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Family Communication of Genetic Risk: A Personalized Approach

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Abstract Genetic information is changing the practice of medicine by personalizing prevention and treatment decisions based on the genetic profile of the individual. The impact of personalized genetic-based medicine extends beyond the individual to include the family, where knowledge of a genetic predisposition to a disease condition has implications for individuals, their siblings and offspring, and for the identity of the family itself. The quality of the family communication process is important in assuring accuracy of risk information and in maintaining the cohesion of the family unit. However, there is increasing evidence that the communication of genetic risk information within families is complex, selective, incomplete, and often ineffective. This paper presents an overview of what is known about the strengths and weaknesses of the communication of genetic risk information within families and suggests a framework for both understanding the process of communication and guiding future counseling models.

Keywords Genomic medicine · Genetic testing · Genetic risk · Family communication · Family systems model

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Introduction

The success of the Human Genome Project has expanded our knowledge of the genetic contribution to health and disease and is influencing health-related decisions for a wide variety of conditions. Genetic information is changing the practice of medicine by personalizing prevention and treatment decisions based on the genetic profile of the individual. The impact of personalized genetic-based medicine extends beyond the individual to include the family, where knowledge of a genetic predisposition to a disease condition has implications for individuals, their siblings and offspring, and for the identity of the family itself. While much of the early literature has come from the field of oncology, an increasing number of non-oncologic adult and pediatric hereditary conditions are being described.

Clinical assays to detect the presence of mutations in genes responsible for hereditary syndromes are widely available, and individuals from families that appear to display a hereditary pattern of disease are increasingly offered genetic testing to determine their genetic susceptibility. The ability to not only interrogate an individual's DNA for a known susceptibility gene, but also 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 [1]. A critical factor in enhancing the effectiveness of genetic risk information is to understand the inheritance pattern within the family in order to identify the genetic risks to other family members and to address their risk-reducing needs [2]. Consistent with the traditional health-care model in which the focus is on the individual, the current policy in genetic testing is to consider the proband (the person undergoing genetic testing) as the gatekeeper of

genetic information for the rest of the family. Relatives with whom genetic information is shared may choose to pursue genetic risk counseling to clarify their own risk of disease, to take advantage of personalized screening and risk-reducing interventions, to inform reproductive decisions, and, if testing is negative, to avoid more intensive and invasive risk management strategies [3–5]. The quality of the family communication process is important in assuring accuracy of risk information and in maintaining the cohesion of the family unit. However, there is increasing evidence that the communication of genetic risk information within families is complex, selective, incomplete, and often ineffective [6, 7, 8•]. This paper presents an overview of what is known about the strengths and weaknesses of the communication of genetic risk information within families and suggests a framework for both understanding the process of communication and guiding future counseling models.

What Do We Know About Family Communication?

The communication of genetic risk information within the family is a complex behavior that involves a deliberative process in which the proband must first incorporate the information into his/her own risk identity, then consider who to tell, decide what information to disclose, and consider how and when to tell other family members [9]. During this process, the proband typically considers the relevance of the information for each family member and the potential benefits and risks for family members associated with receipt of the information [2]. The proband estimates the degree of receptivity of each relative, as well as their potential vulnerabilities, their level of maturity, their coping skills and stage of life, the anticipated benefit to the communication of the genetic information and the potential for harm [9]. Estimates of vulnerability and receptivity are more often related to the life situation and personality of the relative more than the test outcome itself [10]. The complexity of this process is reflected in the selective nature of sharing, or the decision not to share the information at all. Probands may fear negative consequences such as causing distress or anxiety, or having an adverse impact on their relationship [11–13]. Failure to share test results may be a function of poor communication skills within the family, or emotional distance from some relatives. Some probands may rely on other family members to share or disseminate the information within the family, e.g., relying on sisters to tell brothers [14]. In addition to variability in terms of with whom genetic test results are shared, probands also vary in how much information to disclose, and when they plan to share the information [15].

Most studies report that probands share some data about their test results with 75–80 % of their relatives [16–18]. Comprehension of genetics in general, and genetic tests in particular, and confidence in coping with genetic risk are positively associated with communication with the family [16]. Variables positively associated with being the recipient of genetic test results include being female, being a first-degree relative, and having a close relationship with the proband [3]. Once a first-degree relative is informed, there is often the expectation that that relative would inform their first-degree relatives, thus creating a cascade of communication [9].

A systematic review of the literature has identified four factors which motivate individual probands to communicate genetic risk within the family. These include: (1) the need to gain information from other family members to inform their own risk perception; (2) a sense of responsibility to keep family members informed; (3) a desire to promote risk-reducing behaviors among family members; and (4) an attempt to gain emotional support and advice for themselves [2]. The frequency with which genetic test results are shared with family members, however, does not necessarily correlate with the quality of the information shared. The extent of the information shared varies considerably within families and can be influenced by the age and the relationship of the family members. Lack of knowledge about the meaning of the genetic test result and lack of understanding of which family members would benefit from the knowledge can lead to incorrect disclosure [19]. The thoroughness of the information shared may vary from one relative to another based on the proband's interpretation of its significance for each relative. [20]. Factors such as the relative's age, gender, genetic relationship, stage of life and likely reaction to the information are considered [10]. Probands may limit the details of the information shared with their relatives to match their perceived level of readiness, and/or to protect relatives from fear or anxiety [21•]. A familial pattern of disease can constitute a health threat which may impact the stability of the family structure. Probands may be concerned about potential shifts in family relationship patterns leading to emotional distance, avoidance and denial once genetic information is available to relatives. Probands also struggle with finding the appropriate time to discuss genetic test results, especially with their young children [10]. It is not surprising that the attempt to communicate complex and often uncertain genetic information to at-risk relatives often results in poor comprehension and uncertainty regarding its relevance to them. It is also unreasonable to expect that a longstanding history of poor familial communication can be overcome around the issue of genetic risk [22]. As a result, despite relatively high reported levels of communication of genetic test results, a large percentage

of at-risk relatives do not receive adequate information. This is reflected in uniformly low rates of uptake of genetic risk assessment among informed relatives [23–25, 26•].

A Family System Model for Understanding and Improving the Communication Process

Thus, despite the growing availability of clinical genetic susceptibility testing and its incorporation into clinical practice, there is strong evidence that the communication process is often flawed, incomplete and can have negative consequences for the family unit. The promise of "Precision Genomic Medicine" will not be met without a better understanding of the actual process of communication within families. A Family System approach provides a framework integrating the biology of genetic risk information with the social and psychological context of individual and family resources, family relationships and sociocultural beliefs [27]. Using a Family System approach, the complexities associated with the introduction of genetic risk information into a family may be understood as an ongoing process which involves: (1) the nature of the genetic risk; (2) the individual characteristics of the proband; (3) the family structure, the evolving nature of family relationships, health-related family beliefs and values, and stages of life; and (4) the interactive social system both within the family and with the social environment [17, 28].

Nature of the Genetic Risk

The proband, tasked with the responsibility for conveying genetic risk information to family members, must grapple with the many complexities of the risk. Genetic risk can vary in terms of the certainty of the pattern of inheritance, the disease occurrence, the severity of the risk posed, the anticipated age of onset of the associated conditions(s), and the potential for disease prevention or successful treatment. With the recent shift to testing several genes in a panel rather than single gene testing, there has been a rapid increment in the discovery of variants of uncertain significance, for which risk estimates are not available, and incidental findings, or findings which are unexpected and do not correlate with the pattern of disease seen in the family. Germline mutations associated with disease occurrence vary widely in their penetrance or expression of the disease. The expression of the gene may be dependent on complex interactions with other genes and with environmental factors. The disease itself varies in terms of its natural course and severity, factors which will determine the anticipated burden for the family. The projected age and stage of life during which the disease are most likely to occur will shed light on the potential impact on life course tasks of affected individuals. For some inherited conditions, such as hereditary breast cancer, effective primary and secondary preventive strategies are available, while for others, such as Huntington's disease, there are no effective means of either preventing or treating the disease.

Individual Factors

The age and stage of life of the proband, their personal and/or familial experience with the disease, their comprehension of the nature of genetic mechanisms of risk, their perception of personal and familial risk and their communication and coping skills will naturally influence the effectiveness of their communication. High levels of numeracy and self-efficacy [29•] can facilitate effective information sharing. Women are more likely to assume the task of sharing genetic risk information within the family, even for diseases which are not gender based [8•]. Probands who have an authoritative position within the family and/or have a strong sense of responsibility for the well-being of their relatives may be more successful in communicating genetic risk information to other family members than those whose position is less dominant.

Family Level Factors

Families may differ in their organizational structure, roles, and intrafamilial levels of intimacy across diverse ethnic and cultural groups. The definition of "family" has become increasingly comprehensive and extends beyond the nuclear family to include non-biologic members of the functional family unit. An understanding of family level factors is critical for understanding the process of communication within families. Genetic testing is often described as a family affair which is determined by the family's experience with the disease and the family's pattern of interaction, beliefs, communication, and authority [10]. Family causal attributions of disease reflect the family's basic beliefs about personal control of their health. A failure to communicate the existence of a genetic threat may reflect an attempt to maintain equilibrium within the family. Deep-rooted beliefs about genetics may reveal fears about stigma or blame. Patterns of communication within the family vary based on the social and cultural concepts of family and kinship which may have evolved over generations [30]. Open and supportive communication patterns within families at risk for inherited conditions have been shown to improve the accuracy of risk perception on the part of the relative [6], to encourage relatives to seek genetic counseling for themselves and to promote risk-reducing behaviors [31, 32]. The impact of genetic risk information on the family varies depending on the stage of life of different family members, and the life cycle

challenges which they are facing. The receipt of genetic risk information within a family can activate openness, support and strengthening of family ties in response to a potential health threat. On the other hand, the information can restrict communication, alter family membership roles, reinforce patterns of silence or result in patterns of denial [21•].

Societal Level Factors

Societal level factors, often derived from ethnicity, race or religion, have a significant impact on the family's beliefs, values and health behaviors. These factors constitute cultural norms which influence beliefs about the value of science and medicine, the contribution of genetics to disease, willingness to undergo genetic testing, the nature of family ties and the exchange of experiences within the family. Cultural beliefs about kinship often dictate norms for communication, the value of privacy and autonomy, and the types of verbal vs. non-verbal messaging used in the family. Cultural variation exists regarding which family member should make decisions about the health of family members, and which medical interventions are acceptable. The importance of ethnocultural variables are reflected in different levels of concern about privacy, stigmatization and discrimination, as well as concerns about misuse of genetic information and lack of trust in health care providers [33].

Implications for Providers: A Personalized Approach

Conclusions

In summary, it has become clear that sharing genetic risk information within the family is a process with several stages. Compounding its complexity, it can be highly selective and is subject to many individual, familial, and sociocultural factors which impact the communication process. Some probands experience tension between their perceived responsibility for the health of family members and their own needs. Some find the task of sharing information burdensome, and some would prefer more active involvement of a genetic professional [26, 34]. Professional guidelines support a role for genetic practitioners in guiding and supporting patients to communicate relevant genetic information to their families [35, 36]. Since the advent of clinical genetic testing for hereditary susceptibility syndromes, the practice of genetic counseling has moved from a strictly non-directive process to one in which the counselor takes more active steps to promote communication within the family [37•]. This requires attention to family relationships in the dissemination of genetic information. Methods of professional support which have been proposed take the form of encouraging probands to share information with their family, providing educational materials and psychosocial tools to assist probands in sharing information, and making direct contact with relatives of probands, usually in the form of a letter explaining the relevance of the genetic information for their own health. Most of the tools developed focus on information content and do not address the importance of family dynamics and patterns of communication [37•]. In general, professionals feel a responsibility toward informing at-risk family members but are reluctant to breach privacy constraints or threaten family dynamics [38•]. In recognition of the barriers inherent in the communication of information with significant relevance to the health and well-being of family members, the Dutch Society for Clinical Genetics has developed a set of recommendations which acknowledge the primacy of the proband as the primary conduit of the information, but which allows for direct contact with family members when communication barriers cannot be overcome [39]. This option, however, is rarely adopted [37•].

The clinical and research application of genetic testing is fast outpacing our ability to provide appropriate support for individuals and families challenged with understanding and acting on the basis of genetic risk information. Since clinical genetic testing has become available over the past 20 years, there has been a growing appreciation of the complexities of familial communication of genetic results, but there has been little success in overcoming the identified barriers. A Family System approach underscores the need to understand the communication process within a family in the context of their unique life experience and may provide new insights into how to maximize the role of the genetic professional. Acknowledgement of the phases of adjustment to genetic risk may inform the timing of subsequent professional interventions [27]. The counseling team, which typically consists of a genetic counselor and/or other health professionals with expertise and experience in genetics, can be instrumental in coaching the proband to prepare for communication and in promoting family discussions. It is important for the counseling team to identify the unique roles of family members such as the caregiver or the health keeper role within the family. Anticipation by the counseling team of possible shifts in roles and identities and impending life cycle transitions as testing proceeds within a family can prepare family members for transitions in their relationships. Finally, health professionals need to be cognizant of the contribution of ethnocultural beliefs to the family's reaction to genetic risk information.

One way to operationalize a Family System approach within the counseling context is to conceptualize the

counseling process as a family affair from the outset by inviting not only the proband but also relatives chosen by the proband to participate [25]. By defining the family as the unit of counseling, this approach allows family members to consider the implications of the genetic testing process for the family members and to negotiate their obligations at the outset [40]. This could facilitate a fuller expansion of the family pedigree as well as identifying the family's experience with genetic issues, and their educational needs. The counseling team could clarify risks for individual family members and give validity to the information provided. It also gives the team an opportunity to explore family members' patterns of dealing with health threats and beliefs about genetics. Engaging the family as a unit to explore the process of communication within the family and its consequences and to assess unique personal and familial barriers to communication within the family may improve the effectiveness of the counseling [9, 18]. This approach may relieve the proband of some of the burden of transmitting sensitive genetic information and may help to solidify sources of social support within the family, but must be balanced with issues of privacy and confidentiality. In the coming decade the scope of genetic testing will most certainly continue to expand, both in the clinical and in the research setting. Big international databases containing genetic data and clinical outcome data are already making major contributions to our understanding of the genetic basis of many diseases. This enthusiasm for progress in genetics, however, must be accompanied by attention to the information needs of the individual family members from whom the data are derived and for whom the benefits must accrue.

Compliance with Ethics Guidelines

Disclosure Mary B. Daly declares that she has no conflict of interest.

Human and Animal Rights and Informed Consent This article does not contain any studies with human or animal subjects performed by the author.

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