
A Family-Centered Model for Sharing Genetic Risk

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Introduction

The successes of the Human Genome Project and the continuing advances of DNA technology have ushered in a new era of genomic science.¹ Investigators around the world are using genomic technologies to advance our fundamental understanding of biologic and physiologic mechanisms in humans and other species. The ability to sequence the entire human genome exponentially expands our ability to identify the contribution of genetic variation to disease risk and other phenotypic differences within the population.² Information derived from this research has the potential to contribute to disease prevention, disease prediction, and personalized treatment. These scientific advances, however, raise several ethical, legal, and social challenges. Many of these challenges, including personal and societal benefits and risks, and privacy and confidentiality, are mirrored in the current professional and public debate about the perceived conflict between personal autonomy, privacy, and confidentiality, and the potential value of sharing genomic information within the family.

A critical factor in enhancing the effectiveness of genetic risk information is to improve the distribution of the information to appropriate family members.³ The current policy is to consider the proband (the person undergoing genetic testing) as the gatekeeper of genetic information for the rest of the family. Because that genetic information may inform disease risks for other members of the family, it entails a significant responsibility. The proband must weigh the desire to protect family members from potential harm, negative emotional reactions, and loss of privacy with the opportunity to provide information that may offer health benefits associated with having information about their potential genetic risks. While several studies have demonstrated a high rate of sharing of genetic information with family members, they are mainly retrospective, report only on the perspective of the proband, not the family unit, and are focused on whether and when genetic information is communicated, not on the quality of the communication process.⁴ They fail to address many of the complexities of intra-familial communication, which can be highly selective and are influenced by many personal, familial, and social forces.⁵ When deciding with which relatives to share test results, probands may weigh such

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factors as the perceived vulnerability or resilience of the relative, their level of maturity, their coping skills, and their stage of life.⁶ Differences in cultural beliefs about health and illness can also affect the family's reaction to genetic knowledge.⁷ This level of complexity has only been heightened by the recent introduction of next generation sequencing (NGS) technologies, which can interrogate an individual's genome for a broad array of genomic information. NGS yields a large amount of data, much of which is currently uninformative.⁸ While the use of these technologies has great promise for improving health, at this time there is little guidance for their application to the clinical setting or their dissemination within families. Understanding the challenges families face in the context of genetic risk information requires a more systematic approach that takes into account the nature of family relationships, communication patterns, the dimensions of time and life stage, cultural beliefs, and the broader social network.⁹ This paper proposes the model of intra-familial communication most commonly used in the oncology setting, the family-centered model, as a systematic approach to explore the familial dynamic of genetic risk communication. The parallels are listed in Table 1.

A Family-Centered Approach to Cancer Care

Unlike the current uncertainty associated with the risks and benefits of sharing genetic information within the family, it is widely agreed that communication with and active involvement of the family is an important component of the delivery of medical care, especially in the setting of serious and life-threatening illnesses such as cancer. A family-centered approach is particularly applicable to the experience of a cancer diagnosis within a family because it not only acknowledges the support needs of patients, but also the needs of family members. These needs include information about the patient's illness, their role in care giving, anticipated changes in family role functioning, and their access to resources and support. Although cancer can vary in terms of presentation, course, and outcome, a cancer diagnosis in an individual is often met with fear, depression, anxiety, isolation, and uncertainty about the future and can create a crisis which can threaten the family with potential loss. Individuals facing a diagnosis of cancer often rely on their family to help them understand and adjust to their situation, and help them to navigate the decisions they are facing. Thus, it has become a central tenet of the model of cancer care that the family is an indispensable component of the patient's care team and

Table 1

Dimensions of the Family Systems Model

Model Characteristics	Application to Cancer	Application to Genetic Risk
Features of the Illness	Type of cancer	Type of genetic risk
	Mode of onset	Likelihood of developing disease
	Phase of life cycle at cancer diagnosis	Phase of life cycle at receipt of test results
	Availability of therapy	Availability of preventive or therapeutic options
	Course and severity of disease	Expected severity of disease
Time Phases of Illness	Awareness of symptoms	Awareness of familial risk
	Diagnostic work up	Information seeking
	Diagnosis	Testing decision
	Treatment decision	Adoption of screening, preventive options
	Treatment phase	Active surveillance
	Adaptation to chronic phase	Adaptation to genetic risk phase
	Terminal phase	Outcome(s) of genetic risk
Family System Variables	Coping with cancer skills	Coping with genetic risk skills
	Patterns of communication about cancer	Patterns of communication about genetic risk
	Caregiving skills	Family support structures
	Family experience with cancer	Family experience with genetic risk
	Quality of relationships	Quality of relationships
	Adoption of new family roles	Adoption of new risk identity
	Family values and beliefs about cancer	Family values and beliefs about genetics

support network. The family unit offers several potential strengths needed to adjust to an adverse event like a cancer diagnosis, including shared values and beliefs, a history of sharing information, established family roles, and a stable environment.

The recognition of the importance of family support in the care of cancer patients has led to the development of support networks for patients and their families.¹⁰ As an example, “family conferences,” during which the patient and his/her family are engaged with the health care team in a discussion of treatment options, patient preferences, and communication strategies, are a routine component of the care of a cancer patient.¹¹ These conferences provide an opportunity for the health care team to assess family values,

Family-centered models have been proposed to understand the complex interactions between the individual with cancer and the family.

family structure, and function, and thus can promote both patient and family well-being.¹² Even at the international level, cancer organizations are emphasizing the importance of involving family members in the care of a cancer patient, in identifying information and support needs,¹³ and in developing communication interventions to reduce family members burden, improve their knowledge and enhance their coping resources and their self-efficacy.¹⁴

Open communication about cancer-related issues with family members provides an opportunity to explore emotions, and has been associated with better psychosocial adjustment, higher relationship functioning, and better quality of life for both patients and their relatives.¹⁵ Active family involvement in the health care team can lead to an increased sense of self-efficacy for the family members in taking care of the patient and an opportunity for personal growth.¹⁶ Another benefit of engaged family members is the opportunity to explore the implications of the cancer patient’s diagnosis on their own cancer risks and the risk-reducing behaviors which they may adopt.¹⁷ A diagnosis of cancer in a family member has been shown to be a “teachable moment” which can motivate relatives to adopt screening and other health preventive behaviors.¹⁸

The Family Systems Illness Model

Family-centered models have been proposed to understand the complex interactions between the individual

with cancer and the family. Of these, the Family Systems Illness (FSI) model developed by John Rolland, clarifies the relationship between the trajectory of cancer and the family’s cancer experience.¹⁹ The FSI model considers the family as a complex and interactive social system with its own unique structure, developmental patterns, and communication style. The FSI model creates a useful framework for the evaluation of families dealing with cancer and other chronic illness and for the development of support interventions. The FSI model addresses *three dimensions*: (1) the features of the type of cancer, characterized by the mode of onset, the disease course, the disease outcome and the level of uncertainty about its trajectory; (2) the time phases of the cancer and the attendant psychosocial developmental tasks which the family must address; and (3) the family system variables, including its health beliefs, style of coping adaptation and creation of meaning which help to delineate the family’s strengths and weaknesses. This approach emphasizes individual and family development, the family’s legacy of coping with illness and loss, and family values and belief systems. It facilitates research about illness within the family by creating a framework within which to sort out the importance of different psychosocial variables across the course of an illness, and to guide the design of psycho-educational support systems for the family.²⁰

There are many similarities between the demands of a chronic illness such as cancer and the challenges of genetic susceptibility. A family member’s genetic risk information can have significant implications for other members of the family.²¹ In fact, unlike the involvement of the family with the cancer patient, which is often confined to the nuclear family, genetic risk can have even more widespread implications for the extended family. These similarities have led to the adaptation of the FSI model to the setting of genetic risk.²² The Family Systems Genetic Illness (FSGI) model provides a family-centered approach to the communication of genetic information within a family, maintains a family-centered orientation, and expands the scope to explore how families understand their genetic risks and how genetic risk information influences the family as a functional unit over time.²³ One of the strongest motivations cited for undergoing genetic testing is a sense of familial obligation, the possibility of benefit for other family members. In fact, it has been suggested that the decision to undergo testing be considered as a family matter from the outset, including an assessment of the educational and support needs not only of the proband, but of all family members.²⁴ Framing the communication of genetic

information as a family event and facilitating family communication about genetic results may provide a practical option for optimal feedback to family members.²⁵ Like the dimensions of the cancer experience, genetic risk can vary by the type of threat it invokes, the temporal course of coping with potential genetic risk, and the family system variables which have an impact upon the family's adaptation to the genetic risk. In an attempt to fill many of the gaps in understanding the communication of genetic risk within the family, this paper will review the parallels between the family experience of cancer and that of genetic risk as seen through the dimensions of the FSI model (see Table 1).

Dimensions of the Family Systems Illness Model

Features of a Cancer Diagnosis

The reality of dealing with a cancer diagnosis varies widely depending on the type of cancer, its mode and time of onset, the course and severity of the cancer, and anticipated outcomes. The diagnosis of cancer is often preceded by the experience of symptoms, leading to a period of uncertainty until the diagnosis is made. Alternatively, cancer may be found incidentally, a situation which creates an immediate crisis. In both cases, the family is called upon to deal with highly charged emotions and the need to be involved in role changes and critical decision making. The disease course of the cancer also varies and may involve long periods of stability, or the development of progressive disease. Outcomes of cancer vary widely from cure and the resumption of a normal life, to a progressive and fatal course, challenging the family to anticipate potential losses. Role changes within the family are affected by the timing of the diagnosis within the life cycle of the individual diagnosed with cancer. The onset of the cancer in childhood, early adulthood, middle age, or late adulthood will alter the needs of the patient, and also the unique challenges to family roles and relationships.²⁶

Features of Genetic Risk

The features of genetic risk also have many components. The likelihood of developing the condition based on the genetic mutation, or the penetrance of the disease, can range from a high to moderate to a low likelihood of clinical manifestations of the disease. This variability in penetrance can affect family members in terms of their level of uncertainty and the anticipation of the challenges with which they will be faced. The timing of the expected onset of the genetic condition within the life cycle of both the individual and family members is an important consideration.

The expected clinical severity of the disease, or the expected degree of disease burden for patients and their families, introduces practical and emotional considerations which must be faced. Genetic risk for disorders considered by some as socially undesirable, such as mental illness, may evoke fears of stigmatization and social alienation. The availability of effective preventive or therapeutic interventions will alter the expectations of the family and will impact their sense of anticipated loss, and their hopes for the future.²⁷

Time Phases of Cancer

As the course of cancer evolves over time, so do the distinct challenges and demands experienced by family members. During the initial diagnostic work up and ultimate cancer diagnosis, the family identity is threatened and family loss is anticipated. Concrete roles for family members involved with their relative with cancer are clearly recognized and include: (1) information gathering and recording; (2) providing advice on choice of treatment; (3) negotiating the logistics of treatment; and (4) providing emotional and physical support.²⁸ During the chronic phase, information is needed on lifestyle changes, rehabilitation, social re-entry, late effects of treatment, recurrences, and in some cases, end-of-life care.²⁹ Ongoing demands on the family's time, commitment, and emotional support can create family tension and burnout, and can threaten family cohesion. Conflict may arise between the patient and family members in terms of the choice of treatment aggressiveness. Family members, for example, may want their relative to continue on intense treatment regimens, while the patient is ready to adopt palliative care. The chronic phase can offer the family an opportunity to adjust to the ongoing threat of loss and personal risk and find meaning in the experience. In the terminal phase of cancer, the family must face the inevitable loss of a loved one. Decisions about a critically ill patient are likely to affect not just the patient but other family members whose lives, interests, and relationships are connected to the interests of the patient.³⁰ This phase offers the family the opportunity to realign relationships and incorporate the meaning of death into the family belief system.³¹

Time Phases of Genetic Risk

The FSGI model illustrates the experience of genetic risk across the phases of the individual and familial life cycle. Coping with genetic risk is also a dynamic process that can be conceptualized as involving several temporal phases. The first phase is the initial awareness of the potential for a genetic risk of disease, through learning about one's family history and/or

experiencing a diagnosis and the course of disease in a family member. The degree to which a family member may appreciate a pattern of illness within his/her family may determine the family member's reaction to consideration of their own genetic risk. The awareness phase may lead to information-seeking about genetic causes of disease, and active consideration and preparing for genetic testing. As the implications for one's own health begin to be appreciated, the individual must then process the nature and severity of the threat and consider actions to take to address the threat. An important consideration during this phase is the impact that the decision to pursue genetic testing may have on other family members, and how a positive genetic test would challenge the family

members to counsel family members about the practical and emotional demands of living with a genetic risk.

Family System Variables Related to a Cancer Diagnosis

A family's reaction to and involvement with a relative with cancer does not exist in a vacuum but rather is a reflection of the family legacy of organization, relationships, communication patterns, coping mechanisms, and belief systems that are passed down from generation to generation. Studying the involvement of family members in the care of a cancer patient illustrates the complexity of family structure and function. Not all family members are equally equipped to serve as caregivers. The nature of the relationship

A systematic appraisal of these time phases of genetic risk can identify critical periods of transition and facilitate the provision of targeted support systems. Understanding the dynamics of the process of living with genetic risk helps to identify the landmarks, transition points, and changing demands placed on the individual and the family, and could help clinicians to counsel family members about the practical and emotional demands of living with a genetic risk.

identity. As in the case of cancer treatment decisions, family members may exert pressure on a relative to undergo testing, while that relative may have hesitations and concerns. During the active testing phase, genetic test results are received and processed. The genetic test result allows individuals to better quantify their risk for disease, and confront decisions about screening, prevention options, sharing information with other family members, and adjusting to lifelong vulnerability. The timing and degree of communication of genetic test results to family members is often influenced by the perceived vulnerability and life situation of the relative(s).³² This phase is followed by a long-term adaptation phase during which the genetic information is acknowledged and integrated into the individual and family identity. This is a time of adjustment to the consequences of preventive actions which are adopted, and a time of vigilance for potential symptoms of disease.³³

A systematic appraisal of these time phases of genetic risk can identify critical periods of transition and facilitate the provision of targeted support systems.³⁴ Understanding the dynamics of the process of living with genetic risk helps to identify the landmarks, transition points, and changing demands placed on the individual and the family, and could help clini-

among family members, roles within the family, and the family's experience with cancer can all affect the nature and strength of support provided.³⁵ Coping with a cancer diagnosis in the family is a time when pre-existing family dysfunction can either accelerate, or be put aside for the benefit of the cancer patient. Family cohesion and flexibility have long been identified as important components to the adjustment to a health threat within the family. The ability to adapt to new roles within the family by both the patient and other family members is associated with positive family functioning and reduced levels of distress over the time course of a cancer diagnosis.³⁶ Family members' beliefs about the cause of the cancer, which vary by ethnicity, race, and religion, can also have implications for their causal attributions of the cancer and their own sense of vulnerability and perceived risk.³⁷

Family System Variables Related to Genetic Risk

When confronted with a genetic threat, individuals and families are challenged to attribute meaning to the threat while maintaining their sense of personal control. Individual coping mechanisms and family supports are often used to deal with the cognitive and emotional challenges posed by a genetic risk. An individual's genetic health beliefs are a composite of

personal and family experiences with illness, cultural beliefs, and values.³⁸ The communication of genetic risk information within the family has been linked to established patterns of communication, emotional and physical distance, life stage, and social roles.³⁹ Cultural norms are also an important determinant of the kind and degree of communication about genetic risk within the family and the wider community, and in decision making about genetic testing.⁴⁰ Similar to the situation of communication and decision making about a diagnosis of cancer, there is significant cultural variation in the balance between personal autonomy and the primacy of the family unit in the decision to communicate genetic information.⁴¹ Addressing disease risk based on genetic susceptibility may require family members to confront aspects of their family culture and to interact in new and unfamiliar ways. Knowledge of the individual and family variables related to coping with health risks will help to assess their anticipated strengths and weaknesses in coping with genetic risk, and to set realistic goals in dealing with the risk.⁴²

Cautions

Similar to the situation of a cancer diagnosis, the use of the FSGI model in the setting of genetics raises some questions that must be considered. The family member choosing to be tested may feel that the task of informing other family members is a burden, may feel uncomfortable in explaining genetic results, or may consider the sharing of test results as an invasion of his/her privacy. Family members approached to become involved in the experience may feel an undue pressure to participate in the family dialog about the genetic threat and in the family response to the information. Relatives who do not share the genetic risk may feel isolated or alienated from those who do. Some family members may lack the knowledge and/or resources to participate and may become marginalized within the family. The familial response to a genetic threat is often unpredictable and the receipt of information that was previously unknown or unwanted may compromise family relationships. Finally, the sharing of genetic information within the family must always respect the socio-cultural values within the family. When genetic risk information conflicts with family beliefs and cultural values, its importance may be minimized or may threaten family cohesion.

Summary

As the pace of genomic research continues to increase, the opportunity for genetic risk information to be communicated within families will grow. Individuals and family members will increasingly be faced with

decisions regarding the receipt and use of genetic and genomic information, and the risk management options available to them. Health care providers must be prepared to help family members understand the impact of genomic information in the context of their own life cycle, family structure, health beliefs, cultural norms, and family dynamics. Adopting a family-centered approach to both cancer care and genetic risk illustrates the many parallels between the two situations. It can provide insights gained from the family experience with cancer to that of genetic risk, and can provide a model to guide both the clinical approach to genetic risk, and a research agenda to gain a better understanding of how families cope with genetic risk. Both experiences can be characterized as a process that evolves over time with distinct phases and challenges to the family. Both involve uncertainty and anticipatory loss. Both have implications for the potential caregivers and support networks within the family and profound consequences for life cycle decisions. Both can be positively and negatively impacted by the nature of family relationships, cultural beliefs, resources, and the health care system in which the risk unfolds. Central to both situations is a need for open and ongoing communication within the family and with the larger social network. The success of this model in both situations requires effective and accessible educational tools about cancer and/or genetics for patients, family members, and their health care providers. It requires access to social support systems and to resources to deal with the challenges families face in these threatening situations. Appreciating the parallels between these two situations may provide a systematic approach with which to address the current controversy about the communication of genetic information within the family, and to facilitate a public debate about best ways to incorporate genetic information into the family unit.

Note

This work was supported in part by National Institutes of Health (NIH), National Human Genome Research Institute (NHGRI) #R21HG006594-01 and NIH, National Cancer Institute (NCI) and NHGRI #1-R01-CA154517 (Petersen, Koenig, Wolf, PIs). All views expressed are those of the author and do not necessarily reflect the views of NIH, NCI, or NHGRI.

Acknowledgment

The author thanks Donna Rathgeb for her assistance in article preparation.

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