

MEDICINE AND SOCIETY

How Should Decision Aids Be Used During Counseling to Help Patients Who Are “Genetically at Risk”?

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Abstract

People with genetic predispositions to disease are faced with uncertainty about whether, when, and to what extent an illness will actually develop. This prognostic uncertainty, combined with knowledge that preventative interventions (eg, risk-reducing surgeries for familial cancer syndromes) could significantly affect people’s lives, renders prevention decisions especially challenging. This article illuminates ethical questions about the use of decision aids for people with genetic predispositions and calls for approaching individual decisions in light of ongoing communication and reflection about a person’s life goals and values.

Everyone who is born holds dual citizenship, in the kingdom of the well and in the kingdom of the sick. Although we all prefer to use only the good passport, sooner or later each of us is obliged, at least for a spell, to identify ourselves as citizens of that other place.

Susan Sontag¹

Decision Making and Genetic Risk

As Sontag’s quote boldly illustrates, health and illness are generally seen as dichotomous categories: one is either sick or healthy. Yet the rapid development and implementation of genomic medicine is challenging this duality by increasing the presence of yet another type of “citizenship”—namely, for those who are “genetically at-risk.”² In dealing with this new category, health services face several challenges, including how to communicate complex information on individual and familial risk and how to support decision making on preventative treatment options.³

As people become increasingly aware of their genetic predispositions, more will face decisions about prevention efforts such as lifestyle changes and risk-reducing treatments (eg, surgeries for familial cancer syndromes). Prevention efforts might involve difficult trade-offs between quality of life and risk reduction, because in some

cases reducing risk of future ill-health is only possible with some sacrifice of current quality of life. Furthermore, in the case of genetic predispositions, people are faced with uncertainty about whether, when, and the extent to which an illness might actually develop. This prognostic uncertainty, combined with the fact that any preventative treatments might significantly affect people's lives, renders decision making about such interventions especially challenging. It is therefore all the more important that these decisions take into account people's norms, values, and life goals.^{4,5,6} This article examines the need for genetic counseling and decision aids for people with genetic predispositions and calls for innovation in both communication processes and decision aids in order to embed individual decisions in a broader process of ongoing reflection on personal life goals and values.

Genetic Counseling

Current decision-making supports in the context of genetic risk are proving inadequate. Genetic counselors help patients to assess their genetic risk and consider interventions in a nondirective way, which entails providing complete and unbiased information, refraining from revealing their own preferences,^{7,8} and helping align care with a patient's and family's values.⁹ Genetic counseling services are, however, in high demand, and care and treatment [discussions about genetic risk](#) are increasingly occurring outside of the genetic counselling setting, particularly in primary care,^{10,11} oncology,¹¹ and surgery.¹² Patients also discuss known genetic risks with a variety of health professionals—not all of whom are well informed about patients' stated goals and values.¹³ For example, in the case of patients with BRCA 1 and 2 familial cancer syndromes, the availability of multiple (preventative) treatment and screening options means that some patients with a mutation are cared for by a succession of health care professionals in general practice; clinical genetics; and screening, reproductive, and surgical services. Specialization and fragmentation of care can lead to piecemeal, incomplete, and conflicting information about care and treatment options.

In response to the growing need to support communication and decision making in the context of genetic risk in different clinical settings, a variety of decision aids have been developed. For example, in the case of familial cancer syndromes, decision aids have been developed for [diagnostic genetic testing](#),¹⁴ reproductive decisions,¹⁵ and preventative treatment decisions.^{16,17} Decisions about how to respond to genetic risk, however, pose ethical questions that call for innovation. In what follows, we discuss the goals and ethical challenges of using decision aids in the context of genetic risk.

Need for Innovating Decision Aids

Decision aids have been developed for "preference sensitive" decisions, for which the best option depends on a patient's perception of an optimal trade-off between harms and benefits.¹⁸ They have been designed to increase patient participation in decision making and to enhance rather than replace patient-professional communication.

Decision aids have 3 principle goals: to improve patient understanding of risks and benefits, to help patients clarify their values, and to help patients make decisions consistent with those values.¹⁸ Their development should be guided by decision science, which assumes that, in any given context, a best decision can be revealed—or at least approximated—by using a decision-making process or model.¹⁹ Risks that deserve ethical and clinical consideration include decontextualization, detachment, and fragmentation.

Decontextualization. A patient's familial, social, and cultural context are rarely considered in the development of genomic medicine decision aids, despite evidence suggesting that preference-sensitive decisions are influenced by patients' perceptions of successes or failures of approaches taken by other family members with the same condition,^{20,21,22} by perceptions of familial responsibility (eg, parents can be more inclined to choose aggressive preventative options),²³ and by attitudes and preferences of partners or members of their social networks.²⁴ Decision aids' underlying assumptions and value clarification methods (eg, utility theory in decision tree analysis)²⁵ could muddle a patient's decision-making process or be incompatible with a patient's normal decision-making style. Put differently, decision aids can impose a *system* on the decision-making process that alienates a patient from his or her *lifeworld* of shared social experience.²⁶

Detachment. Using decision aids to guide patients' decisions might be particularly tempting in situations in which it is impossible for clinicians to know whether a patient will develop a disease. This uncertainty could lead some clinicians to delegate to a decision aid the tasks of providing risk information, describing options, and clarifying values. The tendency to "retreat behind a technique"²⁷ in the face of ethically and emotionally difficult communication has been described in other areas of health care.^{28,29} Busy health care professionals might also consider the preferences- and values-clarification exercises associated with decision aids an adequate exploration of a patient's values. However, decision aids' effectiveness in elucidating patients' values remains unclear,^{25,30} and using them to replace rather than enhance discussion of a patient's values and preferences is clinically and ethically problematic.

Fragmentation. Technological advances in genetic sequencing mean that future generations could know their genetic predispositions earlier in life and thus might require support from clinicians to reflect on their life goals and to plan care.³¹ Potentially new developments, such as newborn whole genome sequencing, might result in people learning about genes of lesser penetrance (ie, lower risk of developing disease) and receiving polygenetic risk scores for a range of common diseases. As genomic medicine goes mainstream, the number of "patients" with knowledge of their genetic risks from a young age will increase. When people are aware from a young age of their genetic risks, they can experience pressure to anticipate and plan life events³² and future preventative interventions.²² Furthermore, many of these patients, as in the example of BRCA 1 and 2

mutation carriers, will see a host of health care professionals in relation to their genetic risk over the course of their life. Currently, however, decision aids are not developed to facilitate long-term planning or support continuity of care across different settings and with different health care professionals.

In summary, using decision aids in the process of decision making in the context of genetic risk involves considerable risks of decontextualization, detachment from ethically and emotionally difficult discussions, and fragmentation of decisions. These 3 risks are interrelated and reflect a need to understand and discuss a person's biography, context, and treatment trajectory and to anticipate care needs and provide continuity of care.

Dealing With Decision Aids' Risks

Although we cautiously encourage the use of decision aids in the context of genetic risk, we make the following recommendations to minimize the risks outlined above. To minimize decontextualization, we recommend embedding decision-aid use meaningfully into ongoing patient-clinician communications in which a patient's familial, social, and cultural context and other influences are explored and in which opportunities to include family members and loved ones in the decision-making process are presented. Genetic counselors have unique skills and expertise in familial-based counseling that enable them to assume responsibility for this change, although there is a role for nurse navigators, case managers, or even technological solutions such as patient pathway applications. To minimize some clinicians' detachment from ethically and emotionally complex discussions, we recommend meeting clinicians' unmet [genetics education needs](#)¹⁰ with training in how to communicate about genetic risk and in how to use decision aids appropriately. Finally, to minimize fragmentation of decisions among clinicians, the values and preferences a patient shares and explores with one health professional should be available to another.

Experiences of advance care planning for end-of-life care can inform how clinicians plan personalized care and treatment trajectories informed by patient preferences and values within the context of genetic risk. As is the case for successful advance care planning initiatives,³³ health professionals will need to be convinced of the importance of elucidating and respecting patient preferences and values and of ensuring that information is up-to-date and available in health record systems. We also recommend assessing decision aids' value as perceived by patients over the course of their care trajectory and not just assessing their effectiveness in facilitating comprehension or in improving procedural, psychological, or functional measures in the context of individual decisions. Innovations in the design and use of decision aids for people with genetic predispositions will require educating patients and clinicians about interventions and options from a life-course perspective and fostering carriers' reflection on their values, preferences, and life goals across the entire care trajectory.

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