Familial Cancer (2005) 4: 163–167
DOI: 10.1007/s10689-004-7992-1

Informing one's family about genetic testing for hereditary non-polyposis colorectal cancer (HNPCC): a retrospective exploratory study*

Ilse Mesters[1], Marlein Ausems[1], Sophie Eichhorn[1] and Hans Vasen[2]
[1]*Department of Health Education and Health Promotion, University Maastricht, Maastricht, The Netherlands;* [2]*Director Netherlands Foundation for the Detection of Hereditary Tumours (STOET), Leiden University, Leiden, The Netherlands*

Received 18 June 2004; accepted in revised form 13 December 2004

Key words: colon cancer, communication, disclosure, family, genetic testing, hereditary, HNPCC, psychosocial

Abstract

Background: The family-link approach of case finding is considered the fastest and most efficient approach to trace people with hereditary disease. Therefore, there is a need to understand if, why, and how people with hereditary non-polyposis colorectal cancer (HNPCC) inform their biological family. *Aim*: To explore people's perspective on informing one's biological family regarding a hereditary predisposition for HNPCC. *Method*: In-depth interviews were conducted with 30 people recruited from the database of the Netherlands Foundation for Detection of Hereditary Tumours (STOET). Interviews were transcribed and analyzed thematically. *Findings*: Disclosure was stimulated if people felt morally obliged to do so or when they anticipated regret if something happened because it is preventable. Motivation to disclose seemed to increase if there were, especially fatal, cancer cases in the family. Presence of external cues (e.g. professionals) appeared important for disclosure as well. Disrupted and tense family relations were reasons not to disclose, as well as young age of the message recipients and negative experiences at their first attempt to disclose (a novel finding). Disclosure was merely restricted to the nuclear family. A personal approach in this respect was preferred. With respect to content of the disclosure, participants reported to solely announce the presence of the hereditary defect and the possibility of testing. It was mostly considered the recipients' responsibility and own choice to obtain further (technical/medical) information.

Introduction

In 5–10% of the colon cancer cases a hereditary defect is of overriding importance in the disease development [1]. Different varieties of hereditary colon cancer can be distinguished. The underlying study focuses on people with hereditary non-polyposis colorectal cancer (HNPCC). HNPCC is an autosomal dominant disorder. HNPCC gene mutation carriers are estimated to have an 80% lifetime risk of developing colorectal cancer and are advised to follow high-risk surveillance guidelines [2]. Since HNPCC is hereditary, news about a positive test result of one person becomes automatically crucial for the remainder of the biological family. Current standards of practice rely on the diagnosed family member or index person (IP) to inform the remainder of the family in question. This 'family-link' approach of case finding is considered as the faster and more efficient approach to trace people with hereditary disease compared to case finding in the general population. It has become customary to provide a summary letter to the counselee at the end of the genetic counseling describing aspects of the client's medical history, family history, and genetic circumstances. Such a summary letter contains the facts that can be communicated to family members. In the Netherlands, in case mutations have been found, an additional family letter can be given to the counselee for distribution among family members. What clients do with these letters remains unknown. Literature, however, indicates that the problem of

*The study presented was performed at the Care and Public Health Research Institute (Caphri), which participates in the Netherlands School of Primary Care research (CaRe), acknowledged in 1995 by the Royal Dutch Academy of Science (KNAW).

Correspondence to: Dr I. Mesters, University Maastricht, Department of Health Education and Health Promotion, P.O. Box 616, 6200 MD Maastricht, The Netherlands. Tel: + 31-43-3882227; Fax: + 31-43-3671032; E-mail: i.mesters@gvo.unimaas.nl

complete or partial nondisclosure might be substantial [e.g. 3–6]. Reasons for nondisclosure consist of health providers not informing tested people about the necessity to do so [7], or tested carriers' misunderstanding of this information [e.g. 8]. But, also when potential actors for disclosure are successfully informed, a proportion of them will decide not to inform family members. This is striking, since research has shown that a major motivation for testing for people is to help assess risk to family members [e.g. 9]. Perceptions of high-risk individuals regarding informing their family about the hereditary disease risk are under explored. This study investigates people's perspective regarding informing one's biological family on the hereditary predisposition for HNPCC.

Methods

Study design

Because of the delicacy of the subject, it was anticipated that people may be rather reluctant about, and may find it difficult, exposing their deepest thoughts and feelings on hereditary issues and family disclosure. Therefore, a qualitative research design was chosen using one-hour semi-structured interviews, during which the interviewer used a list of topics that she planned to discuss. Important research themes were: reasons to inform family members or not, the process of information provision (own response and responses of family members to the news, coping of the family with the news and best practices in this respect, and the persons who took responsibility for informing other family members), and the extent to which the family was informed.

Recruitment

The aim was to interview 30 persons. All individuals were recruited from the registration database of the Netherlands Foundation for the Detection of Hereditary Tumours (STOET). This organization aims at the nationwide promotion and coordination of periodical medical check-ups of family members who have an increased risk for cancer because of their hereditary predisposition. An employee of the STOET randomly selected 44 persons to be contacted by telephone to explore their willingness to participate and to assure confidentially of personal particulars. From all participating interviewees written informed consent was obtained. The interview took place at participants' homes throughout the Netherlands.

Data analyses

All interviews were audio taped on the basis of which a summary was made of each interview. The data were first segmented per main interview question (*a priori* codes). Next, the researcher developed an initial master list of codes (based on the first three interviews) while examining the data using descriptive words (inductive codes) to identify sub-segments. This master list was reapplied to the remaining interviews and adjusted and reapplied to the data in case a new segment was encountered. No attempt was made to 'quantify' the data. Findings from the analysis also underwent expert review by other study investigators to check for researcher biases. The qualitative data are presented as descriptive summaries and interpretations of the key themes, supported and illustrated by quotes from the raw data.

Results

Population

Thirty-eight of the selected 44 persons were contacted by phone (after an average five contact attempts) to recruit 30 persons for the interview. Six persons declined to participate (four women aged between 49 and 61 years and two men aged 55 and 67 years) because they were too busy, considered it too much fuss, or were fed up with research studies. Two interviews were cancelled. One due to unexpected family circumstances and one due to language difficulties. Of the remaining 30 people, both males ($n = 8$, 27%) and females ($n = 22$, 73%) participated. The average age was 53 years (SD = 12 years, range 25–69 years). Eleven respondents (37%) had a confirmed carrier status.

Reasons to inform one's family

To the question why people did inform their family members about HNCCP moral obligation and anticipated regret were frequently mentioned. As an interviewee stated: 'I did feel responsible for the remainder of the family. At least you can do something about it. I sure would feel guilty if cancer would be diagnosed with a family member and this person was not being screened because he was unfamiliar with this risk. I would reproach myself very much.' Additionally respondents talked about the premonitory role of the Clinical Hereditary Centers and the STOET. If these organizations verbally stressed that informing one's family is crucial, this appeared an important stimulus to get into action. As illustrated by one of the participants 'My sister's physician underlined the importance, so we did it together. It felt like doing the right thing.' Thus, although there was an intrinsic motivation to disclose information to one's family, it helped if external cues were present that gave people the feeling that what they were doing was okay. If people were cancer patients themselves they felt a real urge to warn others, but also the family history with respect to cancer cases appeared strongly related to informing one's family. It seemed that if relatives that had been diagnosed with colon cancer and, maybe even more essential, had died

because of colon cancer (respondent: 'It should not happen again,' 'I have seen my mother dying, this I need to prevent'), perceived susceptibility of the person himself and others appeared enlarged and *vice versa*.

There were also circumstances under which information provision seemed to be postponed or not intended to be carried out. If children were considered 'too' young, they were less likely to be informed. 'I did not need to notify any one. My children are too young to understand. This question does not apply to me.' If the carrier's opinion conflicted with the opinion of the partner, their offspring/family was less likely to be informed, or if hereditary knowledge already created conflict in the nuclear family the remainder of the biological family appeared less likely to be informed, 'My daughter got tested, but she never went back for the test results. This created tension in our family, so we did not take it any further. My brother and his family have no idea.' Family members with whom carriers were on bad terms were also less likely to be notified about the potential hereditary predisposition.

Coverage of family members receiving hereditary information

Interviewees were asked from which persons they received the news and which persons they continued to inform about the news. In general, care providers informed interviewees if they were the index patients or participants had been informed by a first degree relative. In a single case more distant family members informed people. Interesting was that all interviewees responded that all family members that ought to receive the information had received it. However, persons had differing definitions regarding the 'family members that ought to be informed.' In many cases this would be the nuclear family only: 'I only told my husband and children' and 'I did not inform others, I heard it from my sister, I myself have no children, so I am the terminal station.'

Apparently in some cases it took years before all relevant persons were acquainted with the news because family members from current generations were unaware that ancestors had died from cancer. Sometimes parents withheld news about the familial predisposition from their offspring because at that time they were too young and 'tomorrow never came,' until one of the children got cancer and the news got out at last.

Process

From the transcripts, it became clear that informing one's family about hereditary cancer was mostly executed as pure and simple notifying family members about the hereditary danger and the possibility to get a hereditary test. There seemed to be a tendency to rely on medical care/experts for more technical information 'Physicians should subsequently provide the information about how and what' or 'Written and verbal information from the STOET is important to get information.' Regarding the process of information giving it turned out that a disappointing contact in the early dissemination process concerning the hereditary information seemed to increase the chance that the IP would give up. As an interviewee stated: 'After initial friction in my own family I reconsidered informing my brother ... You never know maybe I would upset them needless and cause only tumult.' If persons already had had a few 'good' conversations, they appeared better able to handle a disappointing one. So the previous good meetings seemed to outweigh the impact of a bad meeting.

Interviewees also differed with respect to who notified the remainder of the biological family. Five variations were reported. Some index patients done it on their own, others approached family members together with either a partner or another relative, a family member (other than the index patient) informed the family, some choose a domino approach (the responsibility was passed on to the person that just was informed and so on) or it just happened (without a predetermined plan). The latter variant was mentioned least often, equal numbers represented the others. A disadvantage of a no-predetermined dissemination plan was that the knowledge about who was informed appeared more based on assumptions than on actual knowledge.

Several strategies were used to notify one's family. Some people 'just phoned them all,' others paid their family a personal visit and another strategy consisted of mailing a letter drawn up by the STOET. Also combinations of these strategies were reported. When asking how people experienced informing their family some of them indicated that it had been difficult. In some cases family members were shocked by the (mostly unexpected) news, got angry and took it out on the messenger. As a respondent said, 'In the beginning I had the feeling to be up against this on my own. I was blamed for the situation. I have thought at times I quit, I am not going to do this anymore.' As time went by, their relatives appeared to have gotten used to the situation and apparently learned to cope with their feelings. In families where openness about cause of deaths dominated, people had grown up with the presence of colon cancer so they already sensed something could be wrong, it was only a confirmation of an assumption. Under these circumstances news easily and quickly spread throughout the family, 'It wasn't unexpected really.' These family members were more likely to be interested in the news and to sympathize with the messenger.

Discussion

Several reasons were mentioned regarding why people did notify their family. Anticipated regret and moral obligation were the ones often mentioned at the opening of the interview. The motivation of participants that

worse could be prevented was also found in a study by Wilcke [10] as a reason to inform their family about a hereditary predisposition. McConkie-Rosell et al. [11] and Green et al. [12] also revealed that index patients felt obliged to inform one's family about hereditary issues. This revealed attitude is in line with the general public's opinion as well [13]. If the messenger himself had cancer, if the prevalence of colon cancer in the biological family was high or colon cancer had resulted in the death of a family member, these all were reasons that stimulated disclosure. Julian-Reynier et al. [14] found that those being affected by (hereditary) breast cancer intended to disclose this information twice more frequently than healthy subjects. Not found in the literature, but mentioned several times in the interviews, was that people stressed that if medical care providers or significant others urged mutation carriers to inform their family, they seemed much more motivated to do so.

In our study disrupted/suboptimal family relations were reasons given for not informing other family members, a finding confirmed by other studies as well [2, 4, 12, 15–18]. In the latter three studies this reason was mentioned by about 9–15% of the study participants. Furthermore, if interviewees anticipated rejection of their message by a biological family member, the reluctance to inform that relative increased, a factor also confirmed by Koehly et al. [3] and Costales et al. [15]. Young children seemed to have a higher chance of not being informed as revealed in our study, but also in a study by Tercyak et al. [4].

At the moment little knowledge is available on how information about hereditary cancer is communicated in families. Our study (with one exception) revealed a tendency among participants to discuss hereditary information within the nuclear family only, thus first-degree relatives and partners. This finding is in line with studies conducted by Fanos and Johnson [16] Koehly et al. [3], Julian-Reynier [14] and Peterson [19]. Often it is not regarded a responsibility to inform cousins since that is considered the task of the cousins' parents. Peterson [19], however, indicated that if parents of cousins have passed away there is a higher chances that a second-degree family member will inform them. Furthermore, it turned out that the information dissemination process could sometimes take years. A novel observation of this study was that initial failure to inform family members discouraged people to continue, while apparently early successes seemed to buffer people in this respect. This finding implies that coaching messengers to increase the changes of initial success might be indicated. If the message was more or less expected, and therefore was not dissonant from the existing expectations [20], the hereditary information was more welcomed then when the news was rather unexpected.

Present study revealed different ways in which people inform their family, for instance in writing or 'face to face.' Also different actors seemed to play the messenger role in the family communication process, for instance the index patients, the partner, a specific family member or combinations of these. And, although the 'nuclear family paradigm' is not the only type of family relevant in genetic testing, there appeared to be a tendency to restrict information giving to this microculture. Acknowledging that families should be allowed to set their own rules to discuss genetic information, further research might focus on: ways to broaden the counselee's concept of the nuclear family to more disparate family forms; revealing how written information (patient and/or family letter) is being used for family communication by receivers of this information; revealing more and less helpful approaches (skills) to verbal family communication on genetic issues, since many people opt for, at least, a 'face to face' approach, and alternative ways of approaching family members [21], with consent of the counselee, since some people in our study would like to leave this task to health professionals.

Limitations of this study include that findings might be affected by recall bias since the interviews focused on prior experiences about HNPCC disclosure.

Practice implications

The study revealed that messengers of hereditary news in general consent to notify the nuclear family. If it is required or desirable to share hereditary information to other family members this appeared not to be obvious to index patients at this moment. Index patient's concept of their 'family' should be defined during the genetic counseling process, and messengers should be more clearly instructed concerning their informing task.

Stressing the importance of disclose, especially by health processionals seemed to motivate messengers to get into action and served as a justification to do so. Therefore health care professionals should be made aware of their own influence. Furthermore, messengers should be prepared to deal with unpleasant conversations since this seemed to discourage them from continuing to try sharing the information, especially when this happened in the early stage of the information dissemination process. Starting with the 'easiest' family member, planning on how to respond in such a negative situation and/or asking someone for help and support might be indicated to increase the chances of initial success, as well as some coaching from professionals working at clinical genetic centers. Recent more systematic initiatives to build communication skills of messengers in this respect have been developed by, for instance, by Daly et al. [22] called the Six-Step Strategy to better communication (including a six-step protocol for messengers to follow and a genetic resource handbook) and the hereditary high cholesterol educational package for index patients (including a leaflet with suggestions for family communication and an additional package for blood relatives), which can be requested from

an association for patients with hereditary cardiovascular disease [http://www.bloedlink.nl/index.php?structure_id = 779(30-11-2004)]. Both interventions address the issues who to tell, what to tell and how to tell it and provide reference materials or literature.

Finally, messengers feel obliged to inform others about the existence of the hereditary risk and the possibility of counseling and testing. However, they distance themselves from persuading family members to get tested because of privacy and autonomy reasons. If we rely on messengers to motivate others to seek hereditary counseling and testing it might be essential that messengers explicitly consent to this task and subsequently receive guidance and advise on how to proceed and over a longer period of time than just the pre- post-test counseling period.

Acknowledgements

We are grateful to Marianne Jutte, Mary Velthuizen and Alice Donselaar from the STOET for contacting the target population and we express special gratitude to all the people who agreed to participate in this study.

References

1. Lynch HT, Smyrk T, Lynch J et al. Update on the differential diagnosis, surveillance and management of hereditary non-polyposis colorectal cancer. Eur J Cancer 1995; 31a: 1039–46.
2. Burke W, Petersen G, Lynch P et al. Recommendations for follow-up care of individuals with an inherited predisposition for cancer: I. Hereditary non-polyposis colon cancer – Cancer Hereditary Studies Consortium. J Am Med Assoc 1997; 277: 915–19.
3. Koehly LM, Peterson SK, Watts BG et al. A social network analysis of communication about hereditary nonpolyposis colorectal cancer hereditary testing and family functioning. Cancer Epid Biom Prev 2003; 12: 304–13.
4. Tercyak KP, Peshkin BN, DeMarco TA, Brogan BM. Parent-child factors and their effect on communicating BRCA ½ test results to children. Pat Educ Couns 2002; 47: 145–53.
5. Lerman C. Parent-child factors and their effect on communicating BRCA ½ test results to children. Pat Educ Couns 2002; 47: 145–53.
6. Claes E. Communication with close and distant relatives in the context of hereditary testing for hereditary breast and ovarian cancer in cancer patients. Am J Med Genet 2003; 116A: 11–19.
7. Lynch HT, Foillet KL, Lynch PM, et al. Family history in a oncology clinic: Implications for cancer hereditary JAMA 1979; 242: 1268–72.
8. McAllister M. Personal theories of inheritance, coping strategies, risk perception, and engagement in hereditary non-polyposis colon cancer families offered hereditary testing. Clin Genet 2003; 64: 179–89.
9. Meiser B. et al. Attitudes to hereditary testing for breast cancer susceptibility in women with increased risk of developing breast cancer. J Med Genet 2000; 37: 472–6.
10. Wilcke JTR. Transmitting hereditary risk information in families: Attitudes about disclosing the identity of relatives. Am J Hum Genet 1999; 65: 902–9.
11. McConkie-Rosell A, Robinson H, Wake S, et al. Dissemination of hereditary risk information to relatives in the fragile X syndrome: Guidelines for hereditary counselors. Am J Med Genet 1995; 59: 426–30.
12. Green J, Richards M, Murton F et al. Family communication and hereditary counselling: The case of hereditary breast and ovarian cancer. J Genet Counsel 1997; 6: 45–60.
13. Lehmann LS, Weeks JC, Klar N et al. Disclosure of familial hereditary information: Perceptions of the duty to inform. Am J Med 2000; 109: 705–11.
14. Julian-Reynier C, Eisinger F, Chabal F et al. Disclosure to the family of breast/ovarian cancer hereditary test results: Patient's willingness and associated factors. Am J Med Genet 2000; 94: 13–8.
15. Costales JW, Itzen M, Malick et al. Communication of BRCA1 and BRCA2 results to at risk relatives: A cancer risk assessment program's experience. Am J Med Genet 2003; 119C: 11–8.
16. Fanos JH, Johnson JP. Barriers to carrier testing for adult cystic fibrosis sibs: The importance of not knowing. Am J Med Genet 1995; 59: 85–91.
17. Pasacreta JV, Jacobs L, Cataldo JK Hereditary testing for breast and ovarian cancer risk: The psychological issues. AJN 2002; 102: 40–7.
18. Julian-Reynier C, Eisinger F, Chabal F et al. Disclosure to the family of breast/ovarian cancer generic test results: Patient's willingness and associated factors. Am J Med Genet 2000; 94: 13–8.
19. Peterson SK, Watts BG, Koehly LM et al. How families communicate about HNPCC hereditary testing: Findings from a qualitative study. Am J Med Genet 2003; 119C: 78–86.
20. Patty RE, Wegener DT. Attitude change: Multiple roles for persuasion variables. In Gilbert DT, Fiske ST, Lindzey S (eds): The Handbook of Social Psychology. Boston: McGraw-Hill 1998; 323–90.
21. Sermijn E, Goelen G, Teugels E et al. The impact of proband mediated information dissemination in families with a *BRCA1/2* gene mutation. J Med Genet 2004; 41: e23.
22. Daly BM, Barsevick A, Miller MS et al. Communicating genetic test results to the family: A sx step, skills building strategy. Fam Community Health 200; 24: 13–26.