of resources	90% (Reflexivity of the account was not reported)
Stol <i>et al.</i> ³⁶	To explore the attitudes towards informing family members and relevant practices among clinical geneticists	Qualitative, interview-based study. Participants were 9 clinical geneticists from 6 genetic centres in the Netherlands and 7 additional key personnel on the ethical, legal and social aspects of genetics (1 medical sociologist, 2 lawyers, 3 ethicists and 1 philosopher of science). Thematic analysis was used to analyse data	HC syndromes	- The parent's opinion about how this information should be shared was also taken into account. Also, the need to share information with others (extended family, caregivers, school personnel) was explored, including potentially at-risk family members - Clinical geneticists stated that they instruct consultants to inform their relatives instead of making a direct contact themselves - Reasons given for this practice lie in the so-called 'mores' of clinical genetics, that is, the need to follow a autonomy-based and nondirective approach; the right of the relatives not to know; the lack of a universal wish among relatives to be informed; legal restrictions imposed to geneticists regarding direct contact to relatives; the responsibility of the proband and the welfare of the family and lack of resources	62% (Sampling strategy, data collection and data analysis only partially described; no verification of credibility; reflexivity of the account was absent; conclusions only partially supported by the results)
Forrest <i>et al.</i> ³⁷	To investigate genetic health professionals' practice regarding the familial implications of a genetic diagnosis and subsequent family communication	Cross-sectional online survey study. Participants were 626 genetic health professionals recruited internationally through professional bodies and societies (from 796 who initially accessed the survey). The majority of the respondents were from North America (the United States and Canada), Europe and the United Kingdom, and Australasia. Data analysis included descriptive statistics; logistic regression and χ^2 test were used	Disease-scenarios were presented in the survey: HH, FAP, HD and a balanced reciprocal chromosomal translocation	- 98% of the respondents stated they 'consistently' counsel consultants about the familial implication of a genetic diagnosis - During consultations the counseling mainly included: explaining how family members could have also inherited the genetic condition; identification of at-risk relatives and encouragement to communicate with relatives - After consultations, respondents stated: they always send summary letters to consultants (79%); send generic letters for at-risk family members (7% always send and 52% do it sometimes); offering follow-up appointments to support consultants (59%) and giving contact details and information about the genetic condition ('the great majority')	98% (Input variables only partially described)

Table 2 (Continued)

Reference	Aims of the study	Design and method	Genetic condition(s)	Key findings	Quality assessment score and quality issues
Montgomery <i>et al.</i> ³⁸	'To evaluate the efficacy of a communication skills-building intervention to prepare probands to explain their BRCA1/2 test results to first-degree relatives'	RCT. 422 Participants were recruited from the Risk Assessment Program at Fox Chase Cancer Center, US, and randomized; data from 249 participants were analysed (137 control and 112 intervention – 59% of the total of eligible participants). Various psychosocial and communication-based surveys were completed before the disclosure session and after intervention. Primary outcomes were the percentage of probands sharing test results and the level of distress associated with sharing. Data analysis was performed using SAS statistical software	HBOC	<ul style="list-style-type: none"> - Probands shared test results with 80.1% of the relatives - Perceived control and specific social influence were associated with sharing; individuals with higher depression symptoms were less likely to share their test results - There were no significant differences between study groups in the primary outcomes' - Discussions with participants about communication of genetic information to relatives were held in two different moments: during the pre-test genetic counselling session, relatives who could benefit from the information were identified, the communication format to reach them was chosen, and how much relatives already knew and want to know was assessed; after the disclosure session, guidance on an adapted version of the Buckman's 'Breaking Bad News' model (Daly <i>et al</i>, 1995) was provided, including: how to share the test result with relatives, responding to family members' emotional reactions and providing genetic counselling resources to family members. - A resource guide outlining cancer risk factors, family history, the benefits and limitations of genetic testing and a summary of the communication strategy was given to each participant, as well as a copy of the National Action Plan on breast cancer video 'Genetic Testing for Breast Cancer Risk: It's Your Choice' 	95% (Subjects characteristics not sufficiently described)
Roshanai <i>et al.</i> ³⁹	'To investigate the effect of an informational intervention on counselees' knowledge, risk perception, communication of information to at-risk relatives and satisfaction with the service'	Randomized intervention study. Participants were 147 consultands affected by cancer and/or with a family history of cancer and 82 of their relatives. Data on family communication about genetic information were obtained before counselling, and at 2- and 8-month follow-up through a semistructured telephone interview; after the 8-month follow-up, the referred relatives were contacted in writing to answer a questionnaire investigating their psychological distress and aspects about the shared information with relatives	HC syndromes	<ul style="list-style-type: none"> - Level of knowledge about risk in the intervention group increased significantly over time - Correct estimation of personal risk increased significantly in both groups after 2 weeks, but declined at the 8-month follow-up - At the 8-month follow-up, 73% of the consultands in both groups reported that they had informed all their relatives - Consultands in the intervention group were significantly more satisfied with the content of the given information and with the way of informing relatives - During the first genetic counselling session, a clinical geneticist provided information about HC and risks, basic genetics, estimated risks for relatives, genetic testing and surveillance programmes and the importance of communicating this information to at-risk relatives - After the consultation, for the participants in the intervention group, a specialist nurse explained the pedigree and asked consultands to estimate their risk and identify at-risk relatives and explored consultands' intentions to inform at-risk relatives; in cases where they did not plan to share this information, reasons for this were sought and help was provided to try to 	100%

Table 2 (Continued)

Reference	Aims of the study	Design and method	Genetic condition(s)	Key findings	Quality assessment score and quality issues
Forrest <i>et al.</i> ⁴⁰	'To determine whether the provision of additional genetic counselling support could improve the uptake of genetic services by at-risk relatives of probands'	Intervention, cohort-based study. Participants were 150 family members in 19 different kindreds across two cohorts (one comprising patients who were diagnosed with a genetic condition with familial implications and compared with a control cohort) recruited at the Tasmanian Clinical Genetics Service, Australia. Data analysis included descriptive statistics; χ^2 test and logistic regression. Statistical analysis was done using EpiInfo Statistical Program version 3.3.2; centres for Disease Control and Prevention, USA	Balanced reciprocal chromosomal translocation, HBOC, HNPCC, multiple endocrine neoplasia type 1, Peutz-Jehger syndrome or X-linked condition	overcome perceived barriers. Guidance for the disclosure process based on Buckman's 'Breaking bad news' model (Buckman ⁴⁴) was also provided. Consultants were also given aids to be used in the communication with relatives; a pamphlet with basic genetic concepts and information about their type of HC, a videotape of the counselling session and a copy of their medical records and of the pedigree - The proportion of at-risk relatives who contacted the genetic services was 61% in the intervention cohort and 36% in the control group - Female at-risk relatives were 5.9 times more likely to be 'definitely informed' than male at-risk relatives - In the intervention cohort family communication was addressed as follows: before result disclosure, exploration of pedigree and identification of at-risk relatives; at result disclosure, the importance of disclosure to at-risk relatives was stressed and a follow-up letter covering these aspects was sent; after 2-4 weeks post-result disclosure, enquiry, by phone, about which at-risk relatives have been informed and guidance on how to approach uninformed relatives; after 3-6 months post-result disclosure, if all targeted at-risk relatives failed to have contacted the genetic service, probands were contacted by phone exploring reasons for not informing relatives - Also, offer was made to write an informative letter suggesting contact to the genetic service for further information, to be either given to the proband for subsequent delivery to relatives or sent directly to relatives, according to the proband's preference	95% (Randomization of the participants not reported)
Van der Roest <i>et al.</i> ⁴¹	To assess the effects of proband-delivered family letters to their at-risk relatives in terms of referrals to a cardiologist and/or clinical geneticist	Survey-based quantitative study. Two questionnaires were sent to 52 probands who have been given 'family letters' to distribute among their relatives. The first questionnaire focussed the possible delivery of 'family letters' to relatives and possible referrals to health-care professionals. The second questionnaire was on 'non-responding' relatives. All data were analysed using SPSS (version 12.0.2) and differences between groups were tested using Qui-square test	Inherited cardiac disease (inherited arrhythmia and cardiomyopathy)	- In the first visit to the clinic consultants received information about the clinical implications of their disease, its inheritance pattern and risk estimation for their relatives - 'Family letters' were given to consultants, which they were asked to distribute among their relatives - 'Within a mean follow-up of 2 years, the number of relatives actually referred to the clinical geneticist and/or cardiologist was 57% (142 of 249) - From the questionnaire sent to the probands (response rate of 50%), 88% (23/26) were distributed to relatives and for 86% (19/23) probands one or more relatives had been screened - Participants approved the approach for finding carriers, although reluctantly - The packages aided family disclosure by reducing hesitation, but probands only informed first-degree relatives and only communicated with them about risk once	75% (Subjects characteristics not sufficiently described, no random allocation of the subjects, blinding of the subjects and of the investigators was not reported)
Van den Nieuwenhoff <i>et al.</i> ⁴²	'To describe the role of a written information package as support for the identification of carriers of mutations for Inherited High Cholesterol'	Qualitative, interview-based study. Participants were 8 probands and 8 relatives selected from a list of associates. Data collection involved semistructured interviews with participants. Data were analysed	IHC		90% (Reflexivity of the account was absent; sampling strategy only partially described)

Table 2 (Continued)

Reference	Aims of the study	Design and method	Genetic condition(s)	Key findings	Quality assessment score and quality issues
Kardashian <i>et al.</i> ⁴³	To test the acceptability and feasibility of a personalized informational tool to support families in communicating relevant genetic information (ShaRT), and to examine the rates of sharing test results and family testing in relatives and predictors of increased sharing and testing	through content analysis with NVivo 1.3 software and by the 'Plus-minus method'	HBOC	<ul style="list-style-type: none"> - The majority of the relatives reported that the packages were a 'strong cue for action' - The information package consisted of: an introductory letter, a leaflet on how to inform relatives, an informational letter about a screening project to detect people with IHC, an order form to request information packages for relatives and a prepaid envelope - Probands could receive the package through the regular care system (eg, family physician) or on their own request in response to a local public campaign. The package for relatives was similar to the probands', except that a letter for the family physician replaced the leaflet on how to inform relatives - The packages were produced by professionals of patient organization and revised by doctors and patients known by the organization - 'All the women who received ShaRT tool reported that it was a useful resource', while 70% of the participants in the control group reported that additional resources on sharing results would have been useful - No significant differences were found between study groups in terms of the results shared with first-degree relatives; with second-degree relatives sharing was of 38% in the control group and 75% in the intervention group - 'Characteristics associated with increased sharing and testing included: female gender, degree of relationship, frequency of communication and increased knowledge of pedigree' - The tool consisted in a printed binder given in-person to participants during the results disclosure consultation - Materials were developed by physicians and genetic counsellors of the institution where the study was held - Each binder included: the patient's personalized medical information (medical report, BRCA mutation report and recommendations for surveillance and prevention); the family pedigree; letter to family member stating the BRCA mutation identified in relative; fact sheet with frequently asked questions on cancer risk, costs of genetic testing and insurance issues; contact information of genetic counsellors according to each eligible relative; geographic residence; information of support websites and brochures. A CD version of all the documents was also given to participants 	89% (No description of blinding of the investigators and subjects; variance of main results not reported; sample size is small; no verification of credibility; reflexivity of the account was absent)

Abbreviations: FAP, familial adenomatous polyposis coli; HBOC, hereditary breast and ovarian cancer; HC, hereditary cancer; HD, Huntington's disease; HH, hereditary hemochromatosis; HNPCC, hereditary nonpolyposis colorectal cancer; IHC, inherited high cholesterol; PT, predictive testing; RCT, Randomized controlled trial.

permission was reported by professionals as either having not occurred³² or only rarely.^{33,34}

Assisting consultands in the communication of information to at-risk relatives. This theme comprises nine papers with studies focussing on professionals' practice^{35–37} and specific interventions to assist consultands in the communication of information to at-risk relatives.^{38–43} Features of practice included psychoeducational guidance and information aids that were used as adjuncts to clinical practice.

Psychoeducational guidance. These strategies were facilitated by genetic counsellors and by a specialist nurse, and were provided in addition to the standard pre-disclosure genetic counselling sessions, and by phone 2–4 weeks and 3–6 months after result disclosure. Guidance strategies included the provision of supportive and informative elements: face-to-face explanation of the pedigree and risks and asking consultands to estimate their risk and identify at-risk relatives; education about the familial implications and risks of a genetic diagnosis; exploration of the perceived obstacles to passing on information to at-risk relatives; encouragement and 'instruction' to communicate with relatives, and offering follow-up support for consultands; in cases of (non)communication between parents and children or young adults, tailoring the content of the information to the children's age and stage of development, asking the parent's opinion about how information should be shared. Buckman's 'breaking bad news' model⁴⁴ was used as a communication skills-building intervention to educate and assist consultands in the disclosure process, including the exploration of the relatives' level of knowledge about their at-risk status, their willingness to know about it and responding to the relatives' reactions.

Written information aids. Various information aids were used, either given to consultands at the consultation or sent to their homes according to the proband's preference. Materials were produced by professionals from patient organizations and revised by doctors and patients known by the organization, or by physicians and genetic counsellors from the clinical institutions. Tools mainly consisted of letters, leaflets/brochures or resource guides. Materials were also made available through packages/binders integrating separate components containing the following information: clinical implications about the specific disease; recommendations for surveillance and prevention; the importance of family history; the benefits and limitations of testing; guidance on how to inform relatives; a letter to family members stating that a disease-causing mutation was identified in their relative; a fact sheet with frequently asked questions about disease risk, costs of genetic testing and insurance issues; contact information for genetic counsellors specific to each eligible relative according to their geographic residence; information about support websites and a CD version of all the documents provided. Personalized medical information was also made available, for example, including copies of the pedigree and medical reports, and videotapes from the initial genetic counselling session.

DISCUSSION

This review summarises the findings of 14 studies related to the practice of health professionals in clinical genetics and genetic counselling services on the issue of communication of genetic information within families. Although studies were heterogeneous in design and setting, the findings of this review identified three overarching themes representing how family communication about genetics is addressed in practice.

The dissemination of information within families has been shown to be actively encouraged and supported in genetic counselling professionals, following the published guidelines and recommendations from various professional bodies.⁸ In cases of active nondisclosure, a policy of active encouragement and persuasion characterises the professionals' role and only very rarely do professionals override their patients' confidentiality. This suggests that consultands are commonly willing to disclose genetic information to their family members, even though difficulties may be felt on how to actually communicate rather than on deciding whether they wish to inform them. For those requiring support or showing difficulties in this process, there is psychoeducational guidance, and written information aids are available as means through which professionals can assist their consultands in the communication process. These interventions were generally effective 'cues for action' both in terms of intrafamilial disclosure of genetic information and of genetic testing uptake among at-risk relatives. There is also a more direct approach to family communication, whereby genetics services send informative letters to at-risk relatives informing them of their risks and the availability of genetic counselling services for further information. This approach was fully acceptable to the relatives, and effective in promoting further clarification of at-risk relatives' genetic status.

Most of the practices reviewed were part of research interventions rather than describing the relevant routine practices among genetic health professionals. The studies have described the various ways of addressing the issue of family communication about genetics in practice, ranging from more process-focused approaches (such as direct contact) to others that privilege the provision of specific guidance, but they include few details of the actions taken by professionals and of their strategies for facilitating family communication. Most of the interventions used to support patients to communicate genetic information to their relatives focused on information content (as in the 'deficit' model of the public understanding of science) and were delivered as a single transaction with consultands, whereas in only two cases did follow-up contact occur.

Some of the intervention studies in this review are predicated on the need to encourage family communication, and that it would be in the interests of other family members for the consultand to disclose information about him/herself because their relatives may find it helpful to know about their risk of developing a genetic disorder. The issue of family communication in genetic health care may be regarded as an area where a strict adherence to a narrow, 'shallow' conception of non-directiveness may be inappropriate, and where the explicit confrontation of the consultands with information or with a call to consider the potential outcomes of a range of hypothetical scenarios, in which they have made different decisions, could be seen as 'appropriate directiveness'.¹⁵

Family communication depends on a plethora of factors that go beyond consultands' motivations and the pre-determined actions of professionals within genetic counselling. Furthermore, measures of the number of relatives contacting genetic services or the uptake of testing are not necessarily indicative of communication within families. Recent counselling-oriented interventions aimed at facilitating family communication about genetic information are currently being implemented, aiming at fostering patients' autonomy and their self-efficacy towards making informed decisions.^{45–47} Although it may be unhelpful to impose an operational definition of what addressing family communication about genetics in practice should be, without such a definition the question of what guidance of consultands by professionals might be appropriate remains unclear. Whereas an accurate understanding of information is key for the appropriate

communication with relatives and the consultand's autonomous decision-making (nondirective), counselling on this specific topic does not mean just presenting full and unbiased information as something that can be passed, unaltered, from person to person in the family; this is especially salient in families where parents are considering whether and how to communicate genetic information to their young adult children. As others have argued,^{1,18} family communication about genetics is a multistage process that requires genetic information to be understood beyond simplistic models of communication. The voicing by both consultand and professionals of their perspectives and some dialogue about the differences has been proposed as a helpful way for professionals to consider when it may be unhelpful and inappropriate for them to challenge the consultand and where, in contrast, it may be very useful for them to contribute more proactively with their perspective, without coercion and without denying the critical importance of the patient's wider value systems.¹³

Implications for practice and further research perspectives

Perhaps rather than focussing on the content of the information to be given in genetic consultations, engaging with the consultand in a reflective consideration of the relevance and value of transmitting risk information to those relatives identified as at potentially high risk, and exploring the family dynamics and patterns of communication, as well as the possible nature and causes of poor communication within families, may provide more targeted support and facilitate more open communication within the family.^{38,46} The complexity of family functioning perhaps makes it more fruitful and more ethical for professionals to be open to address these issues using more contextual strategies.¹⁸ The family genetic risk communication framework⁴⁸ may be of help in the clinical context as an orienting tool to work with consultands through their communication process. Clinical genetic services and the health-care system more broadly might also want to consider the specificities of the communication throughout the family's life cycle,⁴⁹ namely between parents and children/young adults,⁵⁰ both in the context of genetic counselling practice and in the facilitation of family-oriented psychosocial support.^{51,52} Future research including process studies and observational studies could contribute to the better understanding of how family communication about genetics is specifically addressed in genetic counselling, namely the actual strategies and responses used by genetic health professionals. Further research should also discuss these data with genetic health-care professionals and with members of families affected by inherited conditions, aiming to look at the advantages and pitfalls of these approaches.

Limitations

Some limitations need to be considered within this review. Given that in this review, only English language studies published in scholarly peer-reviewed journals were included, unpublished data or data published by other means or in other languages that could have contributed to a better understanding of this topic were not analysed. These biases therefore regrettably privilege those studies conducted in countries where the English language is well established and where research funding is available and this impedes access to different cultural and socioeconomic research contexts.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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