Living with Hereditary Non-polyposis Colorectal Cancer; Experiences from and Impact of Genetic Testing.

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Life after genetic testing for hereditary nonpolyposis colorectal cancer; experiences and the impact of experiential knowledge

Running head: Experiences from HNPCC testing

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ABSTRACT

Hereditary nonpolyposis colorectal cancer (HNPCC) is one of our most common cancer syndromes and an increasing number of individuals live with knowledge about HNPCC carrier status. We conducted an interview study in order to investigate the perceived impact on life after genetic testing for HNPCC. Three themes emerged; reactions and emotions, family relations, and implications and strategies. The informants referred to experiential knowledge of cancer, information responsibilities, impact on family relations, and coping strategies. Most informants had suspected hereditary cancer, but confirmation hereof was described as an overwhelming experience and often brought about changes in life. We suggest that the importance's identified, e.g. the impact of experiential knowledge and the impact on family relations, should be taken into account during genetic counselling in order to facilitate information spread and prepare family members for the impact that the genetic knowledge may have on life.

KEY WORDS Hereditary nonpolyposis colorectal cancer; Genetic testing; Life; Mutation carriers; Non-mutation carriers; Interview; Experiences; Experiential knowledge

INTRODUCTION

Hereditary nonpolyposis colon cancer (HNPCC) is one of the most common cancer syndromes and close to 1000 such families have been identified world-wide (INSIGHT) (International Society for Gastrointestinal Hereditary Tumours). Hence, a large number of individuals live with knowledge about HNPCC and cancer risks related hereto. Carriers of HNPCC-causing germline mismatch repair gene mutations are at significant risks for colorectal cancer (80% life-time risk), endometrial cancer (40-60% risk), and at increased risks also for ovarian cancer, gastric cancer, urothelial cancer, brain tumors, and skin tumors (Aarnio *et al.,* 1999). The mean age at tumor development is 45 years with a large variability in age of onset, also within the same family. The identification of disease-causing genetic mutations confirms the diagnosis and enables predictive genetic testing with inclusion of high-risk individuals into control programmes aimed at preventing additional cancer cases in these families. The dominant inheritance implies that 50% of first degree relatives will be mutation carriers with most families thus containing individuals with high risks of cancer as well as those without any increased risk. Verified HNPCC allows for predictive genetic testing, and the choice to undergo such a test will either confirm the presence of an allele linked to a high risk of cancer or demonstrate non-inheritance of the allele that caused cancer in affected family members.

Moreover, the confirmation of hereditary cancer in the family often involves duties to confer information about heredity to other family members, concerns related to the disclosure of the information, and worries for other family members (Koehly *et al.,* 2003; Meiser, 2005; Sommerville and English, 1999; Wilson *et al.,* 2004). Nine out of ten individuals who have undergone genetic testing for HNPCC are satisfied with their decision, but 59% still state that the test result had a significant psychosocial impact on their life (Esplen *et al.,* 2001). After

genetic testing, mutation carriers show similar, or sometimes increased, levels of distress, whereas distress decreases in non-carriers (Meiser, 2005; Meiser *et al.*, 2004; Meiser and Halliday, 2002; Wagner *et al.*, 2005). However, current knowledge suggests that predictive genetic testing is not linked to adverse psychosocial consequences and only a small subset of the mutation carriers report persistent and pronounced cancer worries (Broadstock *et al.*, 2000; Meiser and Halliday, 2002; Wagner *et al.*, 2005). Most psychosocial studies in HNPCC have focused on distress, anxiety, and perceived risks, whereas data regarding how these individuals describe their life situation, e.g. regarding thoughts, coping abilities, and psychosocial resources are scarce. Since hereditary cancer may influence various aspects of life we performed an interview study in order to investigate the perceived impact on life after genetic testing for HNPCC.

METHODS

Informants and procedure

Individuals who had undergone genetic counselling and predictive genetic testing at the Oncogentic clinic, Lund University Hospital, Sweden within 1.0-1.5 years were eligible for the study. These individuals had participated in an initial counselling session wherein pedigree data were reviewed, a risk estimate was given, and the genetic defect, i.e. a mismatch-repair gene mutation linked to HNPCC, was explained. The patients were thereafter asked to reflect about whether they wanted to undergo predictive genetic testing for the disease-causing mutation. All patients in the study opted here fore, and the overall uptake rate is >95%. At a second counselling session the test result, which either showed that the individual was a mutation carrier or a non-carrier, was communicated. All individuals also received an individualized letter containing this information, and were referred back to their local hospitals for controls, e.g. with colonoscopies and gynaecological examinations.

Initially, information about the study and a confidentiality agreement was mailed to 49 individuals. One reminder was sent out and totally 22 informants opted for participation, and due to practical reasons we could arrange interviews with 19 informants. The interviews were generally held in the informants' homes, lasted about 45 - 60 minutes, and were transcribed verbatim (Mishler, 1986). With the intention to let the informants "tell their story in their own way" open-ended interviews were used and were initiated; "I would like you to describe living with genetic knowledge as a mutation carrier/non-carrier as you see it". The study sample from 13 HNPCC families consisted of 11 mutation carriers (5 female and 6 male) and 8 non carriers (5 female and 3 male). The mean age among the carriers was 51 (33-75) years and among the non-carriers 47 (36-64) years.

Data analysis

The transcribed texts were analysed by inductive content analysis (Silverman, 2001). The authors - C.C., a nurse specialised in oncology and M.N., a physician specialized in oncology and genetic counselling - read and re-read the transcribed texts with focus on the aim of the study. Words, statements and sentences that were identified to correspond hereto were noted and arranged into primary themes and sub-themes and were separately analyzed for mutation carriers and non-carriers. As the work proceeded, the content of each theme was expanded or reduced and additional themes were formed. The authors reached consensus on the final themes and sub-themes. Excerpts referring to individual informants are used to illustrate the content of themes and sub-themes. Ethical approval was granted by the Lund university ethics committee (569/2005).

RESULTS

The analyses emerged three major themes; reactions and emotions, family relations, and implications and strategies. Hereunder, several sub-themes were identified and the data are summarized in table I.

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Reactions and emotions

Verifying heredity

Carriers as well as non-carriers commonly reported suspecting heredity before the confirmed HNPCC diagnosis, whereas two non-carriers (informants 5 and 15) reported that the knowledge of hereditary cancer came as a surprise. In both mutation carriers and non-carriers these thoughts were evoked by discussion between parents and grandparents who expressed worries for the many cancer cases in the family. Although many informants had suspected heredity, learning hereof was described as an overwhelming experience:

> /…/ it felt like if a samurai sword hit me from behind, made me loose hold, fall, and requested a fresh start/…/ (6, non-carrier).

> /…/A revolutionary year….one has endured it…it's an experience/…/ (5, non-carrier).

Feeling anxious

Mutation carriers as well as non-carriers reported feeling tense and worried before, during, and after the genetic testing process. One female mutation carrier (informant 4) described how worries about being a mutation carrier made her postpone the appointment at which the genetic test result should be given since she needed time to mentally prepare for

being a mutation carrier. Sorrow and affected sleep were also reported by some mutation carriers. A non-carrier (informant 10) described how she also handled her sister's (who is a mutation carrier) anxiety and worries, which demonstrates that, besides their own fear and worries, family members, may have to handle also that of the relatives. Being a mutation carrier with or without previous cancer also evokes fear of cancer when new symptoms are encountered, even if these are vague and most likely related to e.g. a cold or a stiff back. Some mutation carriers also described an increased anxiety when family members fell sick from whatever cause with increasing worries if several family members were affected.

Feelings of injustice and guilt

Both mutation carriers and non-carriers refer to feelings of injustice and guilt, but from somewhat different perspectives. Non-carriers report feeling guilty about the test result, whereas carriers refer to guilt in transferring the defective gene and to injustice in a more global sense e.g. how cancer strikes in different situations and phases of life with young family members affected while elderly relatives are not.

Experiential knowledge matters

Several informants (males and females, mutation carriers and non-carriers) describe experiences from being close to or caring for relatives with cancer and refer to symptoms, diagnostic investigations, side-effects from treatment, and memories from when old and young family members fell sick or died. Situations in which the informants have been active themselves raise emotional reactions yet to this day. This can be exemplified by a 19-year-old girl who talked about how she was asked to open and read a letter from a doctor and how she hereby delivered the news that her mother had cancer:

> /…/ so I opened the letter and read: Dear Ms NN, the test shows cancer'…there I sat and read it to my mother…it was terrible/…/ (15, non-carrier).

One mutation carrier also described how he from the number of individuals' affected calculated not being a carrier (which he indeed was):

/…/ I was completely convinced that I did not have it (the mutation)….it is very strong that gene, the genetic defect in our family. Almost 100% of those tested had it so I was convinced not having it/…/ (7, mutation carrier).

One non-carrier reports how the positive information (of not being a carrier) doesn't always mean that everything is forgotten, but describe how certain situations her as well as her children of the time of genetic testing with a need to talk about the process:

> /…/ it may occur when we watch TV….and are reminded about cancer….now a teacher at school died from cancer….then we talk about it again/…/ (5, non-carrier).

Family relations

Responsibilities for conveying information

The importance of understanding and revealing the genetic test result was referred to by almost all carriers and half of the non-carriers. The informants used the expressions "advocate", "support", and "engage" to describe feeling responsible for ensuring that family members at risk may be tested and included into control programs. The responsibility for information includes close family members as well as family members in other regions and countries. Particular difficulties may be experienced in conveying the information about hereditary cancer to family members with whom one has only sporadic contact. When informing family members, personal views on expected reactions and vulnerability come into play, e.g. who is likely to react negatively, to whom this information may be harmful rather than beneficial? One mutation carrier (informant 19) describes how she arranged genetic counselling for her brothers since she felt they would not independently take such action. The

responsibility and wish to find out what is best for the children is illustrated in the following quotation:

> /…/Yes, it became my responsibility…my responsibility and I feel I bit guilty about these things, but somehow it´s my responsibility…..you wish the best for your children some way/…/ (13, mutation carrier).

Informing teenagers may be difficult since predicted reactions may go unnoticed:

> /…/ it wasn't difficult to tell….of course it was not as hard since it was positive (non-carrier)…..but there were no questions or reactions, but teenagers have another focus like…..I have to go to the Internet now/…/ (6, non-carrier).

Furthermore, the informants referred to how the information has to be put forwards in a manner that underscores the seriousness, but also provides hope for preventing cancer. One specified difficulty related to mutation carriers who chose to deliver the information via mail. Also, the best time for delivering the news is referred to and one mutation carrier (informant 4) described how diseases or deaths within the family delayed the spread of information.
A difficulty in informing relatives is illustrated by a the same mutation carrier's (informant 4) story about how she had abruptly received the information about the hereditary from her cousin in a parking lot when loading groceries into the car. When she was to pass on the information to her husband her courage failed her and she postponed informing him for several days, and thereafter delivered the information just as abruptly as she had received it herself.

Different reactions among family members

Mutation carriers as well as non-carriers describe different reactions among family members; silence, confusion, positive responses related to the option to participate in control programmes, irritability, and refusal to know, which is exemplified in:

> /…/ some family members argue. They claimed it [HNPCC in the family] was a lie/…/ (4, mutation carrier).

Different attitudes and reactions may also be encountered within a partnership. One mutation carrier (informant 7) refers to himself reacting in "black and white", whereas his partner rather tends to ponder and reflect and besides personality. Life phase and problems related to e.g. economy or diseases also contribute to the differences in information handling.

Relations and communication

Both mutation carriers and non-carriers describe how family relations were affected when hereditary cancer was verified; old disputes and new aggressions occurred in families with poor relations, whereas difficulties brought the members closer in families with good relations. The latter is exemplified in:

> /…/ we had been rather close before as well….we have talked quite a lot…I think we talk more freely and personally than we did before/ (8, mutation carrier).

Within three families (informants 4, 5, 9, and 10) discussions around hereditary cancer were described to have a negative impact and brought up old conflicts related to e.g. parents not taking enough responsibility or being perceived as unfair during childhood. Some informants also described how they, because of the genetic investigations, met with relatives with whom they did not wish to continue relationships. In one family the members found it hard to spend Christmas together after learning about the genetic defect and gave up this tradition, which the father found hard to accept. Adverse feelings for some relatives are

described and may extend also to diseased family members; a mutation carrier (informant 4) described how she, when upset, can envisage pictures of family members who “gave her” the trait. Disputes were also described to occur while waiting for test results and informant (5) described how her teenager worried and affected while waiting for their mother’s result. Although this showed that she was a non-carrier, the tensions remained for some time, reflecting that adaptation to new circumstances may take time. Other informants describe how they may tease or disturb each other referring to the genetic defect, and such small gibes are illustrated in the following quotation:

> /…/I can disturb and joke with my mother when I say ”thank you for the defective gene”/…/ (1, mutation carrier).

Genetic diagnostics may alter the previously known family tree when e.g. step/half siblings and extramarital children are identified in the process. One non-carrier (informant 6) described how he learnt not only about hereditary cancer but also about new relatives and realized that the father he grew up with was indeed his stepfather, and was deeply disappointed over the family not telling him and neglecting to inform him about hereditary cancer.

Mutation carriers as well as non carriers refer to situations in which they perceive a need for communication aid e.g. optimal timing and situation to inform family members about hereditary cancer, but the need varies between different family members and may change over time. Whereas some informants describe how they perceive that cancer will affect all family members, others rather estimate that the personal risk is lower since so many family members have already been affected.

Worries for family members

Worries for the children and thoughts about how heredity will affect their life predominate among mutation carriers and are described to provoke insecurity. The perceived cancer risk for children and grandchildren is based on calculations and individual experiences within the family, which is exemplified in:

> /…/ each child has a 50% risk of inheritance so – I have three children….it´s just 1.5 /…/
>
> (3, mutation carrier).

In order to handle the worries heredity may also be related to broader aspects of life, which can be illustrated by the following reflection:

> /…/It´s not like a death sentence…..it's just a defect….a gene linked to colorectal cancer/…/ (7, mutation carrier).

Implications and strategies

Coping strategies

Both mutation carriers and non-carriers talk about how emotions related to hereditary cancer may be suppressed; denial (e.g. persuading oneself that it could have been worse), projections (e.g. anger with relatives who have transferred the genetic defect), and strategies for handling emotions (e.g. focusing on work and other tasks, in order to reduce time for reflection about the genetic defect). One non-carrier (informant 6) described his mental preparation already during investigation about the heredity, in which he envisaged life filled with hospital visits for tests and controls and imagined himself affected by the worst, i.e. cancer. The informants exemplify emotional coping by convincing one self that cancer can be overcome, preparing for the worst, and not wasting energy on a situation that can not be changed. Several carriers also report benefit from sharing their feelings with others.

Perspectives and acceptance

Mutation carriers admit that thoughts of heredity often are present, and refer to the need to accept the situation. Herein, they describe learning to appreciate the positive aspects of life and to regard their risk of cancer from a more global perspective and in relation to other aspects in life:

> /…/ there are so many negative events in life…..this little gene has a small impact overall/…/ (7, mutation carrier).

Both carriers and non-carriers described how they kept the knowledge about hereditary cancer to themselves for some time, which likely reflects a need to grasp the situation and to determine whom to inform and when to optimally do this. Mutation carriers describe the process of accepting carrier status, which involves taking in new knowledge, understanding its implications, and accepting the consequences. One individual explains how these thoughts occurred already on his way from the hospital:

> /…/ there were many thoughts and reflections around what could happen…..what could happen to whom/…/(13, mutation carrier).

Adaptation, change and new choices

Mutation carriers as well as non-carriers also refer to their 'individual journey' through genetic counselling, genetic testing, and learning about the estimated risk and recommended preventive measures. Mutation carrier-informants describe how genetic testing have influenced their values in life, e.g. how they catch the moment, find time as precious, and are prone to make dreams within reach come true. Mutation carriers describe how they choose to prioritize important moments and refrain from wasting time on arguments and conflicts and some of them also describe how the insight of being at an increased risk of cancer have evoked serious thoughts about the possibilities to change life.

One woman described how she regretted not having testing earlier since she had calculated being affected by cancer at the same age as her mother and had therefore chosen early retirement, which she partly regretted:

> /…/ so I would probably not have quit work the way a did – as an early retirement – I calculated not living past 65…/…/ (15, non-carrier).

Mutation carriers describe that they believe life differs in several aspects between carriers and non-carriers, including e.g. fear of cancer, arranging life around controls, and worries for the children. On the other hand, several mutation carriers describe how they with new insight and participation in control programmes have not changed their everyday life. Indeed, one mutation carrier (informant 19) described that learning about hereditary cancer was a minor problem related to the complications during her time as a cancer patient. Some carriers also reported that heredity frightened others.

Experiences from control programmes

Knowledge about genetic testing, recommended control programmes and options for prophylactic surgery and attitudes toward the controls were discussed by mutation carriers. The first control is often described as the worst, but the controls confer a sense of security and are viewed as a privilege and mutation carriers describe how risks and controls become part of their life with examination intervals “printed” into their minds. The trust in control programs gradually increase and several mutation carriers described how participation herein becomes a natural part of life. Experiences from removing precursor lesions and from family members who recover from cancer also strengthen the motivation for participating in control programs. The mutation carrier refers to the control programmes and many statements refer to prevention and behaviour in daily life. Even though mutation carriers predominantly report trust in control programmes, they also report worries before the controls. The following

quotations illustrate the temporary worries before the control and preventive thinking of controls and generated preventive behaviour in daily life:

/…/"will they find something this time/…/ (1, mutation carrier).

/…/it's such a small risk I will develop [cancer] again….they will remove it before that/…/ (2, mutation carrier).

/…/ of course you're more observant/…/ (9, mutation carrier).

Uncertainty about the future

Mutation carriers as well as non-carriers envisaged how their future appeared before and after genetic testing and herein identified wished to focus of the positive sides of life and indeed experiencing a lower risk of cancer than previously perceived. However, one mutation carrier described a change of mood:

/…/one of my friends said "you never laugh nowadays"…yes, I rarely do. Before, I could laugh at small, silly things…but I can't do that now/…/ (4, mutation carrier).

Knowledge about hereditary cancer is likely to confer insecurity about the future and specially the future of the children and grandchildren. Being able to witness grandchildren growing up may not become reality according to some mutation carriers. Other people's reaction to hereditary cancer was also referred to by mutation carriers with individual experiences from cancer who described how they sometimes felt that cancer scares friends and relatives.

Family planning issues

Informants (1, 7 and 8) who talked about family planning issues generally stated that they wanted children and that the increased risk of cancer had not changed this wish. The only

male who brought up the question about children was a mutation carrier who described his mother's concern about passing on the defected gene and how he felt about this:

> /…/ then none of us should have children really, since we all have some defective gene…..some more serious than others……so you can't act like that (7, mutation carrier).

DISCUSSION

Genetic counselling for hereditary cancer and, in families where the underlying genetic defect has been identified, predictive genetic testing has largely evolved and made readily available during the last decade. The main motives for undergoing genetic diagnostics of HNPCC are hopes for early detection of cancer, knowledge about the children's risks, and reduction of uncertainty with the latter motive being linked to a reduced stress, particularly for non-carriers (Claes *et al.*,; Esplen *et al.,* 2001; Vernon, 1999). Cancer cases in the family, frequent cancer thoughts, and a better ability to cope with unfavourable results have been linked to the uptake in genetic testing (Codori *et al.,* 1999; Loader *et al.,* 2002). Although most individuals in our study described having suspected heredity, the verification hereof was perceived as an overwhelming experience. Feelings of anxiety and guilt were referred to, although with different contents and implications among mutation carriers and non-carriers.

Individuals seeking genetic counselling commonly use their family history and individual experiences from cancer, rather than statistical facts, as reference points for their own risks perceptions and actions. Indeed, one-third of individuals at risk of HNPCC have been found to use personal theories of inheritance or anticipate being carriers (McAllister, 2003). Experiential knowledge, which may be derived from others, i.e. empathetic, or gained from subjective experiences, i.e. embodied, has been demonstrated to influence understanding of factual information, risk perception, cancer-related worry, and long-term anxiety in

families with hereditary cancer (d'Agincourt-Canning, 2005; Hopwood *et al.*,, 2001). Recent data from HNPCC families also suggest that individuals who have experience from or who have lost close relatives to cancer, particularly if several family members have been affected or if these experiences were gained at young age, are psychologically vulnerable (Esplen *et al.*, 2003; van Oostrom *et al.*, 2006a, , 2006b). Experiential knowledge is thereby likely to have a key role in how individuals with hereditary cancer handle their situation and in our study, 13/19 informants referred to the impact of experiential knowledge. In genetic counselling ascertaining that the individuals have understood the genetic defect and the risk implicated is important, and current data suggest that experiential knowledge should be encompassed herein in order to minimize exaggerated risk estimates (Schneider, 2002). This can include e.g. asking whether the individual has been involved in the treatment or the care-taking of family members affected by cancer and questions related to the individual's views on cancer cure. Hereby, factual risks can be discussed in relation to the individuals' experiences and feelings, which may contribute to a learning process shared by the patient and the genetic counsellor (Shiloh, 2006; Shiloh *et al.*, 2006). The informants described coping strategies and referred to the implications that the genetic knowledge had on their life situation. During genetic testing informants also described how they mentally prepared for being mutation carriers. Although most individuals report leading life after genetic counselling the same way as they did before, several individuals describe how the genetic knowledge served as an initiator for performing a desired change in life.

The responsibilities to convey information about hereditary cancer to other family members was referred to by almost all carriers and by about half of the non-carriers. With the responsibility to inform family members follows decisions about whether contact should be taken with relatives of diseased family members, which family member is best suited and

willing to deliver the news, and choosing the right time. Unaffected women in families with hereditary breast cancer have, in line with our findings, reported that the impact of genetic testing extends to family members and social contexts (Lim *et al.,* 2004). Family members are perceived as the best informants, but support in this process from health care is commonly requested (Pentz *et al.,* 2005). However, probands and mutation carriers are more likely take responsibility for informing family members and encouraging them to seek genetic counselling and undergo genetic testing (Peterson *et al.,* 2003). Hence, information dissemination in the family may be dependent on family member who initially seeks genetic counselling. Communication with families with hereditary cancer is complex, but the individual's certainty in relation to his/her own risk estimated influences the likelihood of informing others (Forrest *et al.,* 2003) and was also demonstrated in our study. A Dutch interview study in HNPCC has suggested that motivation to disclose information increases when cancer cases, especially if fatal, have occurred within the family (Mesters *et al.,* 2005). Some family members also mention situations where they have a need for communication aid. Gender differences have been identified with male family members focusing on age and relation to the individual to inform, whereas female members tend to select members perceived as best psychologically suited and the need for communication strategies have particularly been requested by male family members (Gaff *et al.,* 2005). As was identified in our study, individual experiences of learning about heredity may also affect subsequent disclosure patterns (Forrest *et al.,* 2003). Also, tense family relations and negative experiences from attempts to disclose information, may serve as barriers for information spread (Mesters *et al.,* 2005). Hence, assimilation of genetic knowledge in the family is related to family factors, individual factors, and the social context. Improved knowledge about family communication patterns may improve and facilitated information, perhaps through identification of key members perceived best suited to inform others and the genetic

counsellor may, through referring to these issues, facilitate the spread of information in the family.

The current experiences reflect the first HNPCC-families many of which have been identified through multiple affected individuals. The fact the only about half of the individuals invited to participate responded hereto may lead to selection bias, although gender was balanced and different ages and life situations were represented. Future HNPCC individuals will to a larger extent be born into families with genetic knowledge. It is therefore crucial to optimize information spread and communication within these families in order to ease obtaining and living with knowledge about HNPCC. Our findings suggests that the importance, primarily of experiential knowledge of cancer and family communication issues, need to be taken into account during genetic counselling since it may have an impact on the life situation for current as well as future members in families with hereditary cancer.

CONCLUSIONS

After the identification of the genetic background to HNPCC and optimized diagnostics with e.g. assessment of mismatch repair status and improved mutation detection screening techniques the time has now come to optimize the genetic counselling and psychosocial follow-up, including risk perception, coping possibilities, communication abilities, and psychological well-being. The common referral to the importance of experiential knowledge, the responsibilities for informing family members, and the impact on family relations suggest that these aspects should be taken into account during genetic counselling since focus on these question in the early phases of the genetic testing process could facilitate information spread within the family and optimally prepare family members for the impact that genetic knowledge may have on life.

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Table I Themes and subthemes for each individual carriers and non-carriers

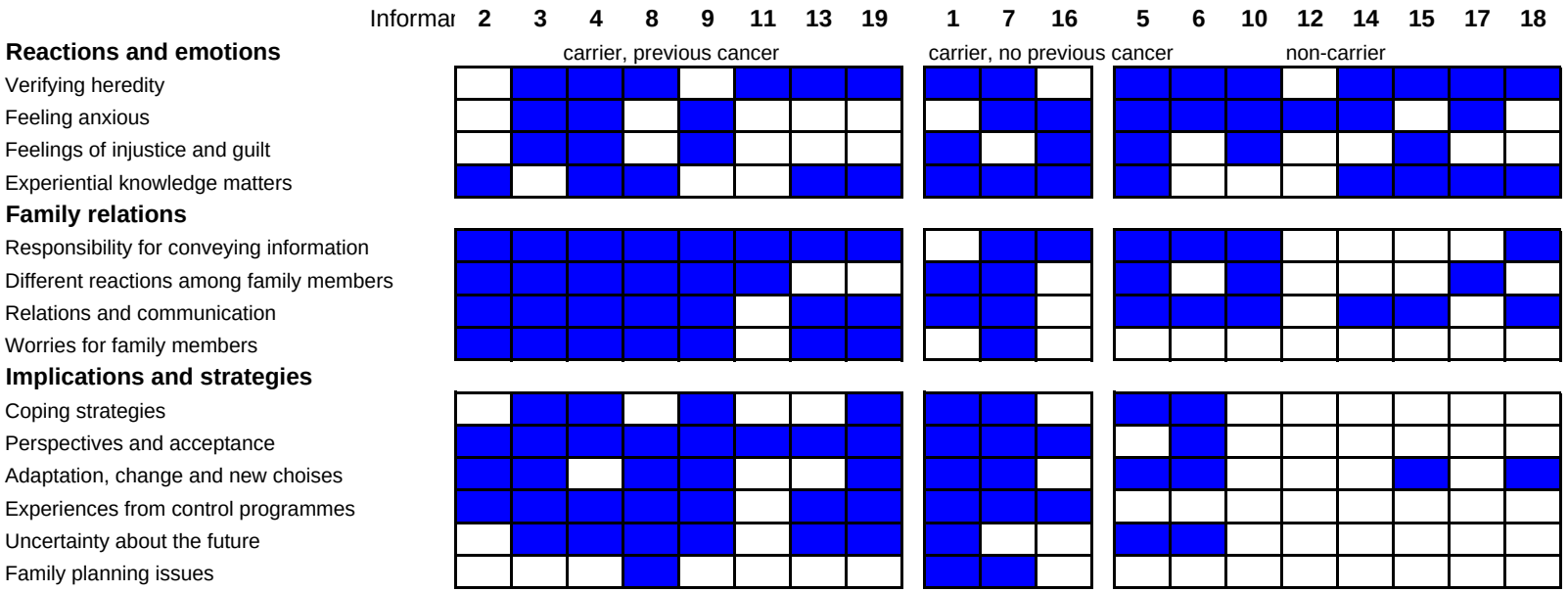

Table 2 Qoutes related to sub-themes (informant)

Reactions and emotions	***Carrier***	***Non-carrier***
Suspecting heredity	/…/yes, I almost suspected that. My father died when he was 42 – and now my paternal aunt is very sick…and there was another aunt and a cousin who died of cancer…so I assumed it was hereditary (3)	/…/both my grandmother and my mother have had cancer…so of course I've been thinking….In principle we knew it before going to the genetic counseling (14)
Symptoms and waiting create anxiety	/…/ there were many thoughts and reflections about what could happen…..what could happen to whom/…/(13) /…/when you get sick…even catching a cold or having headache you think "the time has come" (3) /…/when it pops up…when you get sick from other causes…not really feeling good and energetic….having a pain here or there…now something might be happening (16)	/…/it was a bit spooky before and a lot of thoughts crossed my mind (5) /…/ waiting for the result was the hardest time…that's when your thoughts are going in all directions…do I have it [the gene], or do I not. The closer it got to the day we had our appointment and could expect the result, the more I worried, the more nervous I was….I didn't quite know what to do or where to go. (5)
Feelings of injustice and guilt	/…/it should have been me instead…I'm the oldest and have done more things (informant 16)	/…/ So he [the brother] has three children…it feels unfair somehow, that it should strike his family, while we got out of it (5) /…/ it can happen when we watch TV….and we're reminded about cancer….now a teacher at school died from cancer….then we talk about it again/…/ (5)
Experiential knowledge matters	/…/ between my cancers operations…yes 10 years and 10 days. I cared for my brother at home until he died. The following year I had cancer and cared for my father until he died (19)	/…/ there has been cancer in our family, I knew that. My first memory of my grandfather was that he died of gastric cancer /…/ (15) /…/ when my mother developed cancer, gynecological cancer…she bled, but she was not operated on. I cared for my mother like you care for a baby, she was swollen (15) /…/so I opened the letter and read: 'Dr Ms NN, the test shows cancer'…there I sat and read it to my mother…It was terrible (15)
Coping strategies	/…/ I was completely convinced that I did not have it [the mutation]…it was very strong that gene, the genetic defect in our family. Almost 100% of those tested had it so I was convinced that I didn't have it (7)	/…/I think it's quite natural to envision the worst case and the best case scenarios…you move between the extremes in order to be prepared (6)
Family relations		
Responsible for conveying	My brother finds it harder to talk about these things [heredity] (8	/…/my son is fragile, he is very attached to me (5)

information	/…/it became my responsibility…my responsibility and I feel a bit guilty about these things, but somehow it's my responsibility…..you wish the best for your children in some way/…/ (13)	/…/ it wasn't difficult to tell…of course it was not as hard since it was positive [non-carrier]…but there were no questions or reactions, but teenagers have another focus like…'I have to go onto the internet now (6)
Different reactions among family members	NN [the name of the son] just sat there, staring, and didn't say much…or said nothing in fact…he just stared (4) /…/ some [meant her sister] family members argue. They claimed it [HNPCC in the family] was a lie/…/ (4)	My father didn't want to talk about it – there was complete silence about it (6)
Relations and communication	/…/ we had been rather close before as well….we have talked quite a lot…I think we talk more freely and personally than we did before/ (8) /…/ this Christmas it was just my mother and father…then sometimes they get very sad (10) /…/I can disturb and joke with my mother when I say "thank you for the defective gene"/…/ (1)	/…/ yes, we usually meet then…at Christmas…but we won't this year…yes, that's the way it is (9)
Worry for family members	/…/of course, you get worried and tense about what will happen to NN and NN [children from a previous marriage] when they undergo genetic testing (2) /…/ each child has a 50% risk of inheriting it so – I have three children….it's just 1.5 /…/ (3)	When we went there, my sister and I, I was really worried/…/ I thought about my children and grandchildren (14)
Implications for life		
Adaptation, change and perspectives	/…/ that's what I've said the whole time – that no matter how much I cry, heredity isn't going to go away. No matter how much I feel sorry for myself, it won't go away – and then there isn't any point doing it [crying] (7) What can you say…cry wolf?…no, I kept it to myself (16) I try to achieve my dreams…not look too far into the future…try to do things now instead of later (3)	/…/ so I would probably not have quit work the way I did – as an early retirement – I counted on not living past 65…/…/ (15)
Experience of surveillance programs	I think the colonoscopies are great. I prefer finding the cancer early than late (1) /…/it's such a small risk that I'll develop [cancer] again….they'll remove it before that/…/ (2)	

	/…/ of course you're more observant/…/ (9) /…/will they find something this time/…/ (1) /…/ you get regular colonoscopies so that cancer won't have the time to break out…the doctors know what they're looking for so you don't have to worry (3)	
Uncertainty about the future	[name of the husband] he can't turn on the washing machine or other things, I think about those practical things (4) Not daily uncertainty, but mainly when it's time for the examinations every second year (13) /…/one of my friends said "you never laugh nowadays"…yes, I rarely do. Before, I could laugh at small, silly things…but I can't do that now/…/ (4)	So I saw myself fighting against cancer (6)
Family planning issues	It reduced my chances of having children…and one has to consider whether you want children and then remove [the change] – but we thought it was better to get it done (1)	

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