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Genetic counseling in primary care: longitudinal, psychosocial issues in genetic diagnosis and counseling

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A wave of new interest in genetics accompanied the completion of the Human Genome project in 2000–2003. Ensuing discoveries are affecting medical understanding of rare and common conditions. Reports of new genetic tests promise the ability to diagnose and confront disease well before symptoms are manifested. Yet the effects of targeting prevention in this manner are largely unknown. Patients are bombarded by news accounts and direct-to-consumer advertising claiming the potential for genetics to protect health, cure previously incurable diseases, and enable control over the health prospects of future generations. "Salons" or Internet sites that offer to test individuals for several of 400 or more genetic predispositions are cropping up, raising the likelihood that patients will seek guidance from their personal physicians about whether they "have the gene" or should be tested for a genetic condition.

Often primary care physicians are not well prepared to handle patient inquiries about these new genetic tests and capabilities. They are caught between the popular media and patient curiosity on one hand and the lack of research about the clinical utility of these tests on the other. Despite the fact that primary care physicians counsel patients daily about healthy lifestyles and familial risk factors, they recognize a gap in their own education with regard to the burgeoning volume of new genetic information [1]. Likewise, most patients are unprepared for the myriad of psychosocial issues that arise when genetic testing is undertaken. In many settings, limited access to

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geneticists and genetic counselors also highlights the urgent need for primary care physicians to become better versed in genetic counseling [2]. Beginning the process of genetic counseling, helping patients to decide when to pursue genetic information, arranging for referral if necessary, and helping patients assimilate and act on information from genetics professionals are indispensable elements of primary care in the twenty-first century.

How does genetic counseling fit into primary care?

The "new genetics" includes a rapidly increasing number of known single-gene disorders and growing knowledge of the genetic contributions to multifactorial diseases commonly seen in primary care [3]. Primary care clinicians have an essential role as part of the team providing genetic services, including genetic risk assessment, counseling, and ongoing health care for patients with inherited susceptibility to disease This role is likely to be different than the role played by a consultant, especially because primary care is often longitudinal. Primary care clinicians, among others, need education and information resources if they are to counsel patients seeking genetic information [1,4,5]. Educational initiatives, supporting primary care faculty development and interaction with the genetics community, not only have provided resources, but also have clarified that primary care clinicians are well suited to counsel patients and families in ways that complement the methods of genetics consultants [6,7].

Patient perspectives

From the patient's perspective, the primary care physician is viewed as the first source of genetic information and individual risk assessment. Patients often initially hear about genetic testing for certain disorders from the popular media. When surveyed, they wish to receive more detailed information and counseling about genetics from their primary care physicians [8,9]. Patients want their primary care physician not only to facilitate informed decision making about genetic testing (eg, for susceptibility to colon cancer), but also to counsel them about and oversee the implementation of individualized preventive health care [9]. An underlying process of primary care is to encourage patients and families to engage in risk-reducing behaviors, including screening and healthy habits. This process can be modified to explicitly include family history and other genetic information [10]. Primary care clinicians can address patient needs for information and prioritize education about diseases that "run in the family."

Physician perspectives

From the physician's perspective, genetic counseling fits well into the tasks of primary care. Most common "genetic" diseases represent an

interplay between genotypic and environmental influences [11]. Common diseases, such as diabetes, cancer, heart disease, and psychiatric illness, presumably result from the combined effects of environment, lifestyle choices, and many different genetic variations, each generally of low penetrance [3,10]. Until more recently, however, most physicians have not looked at genetics in terms of common disease. Early genetic tests dealt with rare, highly penetrant mutations (eg, Huntington's disease) in which presence of the mutation undoubtedly led to disease. Although this "genetics as destiny" formulation is true for some unusual genetic conditions, most genetic variations are of low penetrance, giving a probability but not certainty of developing disease and a chance that genetically guided preventive measures could be effective. If one employs a broader definition of genetics to include common disease, it is not difficult to envision a role for genetic counseling in primary care. A primary care physician with a broad understanding of a patient's behavioral risk factors coupled with knowledge of the family history would be well positioned to identify familial risk, refer selected patients for genetic consultation and testing, and suggest preventive measures for many common disorders with a hereditary component.

Primary care physicians, who are used to prioritizing among competing clinical demands [12], can place genetic counseling into the broader context of a patient's overall health status. A 50-year-old woman may present to her physician considering the possibility of *BRCA1/2* testing because of a family history of breast cancer. On review of her family and social history, the physician finds that an elderly aunt recently died of breast cancer, but both of her parents died of complications of diabetes and the patient continues to smoke. Here the question of familial breast cancer risk more appropriately may take a back seat to the strong family history of diabetes [13]. While still addressing the patient's concerns, the primary care physician can help to prioritize which disease processes pose the greatest risks. The physician can reassure this patient that because she is at average familial risk for breast cancer, she should have a yearly mammogram and clinical breast examination, as recommended for all women her age, but she does not need referral for genetic testing. In addition, the physician can explore the patient's responses to the family history of diabetes and her motivation for preventive measures focused on preventing diabetes and its complications, including smoking cessation, exercise, and healthy diet. Genetic counseling in primary care ideally would become part of an overall health risk assessment that translates into targeted behavioral and medical interventions.

The primary care physician's knowledge of a patient's broader context prepares the physician to integrate psychosocial issues into any discussion of genetic information. The determination that a patient has a genetic or familial susceptibility to disease may raise a variety of psychosocial issues. Individuals may worry about stigmatization, insurance eligibility, and

employment discrimination if their risk is disclosed [14]. Individuals who are asymptomatic may exhibit features of ''uncertain wellness'' manifested by increased anxiety over being told they have a potential for a life-changing disease. Others may be galvanized to institute preventive behavioral change or may exhibit a fatalistic attitude and increase their risky lifestyle behaviors. Many of these potential reactions to the determination of genetic risk have not been studied except in selected families with highly penetrant hereditary conditions [15,16]. The ramifications for patients who undergo risk assessment and the ways in which people make sense of family medical history are not yet clearly understood. An in-depth knowledge of the patient's past reactions to illness and of significant life events, including family members' illnesses or deaths, may be helpful as physician and patient decide when and how to pursue genetic information. Primary care physicians draw on their knowledge of the patient and family, anticipating and addressing patients' emotional reactions to genetic counseling and testing.

Another component of primary care that lends itself well to genetic counseling is the longitudinal relationship inherent in the care of patients. There may be particular times in the life cycle when genetic information becomes particularly pertinent (eg, when young adults contemplate childbearing, when a new diagnosis is made, or when a family member dies). Genetic disease often requires that an enormous amount of information be conveyed in a meaningful way to patients; this cannot always occur in one or two traditional genetic counseling sessions [17]. In a longitudinal relationship, information can be presented when the individual is best able to understand and make use of it. Primary care could allow for ongoing patient education and counseling over time, adapted to individual needs, style of information processing, and changing emotional states. A longitudinal clinician-patient relationship allows for a flexible timetable for the provision of information, one that focuses on the patient and is not limited to a single set of meetings with a consultant.

The final aspect of primary care well suited to genetic counseling is that primary care physicians often treat families [18]. Because genetic information often has repercussions for everyone within a family, a working knowledge of the family is invaluable when counseling about genetic testing. Issues of autonomy, privacy, and embarrassment versus disclosure of potentially lifesaving information can be brought up with patients in the context of their own family's experiences and ways of communicating. This discussion could ease potential problems with relaying information to affected persons. Knowledge of the family also could equip physicians for family-based approaches to managing genetic risk [19]. Primary care physicians may be aware of key, influential family members who spread information throughout the family and whose opinions greatly affect their relatives' health care and lifestyle decisions [20,21].

Finally, it is expected that increasing need and limited access to trained genetic professionals will necessitate that some forms of genetic counseling be provided within primary care. Currently, there are 25 genetic counseling training programs in the United States, which have produced about 2000 working genetic counselors [22,23]. The expected need for genetic counseling in implementing large-scale, population-based genetic screening for just one disorder, such as cystic fibrosis, could completely overwhelm all the available genetic counseling services [24,25]. The primary care community will be called on to do a good job with genetic counseling for some conditions and to devise workable consultation relationships with genetics professionals for situations needing their expertise [25]. Widespread carrier screening for mutations associated with cystic fibrosis would not seem feasible unless primary care clinicians offer the test, provide basic pretest counseling, and explain test results to patients who are not cystic fibrosis carriers (see article on prenatal screening in this issue). Practitioners in various settings would have different ways of working with genetics professionals, however, to ensure that cystic fibrosis mutation carriers, and especially couples who both test positive for cystic fibrosis mutations, receive adequate genetic counseling and understand the meaning of their test results [25].

What is genetic counseling?

This section describes, in simplified form, the tasks accomplished when a patient consults a genetics professional regarding genetic testing. This is a highly individualized endeavor that attempts to provide meaningful information for the patient and family in question. A fundamental component of genetic counseling is educational, attempting to present a large amount of complex information in comprehensible ways that allow a patient to make an informed decision. At all times, the counselor strives to protect patient autonomy in the decision-making process. Full disclosure of relevant information is considered the best way to respect patient autonomy [22].

Pretest counseling

Pretest counseling precedes genetic testing. In this session, the genetic counselor interviews patient, assembling a large amount of information about family medical history, ethnicity, patient's concerns and expectations of genetic testing, and information about insurance coverage. Pretest counseling involves the following:

- Construction of a family history and pedigree showing all medical problems
- Analysis of the family history for potentially inherited and congenital diseases

- Verification of the family history (often a process that is pursued after the first counseling session, by contacting relatives and obtaining medical records)
- Assessment and interpretation of the risk for occurrences (or recurrence) of genetic conditions in the family
- Discussion of the nature of the conditions, including the contribution of heredity
- Discussion of the options available to reduce risks, including available testing; sometimes this involves planning which family member should be tested first, if possible, to give the most informative results
- Brief discussion of the meaning of various possible results of genetic testing
- Presentation of risks and benefits of each option, with careful attention to patient comprehension
- Assistance in selecting the option most appropriate for the person consulting, with consideration for the family
- Provision of supportive counseling or referral to community resources when appropriate
- Coordination of testing, when indicated
- Composition of a summary letter to the patient or referring physician, which documents the counseling session and outlines the plan of care

Post-test counseling

Post-test counseling involves providing the results of genetic testing to the person tested. The counselor presents a full explanation of test results and implications for further testing and management. Information is discussed regarding the implications for family members. Finally, post-test counseling involves emotional support, including referral to mental health professionals when indicated and often to condition-specific advocacy and self-help groups. Generally the role of the consulting genetic counselor ends at this post-test session, although individuals can arrange follow-up visits as new questions arise or as more family members request information about the implications of the tests for them.

Preparing patients for referral

Primary care physicians can do much to select and prepare patients referred for genetic counseling. Not everyone who seeks genetic information or has questions about family medical history needs a referral to a genetic counselor. Primary care physicians can "screen" and determine who is most likely to benefit from a session with a genetic counselor. Ideally the primary care physician should perform a preliminary assessment of the family history and other risk factors for genetic disease (see article on family history in this issue). Determination of risk is crucial because typically most

individuals who choose to pursue genetic testing are driven more by their subjective sense of risk than their actual risk status [16]. To help with this risk assessment, a primary care physician may be in contact with a geneticist or may refer to published resources pertaining to specific disease categories (eg, see resources described in the article on hereditary cancer susceptibility in this issue). It may be helpful for the referring physician to outline the genetic consultation process and tell the patient what types of information will be needed by the geneticist. With the patient's consent, pertinent records should be sent.

It is important, if feasible before referral, for the primary care clinician to help the patient clarify the value of pursuing genetic information at this time, as follows:

- What will the patient do with the information presented?
- What are the potential repercussions of possible test results to patient and family?
- What are the possible actions, if any, that can be taken to manage the genetic risk?
- What impact will the information have on the patient's and family's "tasks" in this stage of the life cycle?
- Will medical information or test results be communicated to other family members who may be at risk for a genetic disorder, and, if so, how will this be accomplished?
- How urgent in this case is the process of finding out and responding to genetic information?

Nondirective counseling

To complete the discussion about how the work of genetic counselors complements that of primary care clinicians, one must look at the ideal of nondirectiveness. A defining aspect of traditional genetic counseling is the "nondirective" approach to the counseling session. Nondirectiveness is a way to protect patient autonomy. In part, it is a response to the eugenics movement and other oppressive social engineering schemes of the past century (see the article on eugenics elsewhere in this issue). It also has its roots in the psychology of self-actualization [26]. Nondirective counseling promotes the idea that the genetic counselor is a neutral advisor acting solely as educator about the meaning and use of genetic information for the individual [27]. Advances in genetics, especially the availability of genetic tests to detect disease susceptibility at a stage when disease can be prevented, have prompted some to question whether the nondirective approach is too restrictive [28]. Such discussions also have arisen with regard to how the results of predictive tests would affect family members who did not seek genetic information [28]. The notion of nondirectiveness often has been misconstrued [29]. The ideal of pure nondirectiveness contrasts with other models of the physician-patient relationship prevalent in primary care.

It is difficult for a primary care physician, as an advocate for the patient's health and well-being, to take an entirely neutral stance in conveying potentially life-altering information. Alternatives to nondirective counseling, such as the patient-centered approach [30] and shared decision making, are familiar to generalist practitioners and likely to be appropriate in many situations when helping patients to decide whether to pursue genetic information [6,31]. The model is not one of a language interpreter who simply "translates" arcane genetic science into comprehensible language. The patient must make his or her own decision, but the physician can guide the patient in recognizing and addressing the medical and emotional impact of the information and considering the consequences of any given plan of action. Although respect for the patient's final choice is crucial, leaving the patient "at sea" during the decision-making process may impair his or her ability to arrive at a fully reasoned conclusion. The primary care physician "worries" with the patient and for the family, providing guidance based on eliciting their values, needs, and beliefs about health, aiding the patient in making the best possible decision for himself or herself.

Model for genetic counseling in primary care

After describing a rationale for locating some genetic counseling functions in primary care and reviewing the tasks that genetic counselors aim to accomplish in consultation, a framework can be developed for integrating genetic counseling in primary care with that provided by consultants.

Family history

The first step in genetic assessment involves the acquisition and interpretation of accurate family medical histories. The family history encapsulates information about shared behavioral, environmental, and genetic factors in families, often useful in primary care. Family history has become a prominent tool for risk assessment and possible intervention in genetic disease [32,33]. An excellent method of recording and communicating family history is to display it graphically as a genogram (pedigree) [34–36]. The genogram is the cornerstone for understanding family structure, patterns of illness including genetic inheritance, and biopsychosocial dynamics in families [37]. Methods for recording and using a family pedigree or genogram are described in detail in the article by Bennett in this issue.

Clinical utility of genetic testing

When the family history or other clinical information indicates further genetic assessment, the clinician and patient can discuss whether pursuing it makes sense at this time. Primary care clinicians are especially concerned

with the clinical utility of genetic testing [6]. *Clinical utility* is defined as the usefulness of a particular test in a specific clinical situation [10]. Patient, test, and intrinsic disease factors drive clinical utility. Tests of high clinical utility are likely to be of more benefit for patients. Conversely, tests of lower clinical utility may be of no clinical value or outright harmful to patients [38]. Characteristics of tests with higher clinical utility include the following:

- The test has high predictive power. The test would reduce greatly uncertainty about development of the disease in this patient or family. In general, this is more likely to be true for genetic alterations with high penetrance. That is, the presence of the mutation generally means the patient will have the disease (eg, multiple endocrine neoplasia, Huntington's disease, inborn errors of metabolism).
- The disease in question has highly effective and acceptable pre-symptomatic intervention (ie, hemochromatosis, phenylketonuria, glutaric aciduria).
- The pretest probability for detecting a disease-causing mutation is fairly high (this is where the genogram becomes an invaluable tool.) The pretest probability increases based on the following familial factors:

  High number of affected individuals
  Multiple generations affected
  Disease characterized by early age at diagnosis
  Recognized clustering of findings associated with the genetic disease (eg, ovarian and breast cancers for *BRCA* mutations; uterine and colon cancers for *HNPCC*)
  Ethnicity in which the prevalence for genetic disease is increased (ie, *BRCA1/2* in Ashkenazi Jews; maple syrup urine disease in Mennonites)

- The test has high perceived usefulness to the patient and family. A person with no offspring or siblings at risk for familial breast cancer may perceive genetic testing differently than someone with numerous offspring and siblings.

Psychological implications

After a determination of the clinical utility of genetic testing, which focuses primarily on the characteristics of the disease and test in question, an exploration of the psychological implications of genetic testing for the patient is warranted. It is necessary to assess the patient's ability to understand the intricacies of genetic testing. Often individuals have heard from family members and other sources about the pros and cons of genetic testing. It is important to "reality-test" with the patient about the meaning of a positive or negative result. What are the reasons for seeking testing in the first place,and does the patient have a realistic view of how testing can and cannot achieve their stated goals?

A discussion also should include what the patient will do with the information when genetic testing is completed. Genetic testing has implications far beyond the individual being tested, and a discussion of these implications should occur *before* someone undergoes testing. Has the individual discussed with other family members that he or she is considering testing? Has the individual thought about who might disclose information if it is found that family members are at risk for a certain mutation? How might other persons in the family react to hearing about health information that they did not seek or want?

Genetic counseling can have broad implications for persons' perceived health status regardless of whether they undergo genetic testing [39]. Would a positive result make them think of themselves as "sick" even though they do not yet express any symptoms of disease? Do they understand that most mutations have incomplete penetrance, meaning that a positive test does not mean that they will ever manifest the disease? Do they understand that for some disorders clinicians do not yet know of all the mutations and a negative test result may not exempt them from risk? Would testing positive motivate individuals and families to work on preventive measures, or would it make them feel fatalistic, leading to lifestyle choices that increase risk for a certain disorder [15,16]? How would individuals feel if they test positive for a mutation that they have passed to their children?

Finally, a discussion about the prospect of "misattributed paternity" should precede genetic testing [40]. According to some studies, in families that undergo genetic testing the prospect of finding out that children are not from the presumed biologic father may be 10%.

When to refer

When the primary care physician has explored the medical and psychological implications of genetic testing with the patient, a decision about referral should be made. Referral is warranted in the following situations, among others:

- The primary care physician or patient wants more specific information about the genetic condition in question.
- The clinical utility for testing in the particular case is high.
- The individual has a basic understanding of what genetic testing can and cannot do and of the potential psychological consequences of genetic testing.
- The individual has discussed the issue of testing with other involved family members.
- The individual desires genetic consultation after available options for prevention, treatment, and managing reproductive risk with this disorder have been outlined.

Example

A 49-year-old, female patient of a family physician came to the emergency department with a 1-week history of shortness of breath and chest pain. The emergency physician ordered a spiral CT scan of the chest. The result came back negative for pulmonary embolism but showed a large mass eroding into the mediastinum from the anterior chest wall. On physical examination, the physician noted a large, fungating, and bleeding tumor of the left breast that obviously had been present for some time. The patient was admitted to the family practice inpatient service. Fig. 1 shows a genogram that was generated during her inpatient admission.

On further discussion about the breast mass, the patient confided that she had noticed bleeding from the breast for at least several months. Review of outpatient records revealed that she had declined clinical breast examinations and mammography. Outpatient records had an extensive family history but no mention of prior breast cancer in the family.

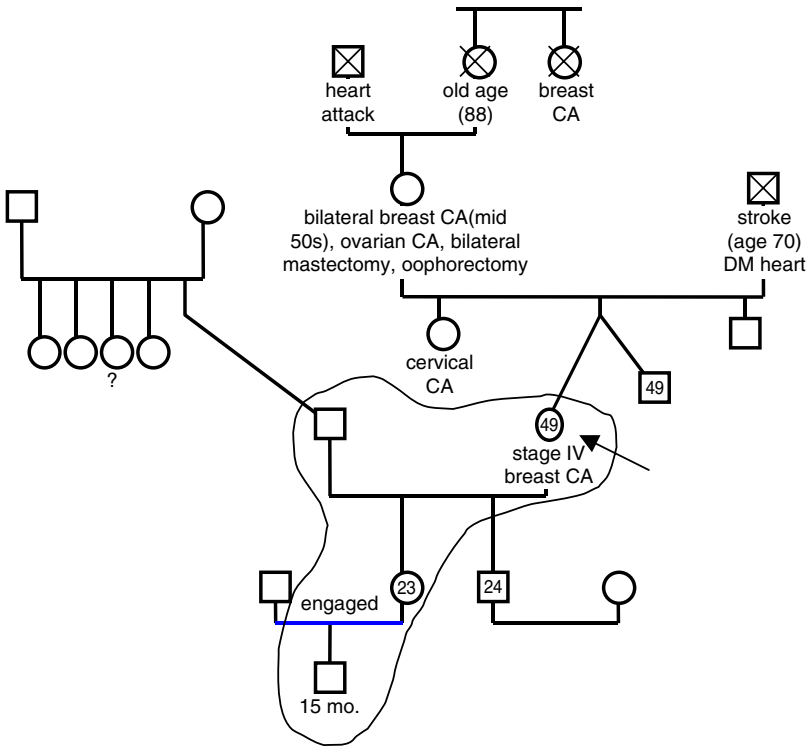

Fig. 1. Genogram for a woman who is a candidate for *BRCA1/2* mutation testing. Arrow indicates the identified patient. Enclosed line indicates household members. CA, cancer; DM, diabetes mellitus.

The woman reported that her husband had a long history of alcoholism and that she considered herself the "keystone" of the family. Although she obviously had a serious medical problem, she did not feel that she could afford to be sick at this time. She felt that she was holding together a family in crisis.

The patient's 23-year-old daughter had a conflicted relationship with her father. The father did not approve of the daughter's boyfriend, who was the father of her child, the only grandchild in the family. The patient reported a close relationship with her mother, who was diagnosed with bilateral breast cancer and ovarian cancer in her 50s and is still living today. The patient could not provide any medical information about the paternal side of the family.

The genogram presents several medical and psychological issues. From a medical standpoint, it appears that the woman is at a high risk for having a *BRCA1/2* mutation. Multiple generations are affected by breast and ovarian cancer. The cancers generally are diagnosed at a young age. There is an individual with bilateral breast cancer. From a medical standpoint, this woman has a high pretest probability for a *BRCA1/2* mutation.

The genogram presents psychological issues as well. The patient identifies herself as the "glue" that holds the family system together. Yet from a genetic perspective there is a strong possibility that she is a carrier of a mutation that could be destructive, if not fatal. It might be difficult for her to think of undergoing testing because of the possibility of having passed a mutation on to her daughter. The thought of conferring increased risk on a member of the family may cause distress that interferes with her ability to think through appropriately all the consequences of testing. If the patient were to be tested and a cancer-associated mutation found, testing for her daughter could have some beneficial outcomes. If the daughter tested negative for this mutation, she would not need to worry about being at markedly increased risk and could continue routine preventive care. If the daughter tested positive, she could begin an intensive program of surveillance and could consider medical or surgical prophylaxis that potentially would prevent the development of cancer (see article by Culler in this issue). Based on the genogram and the patient's perception of her role in the family system, it would make sense to present the prospect of genetic testing as another way for her to help and care for her family.

Finally, the stage of the life cycle at which genetic testing is being considered is likely to affect the patient's and family's responses greatly. In this example, the patient may not have long survival from an advanced cancer. Although her test would be most informative for her daughter's cancer risk, anticipatory grief and denial may complicate the process of deciding whether to undergo mutation testing. If resolving this grief and going through genetic testing simultaneously with cancer treatment is unrealistic, it may be feasible for the patient to choose to "bank" a specimen that could be used later for DNA testing, with the patient's instructions for who should have access to it after her own death.

Longitudinal aspects

The longitudinal nature of the physician-patient relationship in primary care affords several benefits when patients present with genetic issues. Genetic counseling need not end at the typical ''post-test'' counseling session. Perceptions about risk and its potential reduction can influence greatly a patient's willingness to undertake behavioral change. Because these perceptions may change over time, the primary care physician is well positioned to provide ongoing monitoring and additional advice or intervention as necessary. In contrast to genetic consultants in many settings, the primary care physician is ideally positioned to continue the dialogue on genetic issues for as long as necessary after test results are obtained.

Utility changes over time

Clinical utility is a moving target. As new research becomes available and better treatments are developed, the clinical utility of a particular test needs to be reassessed [37]. A test that someone decided to forgo 2 years ago may now for various reasons be of higher clinical utility. A disease such as Alzheimer's disease, which lacks effective presymptomatic intervention, may be amenable to some as yet discovered future treatment, such as immunization. When and if that happens, predictive genetic testing (eg, genotyping for APOe) may take on higher clinical utility. Likewise the patient's perception of utility also may change over time (see subsequent section on life cycle triggers), and it is important to reassess continually what patients are thinking with regard to genetic testing.

Long-term sequelae

At least one study suggests that persons undergoing cancer genetic counseling who refuse testing can experience unintended psychological sequelae for 6 months after the counseling session [38]. It can be important to monitor patients and ''check in'' with them on a regular basis to see how they are processing and coping with the assessment of risk presented to them at a pretest counseling session. Patients who decide to undergo testing also can have significant psychological reactions to testing that manifest at a later date. These persons benefit from longitudinal care as well [39,41].

Life cycle triggers

Various family life cycle triggers present the opportunity to revisit genetic information with patients over the life span [41], as follows:

- Someone with inherited disease susceptibility now becomes ill.
- Someone is about to marry and start a family.
- Someone's children grow into adulthood and are considering having children of their own.
- More family members are diagnosed with a particular disorder.

Summary

As medical science evolves, so must the primary care physician's role in patient care. Primary care physicians can provide the initial genetic risk assessment and explore with patients the clinical utility for genetic testing in specific situations. Patients look to their primary care physicians for expertise about their families and about prevention of diseases that "run in the family." The emphasis on continuity relationships and the contact with multiple family members inherent in primary care provide the physician ongoing opportunities to communicate genetic information effectively. When a patient is referred for formal genetic counseling, the primary care physician sometimes can serve as a resource in working through complex family dynamics. Almost always, after genetic consultation, the primary care clinician has an ongoing role in follow-up, treatment, or prevention. It can be particularly important to revisit and update genetic information in response to individual and family life events. Genetic information is just one aspect of the whole person, whom the primary care physician treats within the context of a longitudinal relationship.

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