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Understanding Patterns of Health Communication in Families At Risk For Hereditary Non-Polyposis Colorectal Cancer: Examining the Effect of Conclusive vs. Indeterminate Genetic Test Results

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Abstract

In families meeting criteria for hereditary non-polyposis colorectal cancer (HNPCC), genetic testing may or may not identify a mutation. Communication about genetic testing and risk in families with identified HNPCC mutations is associated with individual and relational factors. Similar communication patterns would be expected in families with similar clinical and pathological characteristics, but without an identified HNPCC mutation; however, previous studies have not included such families. Social network analysis was used to compare communication networks and associated individual and relational factors in families with and without identified HNPCC mutations. Respondents from families without identified mutations communicated about genetic counseling and testing and risk for HNPCC with a significantly smaller proportion of network members, compared to respondents from mutation-positive families. Members of families without identified mutations were also more likely to share thoughts about risk for HNPCC with network members whose advice they take, compared to members of families with known mutations. These findings extend our knowledge of communication in families at risk of HNPCC to include the many families in which a causative mutation has not yet been identified. Differences in the breadth of communication about genetics and risk for HNPCC, and the possibility that members of families without identified mutations may seek advice from those with whom they communicate about risk, provide new avenues for future research. Understanding existing communication patterns could help improve education and counseling processes, and facilitate the development of interventions designed to assist in family discussions of risk.

Introduction

In families at risk of or affected by hereditary conditions, family communication is an important mechanism by which individuals learn of their risk for disease (Carlsson & Nilbert, 2007; Mesters, Ausems, Eichhorn, & Vasen, 2005; Peterson, 2005). Health care providers do not disclose genetic risk information to family members who are not their clients (The American Society of Human Genetics Social Issues Subcommittee on Familial Disclosure, 1998). Thus, individuals who have consulted with professionals and received

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genetic risk information are responsible for disclosing it and discussing its implications with family members.

Multiple members of families with hereditary cancer syndromes may be at risk, providing one context for examining communication about genetics and risk. Hereditary non-polyposis colorectal cancer (HNPCC) increases risks for several cancers, the most prominent of which are colorectal (CRC), endometrial, gastric and ovarian cancers (Barrow et al., 2009). Awareness of risk is crucial; lifetime risks for CRC may reach 69% for men and 52% for women in the absence of cancer screening (Quehenberger, Vasen, & van Houwelingen, 2005). Colonoscopy screening starting early, and occurring every 3 years, more than halves the risk of CRC, prevents CRC deaths, and decreases mortality by about 65% in HNPCC families (Jarvinen et al., 2000).

Genetic testing for HNPCC begins with an individual diagnosed with an associated cancer (the *index case*). Genetic testing of the index case can detect a deleterious mutation (mutation-positive results), a variant of uncertain significance (equivocal results), or fail to identify a mutation (indeterminate results). When a mutation is identified in the index case, other at-risk family members can pursue testing for that mutation. Those who carry the mutation have significantly increased cancer risks and should pursue recommended cancer screening. Family members who do not carry the family mutation are spared early and frequent cancer screening. Family members of index cases with indeterminate genetic test results would not benefit from genetic testing, as no mutation has been identified for which they can be tested. They are encouraged to follow the screening guidelines for confirmed mutation carriers until more definitive genetic testing clarifies their cancer risks (Lindor et al., 2006). Families suspected of having HNPCC tend to have multiple generations in which a member has been diagnosed with HNPCC-associated cancers. The primary difference between these families is identification of a deleterious mutation, and the availability of genetic testing for at-risk family members.

Previous studies examined communication in families with known HNPCC mutations. In these families, communication occurs in the context of individual factors (e.g., gender, age, risk status) and relationship characteristics (e.g., provision of advice and support, perceived closeness, kinship tie) (Carlsson & Nilbert, 2007; Koehly et al., 2003; Mesters et al., 2005; Peterson, 2005; Peterson et al., 2003; Stoffel et al., 2008; Wilson et al., 2004). This is consistent with the Family Systems Genetic Illness model (Rolland & Williams, 2005), in which characteristics of the family system, or family social network, and the illness interact to create a unique climate for family communication and response to risk for disease.

Communication about genetics and risk for HNPCC may be particularly important for families without identified HNPCC mutations. It is less likely that members of these families will receive education, counseling and cancer screening recommendations from genetic health care providers. They may rely on the index case to share genetic risk information with other at-risk family members. Index cases with indeterminate genetic test results are less likely than mutation-positive index cases to share test results with family members (Stoffel et al., 2008). And, at-risk individuals who do not discuss risk with family members are less likely to engage in recommended cancer surveillance (Ersig, Williams, Hadley, & Koehly, 2009).

Given the extensive similarities between families with and without identified HNPCC mutations, it seems likely that patterns of communication about genetics and risk would be similar. However, previous studies have not compared communication in families with and without identified HNPCC mutations. The purpose of this analysis was to examine the association of selected individual and relational characteristics with communication about

HNPCC, and compare these associations between families with and without identified mutations.

Methods

Methods & Framework

Social network analysis (SNA) was used to examine communication patterns and the association of selected individual and relational characteristics with communication about genetics and risk for HNPCC. SNA focuses on relationships among individuals (Wasserman & Faust, 1994). This study is based on a framework that builds on the premise that social networks are essential to health and well-being (Berkman & Glass, 2000; Berkman, Glass, Brissette, & Seeman, 2000). Social networks function through psychosocial mechanisms, including provision of informational support via interpersonal communication (Berkman & Glass, 2000). These exchanges can influence health in the face of risk for disease. Exploring informational support and communication in families at risk for HNPCC may provide insight into the health behaviors and health status of those at risk (Berkman & Glass, 2000). The network of interest for this study was the family system, which included all people who were considered to be family, and the social resources they provide. The relations of interest were those within which communication about genetic counseling, genetic testing, and risk for HNPCC occurred (Koehly et al., 2003).

Sample

Data were gathered from multiple members of families with and without identified HNPCC mutations. Participants included index cases and their first-degree relatives (FDRs). Index cases received either indeterminate or mutation-positive genetic test results; in this analysis, their families are defined as families without identified mutations and those with identified mutations, respectively. FDRs were the adult biological siblings and children of the index cases. These individuals were included because they are considered to be at increased risk of cancers associated with HNPCC, regardless of whether a family mutation was identified.

Data were collected through an IRB-approved addendum to a longitudinal study (National Human Genome Research Institute, 95-HG-0165), which offered genetic education, counseling and the option of genetic testing to individuals suspected of or at risk for HNPCC (Hadley et al., 2003; Hadley et al., 2004). All participants gave verbal informed consent.

Procedure

All index cases and FDRs of mutation-positive index cases had previously participated in the ongoing longitudinal study, and were sent a flyer describing the addendum. Interested individuals contacted the study team and completed a semi-structured telephone interview. FDRs of indeterminate index cases had not participated in the parent study, and were identified using modified cascade sampling (Beeson & Doksum, 2001). Indeterminate index cases enumerated FDRs to whom they were willing to distribute materials inviting them to participate. Interested FDRs contacted the study team and completed the telephone interview. Following the interview, all respondents received retail gift cards.

To facilitate comparison of communication patterns, families with identified mutations were matched to families without identified mutations. Indeterminate index cases were characterized by gender, age, and type of cancer, then matched to mutation-positive index cases based on these characteristics, resulting in matched family pairs. All participating FDRs from matched families were included in the analysis.

Measures

The first question in the semi-structured telephone interview generated each participant's family network. Participants were asked, "When I say family, who do you think of?" Individuals enumerated in response to this question could be biological, nonbiological, or social kin. Follow-up questions were used to obtain limited demographic data on these family network members, or *alters*.

Outcome variables—The outcome variables of interest for this analysis were two communication networks: communication about genetic counseling and testing for HNPCC and communication about risk for HNPCC cancers. These were assessed using two questions: "With whom in your family have you talked about genetic counseling (information) and testing for HNPCC?" and, "With whom in your family have you shared your thoughts about your HNPCC cancer risk status?" In the first question, "genetic information" was used with FDRs of indeterminate index cases, who had not received genetic counseling through the parent study.

Predictor variables—Predictor variables of interest were individual and relational factors hypothesized to affect communication about genetic testing and risk for HNPCC, based on findings in the literature. Predictor variables include: generation relative to that of the respondent, gender, kinship tie to the respondent, social support, social influence, perceived similarity, and HNPCC risk status (Carlsson & Nilbert, 2007; Koehly et al., 2003; McPherson, Smith-Lovin, & Cook, 2001; Mesters et al., 2005; Peterson, 2005; Peterson et al., 2003; Stoffel et al., 2008; Wilson et al., 2004). Respondents provided data for all predictor variables except HNPCC risk status. Generation and kinship tie were treated as categorical variables. Alters were assigned to one of three categories for generation, based on their relationship to the respondent: older, younger, or same generation as the respondent. Unless otherwise indicated, friends were considered to be in the same generation as the respondent. Respondents indicated each alter's gender and kinship tie with the respondent. Alters could only have one kinship tie: partner/spouse, biological family member, nonbiological family member, or social kin. Respondents also listed alters who provided social support; dispensed advice (i.e. exerted social influence); those who respondents said they would listen to; those with whom respondents said they were close; and those with whom respondents perceived similarity in personality or physical appearance.

HNPCC risk status was extracted from family pedigrees. For biological kin who had undergone genetic testing for known family mutations, risk status was based on genetic test results. Biological kin who had not had genetic testing, or who were members of families without identified mutations, were all considered to be at risk. Partners/spouses, nonbiological kin, and social kin were considered not-at-risk for HNPCC.

Covariates—Covariates controlled for dependencies within the family communication networks. Density controlled for the overall rate of discussion in each family network, and variability across families. Since respondents may exhibit individual differences in their propensity to discuss genetic information or personal risk with others (out-stars) and respondents from the same family may engage the same people in these discussions (in-stars), analyses controlled for these network structures. Out-stars controlled for variability in the number of persons nominated by the respondent on each outcome variable of interest (outdegree). In-stars controlled for variability in the number of nominations alters received from participants within the same family (indegree) (Anderson, Wasserman, & Crouch, 1999).

Data Analysis

Hierarchical nonlinear models were fitted to examine associations between individual and relational characteristics and communication about genetic counseling and testing and risk for HNPCC. Hierarchical modeling accounted for the nesting of network ties within families (Bryk & Raudenbush, 1992; Snijders & Bosker, 1999). Models were fitted using HLM 6.06 (Raudenbush, Bryk, & Congdon, 2004). Separate analyses were completed for each outcome variable. Same generation and nonbiological family member were the referent categories for analyses examining generational differences and kinship tie, respectively. To control for dependencies within each family communication network, density, in-stars, and out-stars were included as covariates in all fitted models. A total of 599 alters were included in each analysis. A Type I error rate of .05 was used to evaluate the statistical importance of each predictor variable.

The “step up” method was used to fit multivariate models (Bryk & Raudenbush, 1992). The level-1 model was built using a stepwise approach. For each outcome variable, predictor variables that were significant in bivariate models were entered into the multivariate model. The primary aim of the current paper is to assess whether associations between selected individual and relational characteristics and patterns of communication about HNPCC vary according to family mutation status. To assess this, moderational analyses were conducted by testing the interaction between mutation identification within the family and each predictor variable (Baron & Kenny, 1986). The variable representing mutation identification is a family level variable, and was entered at level 2. A main effect of mutation identification and interactions with each level 1 predictor were considered. Using a stepwise approach, nonsignificant interactions were removed from the model, suggesting that there were no differences in these associations between families with and without identified mutations. Nonsignificant main effects were removed if they did not contribute to a significant interaction.

Results

The final sample included 20 index cases (10 with indeterminate genetic test results) and 31 FDRs (16 from families where the index case received indeterminate results) (Table 1). Most participants were female and middle-aged, and all identified themselves as Caucasian. All index cases had a personal history of at least one HNPCC-associated cancer; a minority of FDRs had a personal history of cancer. Data were gathered 3-12 years after index cases received genetic test results.

Network Members

For all respondents, an average of 11.75 (3-43) persons were enumerated as family members. Half of alters were in the same generation as the respondent (50%), while 33% were in the younger generation. Most alters (55%) were female, at risk of HNPCC (53%), and members of the biological family (70%). A substantial minority of network members were social kin (10%) or nonbiological kin (10%). Respondents indicated that they were close to over half of their network members (52%). Just over half of alters were identified as providing emotional support (51%); a smaller percentage (38%) provide tangible support. Respondents would take advice from 42% of their alters, and would listen to the vast majority (72%) of their family network members.

Breadth of Communication Networks

On average, members of families without identified mutations enumerated 3.74 more family members than those from mutation-positive families (13.58 vs. 9.84; $p=.035$). On average, respondents from families without identified mutations shared their thoughts about being at

risk for HNPCC with fewer family members (3.08), compared to respondents from families with known mutations (7.60; $p<.001$). Subsequent analyses examined the proportion of family network members with whom respondents communicated, relative to the total number of named family network members. Respondents from families without identified mutations talked about genetic counseling and testing with a smaller proportion of enumerated network members, compared to respondents from families with identified mutations (.36 vs. .66, $p=.002$). Respondents from indeterminate families also shared their thoughts about risk for HNPCC with a significantly smaller proportion of enumerated network members (.23 vs. .77, $p<.001$).

Communication about Genetic Counseling and Testing

Respondents were significantly more likely to discuss genetic counseling and testing with female alters (Table 2). Compared to nonbiological family members, respondents were more likely to talk to their biological family members, and less likely to talk to their social kin. Respondents were more likely to talk to family network members with whom they are close, and who provide them with tangible support. No significant interactions with mutation identification (i.e. family mutation status) remained in the final multivariate model.

Shared Thoughts about Risk

Respondents were also significantly more likely to talk about their risk with network members who are female (Table 2). Compared to network members who are in the same generation as the respondent, respondents were significantly less likely to talk to network members who are younger. Respondents were more likely to share thoughts about risk with family network members who are close and who provide them with emotional support. Compared to nonbiological kin, respondents were less likely to share their thoughts about risk with social kin, but were more likely to share thoughts about risk with biological kin. One significant interaction with mutation identification remained in the final model. Respondents from indeterminate families were more likely to share thoughts about risk for HNPCC with network members whose advice they took ($OR = 17.81$), compared to respondents from mutation-positive families.

Discussion

These findings provide important insights into the selection of network members for communication about genetics and cancer risk in families diagnosed with HNPCC. This analysis expands on previous studies by directly comparing communication patterns in families with and without identified mutations. Including FDRs of index cases with HNPCC but without identified mutations provides more extensive, novel data on the communication process in families without identified mutations. Patterns of communication in families with and without identified mutations were largely congruent. Differences emerged in the breadth of communication about HNPCC, and that members of indeterminate families were more likely to tap advice givers within their family networks when sharing thoughts about risk for HNPCC.

Members of families without identified mutations talked about genetic counseling and testing with a smaller number and proportion of family network members. This has implications for the spread of genetic risk information and cancer screening recommendations in families without a molecularly confirmed diagnosis. Following recommendations for early and frequent screening is important for prevention of CRC and identification of cancer in its early stages, influencing long-term health outcomes in at-risk families. Thus, it is important for this information to reach all at-risk family members. In previous studies (Stoffel et al., 2008), index cases with indeterminate or true negative

genetic test results shared those results with a smaller percentage of second- and third-degree relatives than mutation-positive index cases. The present analysis included communication about genetic counseling and testing, not just sharing of genetic test results. FDRs of indeterminate index cases were not eligible for genetic counseling and testing through the parent study, which may have limited their discussion of these topics. Whether access to genetic counseling and testing is associated with communication about these topics should be examined in future research.

Members of families without identified mutations were also less likely to share thoughts about their risk with family network members. This is surprising, since an extensive family history of HNPCC-associated cancers contributes to thoughts about risk among members of families with indeterminate HNPCC genetic test results (Ersig, Ayres, Hadley, & Koehly, 2010) and most families in these analyses had strong family histories of cancer, regardless of family mutation status. Unlike members of indeterminate families, members of mutation-positive families can pursue genetic testing to clarify their risk for HNPCC-associated cancers. Obtaining concrete risk information through genetic testing may facilitate sharing of this information with other family members. Being able to discuss genetic testing could also influence with whom at-risk individuals choose to share information about their risk status. Lower rates of communication in families without identified mutations could also reflect less discussion about HNPCC in general. A more in-depth analysis of the content of communication in families without identified mutations is warranted.

Respondents from indeterminate families were more likely to share thoughts about risk with network members who also provide them with advice. This could reflect the uncertainty of indeterminate genetic test results (Stoffel et al., 2008; Wilson et al., 2004). Ambiguous risk information could cause emotional difficulty, and respondents may seek help with decisions about how to address their risk (Koehly et al., 2003; Shiloh, Koehly, Jenkins, Martin, & Hadley, 2008; Vadaparampil, Wey, & Kinney, 2004). This finding should be explored further, perhaps by asking at-risk individuals about their motivations for selecting particular communication partners.

Results from this study parallel those from earlier studies demonstrating that women, regardless of risk status, are central to genetics-focused communication in families (Koehly et al., 2009; Koehly et al., 2003; Patenaude et al., 2006). In the present study, women were central to communication about genetic counseling and testing and risk for HNPCC in families with and without identified mutations. Similar findings in families without identified mutations indicate that women are kin keepers of family health information, not just genetic test results. Examining more extensive communication networks could improve our understanding of the unique role of women in the dissemination of risk information within the family system. This information may be important for extended family members, such as second and third degree relatives, who were not considered in the current report. The importance of women in disseminating risk information within the family system suggests that they may play a particularly important role in educating family members about behavioral approaches to preventing CRC or identifying it in its early stages. These women may be optimal family health educators within social network-based interventions that target these behaviors.

Kinship ties were important in respondents' communication patterns. Respondents were more likely to talk to biological family members about genetic counseling and testing and risk for HNPCC than nonbiological kin. Interpretations of the family history of cancer, and their implications for perceptions of who within the family is at risk, could lead respondents to target biological kin when discussing genetic counseling and testing and risk for HNPCC. Family mutation status did not moderate the association between kinship and

communication; however, reasons for communicating with biological family members may differ. In mutation-positive families, biological relatives may be targeted because of increased risks of carrying the mutation, and eligibility for genetic testing. In families without identified mutations, communicating with biological family members may help ensure that all at-risk individuals receive appropriate information. Family history and perceived duties and responsibilities motivated communication of genetic risk information in families at high risk for hereditary forms of CRC (McCann et al., 2009). Comparing the motives behind communication in families with indeterminate vs. mutation-positive test results would add to our understanding of these processes.

Similar to studies of mutation-positive families, this study found that the nature of relationships is associated with communication about genetics and risk (Koehly et al., 2003; Wilson et al., 2004). Respondents in this study communicated with network members who provide them with different forms of social support. These measures may be proxies for relationships among family members, which were not explicitly examined in this analysis. In previous studies, at-risk individuals shared genetic risk information in a bid for emotional support (Stoffel et al., 2008). It is possible that respondents chose to communicate with network members who had previously provided some form of support, assuming that they would provide similar support in this situation. Communicating with close network members may help achieve similar goals; working to build interpersonal ties among already close family members may facilitate coping with genetic risk information (Peterson, 2005).

Respondents were also more likely to share thoughts about their risk for HNPCC with members of the same generation. This may reflect shared experiences within the family, which influence topics of discussion within families with identified HNPCC mutations (Palmquist et al., in press). Reasons for selection of different family members were not explored in this analysis. Exploring motivations for communication with different family members would provide valuable insight into these processes, as reasons for limiting discussion with older or younger family members may be similar or different.

Findings from this study were congruent with the Family Systems Genetic Illness model (Rolland & Williams, 2005); the social network among family members has important implications for the health status of at-risk members (Berkman et al., 2000). Family system characteristics, including different aspects of family relationships, affected patterns of communication about genetics and risk for HNPCC. Of note were the psychosocial mechanisms that operated through the family networks, which were associated with communication about genetics and risk for HNPCC.

Future Directions

Additional research would provide important data on the choices behind communication processes, and the potential implications of family communication. Very little is known about perception of genetic risk information and reasons for sharing this information with particular network members. Of particular interest is what information is shared among family members, specifically, in-depth examination of the content of communication about genetics and risk. The outcome measures in this analysis focused on very broad topics. Although there were few differences in the communication patterns of members of families with and without identified mutations, there may be significant differences in the content of that communication. In addition to including families with different genetic test results, future studies could compare types of family members (e.g., index cases, children) to determine whether family role influences communication about HNPCC, and provide important insights into family relationships and interactions. Exploring the naturalistic communication of genetic information within at-risk families could provide a basis for

interventions designed to take advantage of naturally occurring social networks in families at risk for hereditary conditions.

Limitations

Factors limiting the generalization of these findings include the small number of participants from each family, lack of ethnic diversity, and recruitment from an existing research population. On average, 2-3 people per family participated. This resulted in incomplete network data, which do not capture the full complexity of the family structures.

Conclusions

Findings from this study extend our knowledge of communication in families with HNPCC to include the many families in which a causative mutation has not yet been identified. In light of these findings, genetics education and counseling should be offered to families meeting clinical and pathological criteria for HNPCC, even in the absence of an identified disease causing mutation, to facilitate communication about cancer risk and cancer screening. This study identified few differences in the communication patterns of families with and without identified HNPCC mutations, despite findings in the literature suggesting that individuals and families without identified mutations might have more difficulty understanding those results and sharing them with others (Stoffel et al., 2008). Continued inclusion of families without identified mutations in studies of families at risk of hereditary cancer syndromes will improve our understanding of communication and dissemination of risk information.

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Table 1

<i>Respondent Demographics</i>		Members of Mutation-Positive Families (n=25)			Members of Indeterminate Families (n=26)		
Variable	All	Index Cases (n=10)	FDRs (n=15)	All	Index Cases (n=10)	FDRs (n=16)	
Female	21 (84%)	8 (80%)	13 (87%)	23 (89%)	8 (80%)	15 (94%)	
Age	49.12 ± 15.04	56.60 ± 8.71	44.13 ± 16.51	48.58 ± 13.29	54.70 ± 8.26	44.75 ± 14.58	
College Degree	52%	40%	60%	65%	78%	63%	
Partnered	72%	80%	67%	65%	70%	63%	
History of CRC	10 (40%)	8 (80%)	2 (13%)	8 (31%)	7 (70%)	1 (6%)	
History of CRC + another cancer	7 (28%)	6 (60%)	1 (6.6%)	4 (15%)	3 (30%)	1 (6%)	
Other HNPCC cancer	4 (16%)	2 (20%)	2 (13%)	3 (11.5%)	3 (30%)	0	
Other cancer	0	0	0	1 (4%)	0	1 (6%)	

Table 2

Final Multivariate models

Variable	OR	95% CI	p-value
Communication about genetic counseling and testing			
Female alter	1.64	1.02- 2.63	.011
Partner	4.23	1.54- 11.60	<.001
Biological family member	7.34	3.83- 14.05	<.001
Nonbiological family member	<i>Referent</i> ¹		
Close	4.14	2.40- 7.15	<.001
Tangible support	2.56	1.49- 4.41	.004
Shared thoughts about risk			
Nonbiological family member	<i>Referent</i> ²		
Social kin	.16	.06- .43	.015
Close	5.44	2.85- 10.38	<.001
Advice	.77	.34- 1.73	.676
x Indeterminate	7.17	2.42- 21.22	.008
Emotional support	4.09	2.26- 7.39	.006

¹ To reduce instability in the model, kinship categories of health care provider and social kin were collapsed into the referent to construct the model for communication about genetic counseling and testing. The model controls for density, out-star, and in-star.

² To reduce instability in the model, kinship categories of partner, biological family member, and health care provider were collapsed into the referent to construct the model for sharing risk status with others. The model controls for density, out-star, and in-star.