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Am J Nurs. Author manuscript; available in PMC 2009 May 06.

Published in final edited form as:
Am J Nurs. 2009 April ; 109(4): 65–69. doi:10.1097/01.NAJ.0000348607.31983.6e.

Disclosure of Genetic Information Within Families:

How nurses can facilitate family communication

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When someone has or is at risk for a hereditary condition, the entire family is affected. Communication within families about such risk is important—it's central to independent decision making—but it can be difficult: whether someone can disclose such information, even to family members, depends on many factors. In order to help patients and families make decisions about disclosing information regarding genetic risk, and avoid undo distress in the process, nurses must understand the many issues that affect families when genetics is discussed.

TO DISCLOSE OR NOT TO DISCLOSE?

The fields of genetics and genomics have seen enormous growth in recent years. (*Genetics* is the study of single genes and their effects, and *genomics* is the study of all of the genetic material of a species—the genome—and the relations among different genomes; because some diseases or conditions are caused by interactions among more than one gene or among genes and the environment, some scientists consider the study of such cases to be part of the field of genomics.1) As a result, the disclosure within families of the results of genetic testing and other information on hereditary risk has emerged as an important area of research. Many studies focus on genetic conditions such as Huntington's disease,2, 3 cystic fibrosis,4-6 hemophilia A,7 and hereditary breast and ovarian cancer.8-10 Whether people with these diseases disclose genetic information to family members depends on what they perceive to be the risks and benefits of doing so, how close they feel to family members, their sense of responsibility to family members, how they expect the family members to react to such information, their level of certainty about their own risk, and their own emotional readiness to disclose the information.2, 4, 7, 8

The most common reasons cited for disclosure within a family include a perceived need or obligation to disclose,2, 4, 7, 8, 11 the fear that the relative carries a reproductive risk,5, 6 a close social relationship with the relative,2, 5, 7, 8 the need for support,5, 11, 12 a feeling of responsibility toward the younger generation,2, 13 and a perceived need to retrieve information about familial risk.6 Those at risk for or diagnosed with hereditary breast and ovarian cancer also cite their desires to encourage genetic testing, obtain advice about medical decisions, provide information about risk and possible discrimination,11 and be open about the cancer diagnosis.8

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The reasons given for not disclosing within a family include a lack of closeness,2, 4-6, 8, 9, 11 a desire to protect family members from troubling information,2, 3, 8, 11, 13 a perception that the relative has a lower risk of passing on the disorder because she or he is unmarried or childless or plans to have no additional children,4, 5 test results that were uninformative or negative for specific mutations,11, 13 a relative's youth or immaturity,5, 9, 13 family disagreements,6, 8 assumptions that information had been imparted by other family members,8, 9 an antiabortion stance,4 a lack of “openness” regarding cancer,8 and happenstance (it “never came up”).9 Nondisclosure decisions also may be influenced by guilt or anxiety.11

WHAT TO DISCLOSE?

Huntington's disease is an autosomal condition, as is hereditary breast and ovarian cancer (separate cancers related to the same mutations and therefore grouped together), meaning that the mutation causing them is transmitted by autosomes, or nonsex chromosomes, and they are dominant traits, which means that they may be expressed even in those who receive the mutation from only one parent. When one parent carries an autosomal dominant mutation, the chance of passing it on to their offspring is 50% for each pregnancy. But there are substantial differences between Huntington's disease and hereditary breast and ovarian cancer, in terms of genetics and treatment.

HIPAA and Genetic Disclosure

Does the law affect families?

The privacy rule of the Health Insurance Portability and Accountability Act of 1996 (HIPAA) went into effect on April 14, 2003. The rule created new requirements for health care providers, third-party payers, and health care clearinghouses to protect the privacy and security of health information, including genetic information, that could identify a person.

A person's decision not to disclose her or his risk of developing a genetic condition to a family member may pose a dilemma for the health care professional, who has to choose between ethical obligations to inform the at-risk party1, 2 and legal requirements to respect and protect the patient's privacy. The potential for harm to family members often plays a large role in the decision. For example, in rare cases, the use of a certain surgical anesthesia can cause death in people with the mutation that disposes them to malignant hyperthermia. Likewise, strenuous exercise can bring on sudden death in people with hypertrophic cardiomyopathy. In such cases, clinicians should carefully explain the reasons for sharing information with family members, strongly encourage patients to notify at-risk relatives, and offer to help develop a plan for notifying them. When patients choose not to notify family members, clinicians typically respect the decision. There are situations, however, in which the clinician may consider overriding the patient's preferences because the family member is judged to be in danger of serious or immediate harm or there is a high likelihood that a dangerous condition will occur that can be either prevented or adequately treated with early recognition. Clinicians who believe it is necessary to override a patient's wish regarding the notification of family members, should consider consulting an ethics committee or legal council.3 In most situations, though, the risk of serious harm associated with nondisclosure is not clearly immediate. For instance, if a woman has tested positive for the BRCA1 or BRCA2 mutation but doesn't want to upset her two sisters by sharing that information with them, the need for disclosure isn't clearly urgent. Each sister has a one in two chance of carrying the same mutation. It would be understandable for the patient to try to learn more about what the

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mutation means for her and her children and to give careful thought to whether and how she'll share her test results with her sisters.

Clinicians can best help family members by clearly informing patients who test positive for genetic conditions or the mutations that cause them about the risks faced by their family members, discussing the value of disclosure and offering assistance with it.2, 4-6

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The appearance of Huntington's disease symptoms, their severity, and the age of onset are highly variable, depending on the number of times a DNA segment known as the CAG (cytosine–adenine–guanine) trinucleotide repeats. Currently, no treatment is available to stop the progressive symptoms and life-shortening effects of Huntington's disease.

Those who test positive for mutations in tumor suppressor genes BRCA1 or BRCA2 are at increased lifetime risk for developing breast and ovarian cancer, but not all will do so. They are given recommendations for surveillance that include mammography and a clinical breast examination, avoidance of behaviors that increase the risk of cancer, and when appropriate, chemoprevention or prophylactic surgery.

Research has suggested that those at risk for hereditary breast and ovarian cancer tend to discuss with family members the meaning of test results, the need for others to be tested, the obligations to children, treatment options, and possible effects on reproductive choices. By contrast, people at risk for Huntington's disease tend not to disclose information beyond test results unless asked by family members.3

Among women at risk for hereditary breast and ovarian cancer or X-linked hemophilia A, disclosure of genetic information is largely influenced by sex, with a greater number disclosing to female than to male relatives, reflecting a common misperception that these conditions concern only women.7-11 Many people do not realize that men—especially those with a relevant family history—can develop breast cancer and would, like women, benefit from genetic counseling and testing.

When people choose to disclose to family members is often influenced by major life events such as graduation, marriage, pregnancy and childbirth, or the manifestation of symptoms in children.2, 3, 6 Those with Huntington's disease and hereditary breast or ovarian cancer may view disclosure as a “process of telling” rather than an act2 and plan the “right” time to disclose information to family members, based on how they assess their own and their relatives' readiness.3 When such events arise, they heighten everyone's awareness of the ramifications surrounding the hereditary nature of the illness. According to one study of 21

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Australians with one of four genetic conditions, “At such times, the significance of the distinctly genetic aspect of their illness adds a dimension to their experience that may be lacking at other times. … Individuals may be prompted to embark on a quest to track risk through the family.”6

DISCLOSURE TO CHILDREN

Is there a right time to give children information on hereditary conditions? Concerns include the potential for psychological harm; the child's lack of autonomy in deciding whether to undergo genetic testing (if, for instance, a parent's desires outweigh the child's); and the possibility that the child won't be able to understand the full implications of the information.14 A common view among those with a heritable disease is that parents are responsible for imparting genetic information to their children15 when they feel their children are old enough to understand3, 4 or when it's time for the children to make important decisions.2

In our own research, which focuses on information management in families in which a child has a genetic condition, parents report disclosing information when they feel their child is developmentally ready, in the hopes of helping the child adapt to the condition.16, 17 They describe disclosure as a process that continues through childhood and adolescence. When only one child in the family is known to be affected, parents and health care professionals must consider what to tell siblings who may or may not have the same condition. In such cases, parents may have markedly differing concerns about disclosure to the siblings and to the affected child.

Our research further suggests that both the nature of the condition and the child's developmental stage influence parents' willingness to disclose genetic information to that child. As children pass through developmental stages, their parents' attitudes toward communication about genetic conditions often change. We offer the following timeline as a guide to help health care professionals better understand what, during clinic visits, might be on the minds of parents whose children have or are at risk for a genetic condition.

Diagnosis

Most states now screen newborns for a variety of genetic conditions. Often the results are reported to the infant's provider or a facility contact person by telephone, followed by a letter or fax, reporting the abnormal results and recommendations. Frequently, parents are informed of the results by telephone or letter from the provider and referred to specialists for consultation and additional testing. The disclosing health care professional, however, needs to follow up to make sure that the parents understand the implications of the diagnosis and the importance of follow-up and possible treatments. The diagnosis might imply that at least one parent carries or has the condition and that other family members may be carriers or at risk. The health care professional can encourage discussion on how to share information with family members and refer them to reliable Web sites and other sources of information.

Resources for Families

Families who want more information about genetic disclosure can visit the following online resources:

- Sharing Genetic Test Results with your Family, at Cancer.net: www.cancer.net/patient/Learning+About+Cancer/Genetics/Sharing+Genetic+Test+Results+With+Your+Family

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- Talking to your Family About Risk, at GeneticHealth.com: www.genetichealth.com/Resources_Talking_to_Your_Family_About_Risk.shtml
- Family Ties: Communicating Genetic Risk with Family Members, at the VCU Massey Cancer Center: www.massey.vcu.edu/treatment/?pid=2204

The preschool stage brings a dawning awareness of symptoms, treatments, or physical differences from peers and may prompt children with genetic conditions to ask parents or health care professionals why they have to take medicine, eat certain foods, or undergo treatment. Even within a family, parents have different ideas about what information should be shared with the child, how it should be presented, and who should do the sharing. In providing guidance, the health care professional must recognize parents' concerns. Some parents prefer to inform their children through a health care professional, either directly (by having the professional discuss the subject with the child) or indirectly (by letting the child overhear what is said during clinic visits).

The school-age years usher in a deeper awareness of family relations, and a child may question parents about the implications of a genetic condition for themselves and others in the family. As they age, and when science and sex education are introduced in school, these children may become more aware of their own reproductive risks.

Adolescence brings with it a new awareness of self, reproductive capacity, and the larger world, which has important implications for sharing information on genetics. As the child develops an understanding of genetic conditions, parents may wish to discuss the teen's specific reproductive risk. When the adolescent begins considering leaving home or starting a family, questions about the genetic condition might change or become more numerous or pressing. For example, older adolescents with autosomal recessive conditions may have more specific questions about their chances of having a child who has or is a carrier of the condition.

IMPLICATIONS FOR NURSES

To assist patients in disclosing to family members, clinicians can ask about familial relationships and how family members tend to discuss health and illness.

Those who feel confident in disclosing genetic information often encourage other family members to learn more about their own risks and to consider genetic testing, if appropriate. Nurses can counsel patients on different means of contacting relatives: in person; by telephone, e-mail, or letter; through another trusted relative; or by bringing family members with them when results are given. (See *HIPAA and Genetic Disclosure.*)

When a person contacts family members, she or he should have up-to-date information on the condition, genetic risk, and genetic testing. To reach a number of family members at the same time, especially distant relatives or family members who are estranged, a patient may write a family letter, which allows the patient time to formulate thoughts and gives relatives information in writing that they can review and keep for future reference. Family members can be encouraged to talk with clinicians and meet with a genetics expert to discuss risks, benefits, and limitations of genetic testing.

As children and adolescents mature, their ability to understand and their communication skills increase. Clinicians should tailor the information they provide to parents and children according to the children's developmental stages and the parents' preferences, actively assisting parents in discussing the condition and its inheritance with children.17

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To the parent of a seven-year-old, a nurse may simply ask, “Have you ever talked with Jake about his cystic fibrosis?” If the parent indicates that the child doesn't seem ready to discuss any aspects of the condition besides treatment, provide reassurance and advice: “Other parents of young children feel the same as you do. When Jake is ready for more information, he'll start asking questions. You know your own child best. If he is attentive when you talk to him about it or has questions, then he's ready for more information.” End the conversation by emphasizing your availability for continued support: “You can always give me a call if you or Jake have any other questions. Here are a couple of pamphlets about talking to your children about cystic fibrosis. Other parents have found them helpful.”

As the children approach adolescence, science classes may prompt them to initiate a conversation with their parents. At that point they may be ready for more detailed explanations, informational pamphlets, or a meeting with a genetics expert.

Parents often wonder whether their healthy children should have genetic testing. Clinicians can advise parents that, unless the hereditary condition affects children, most experts recommend allowing children to make this decision independently, when they are old enough to understand the implications of testing, after being fully informed of the pros and cons.18, 19 Children who should be tested for genetic conditions include those with a family history of hereditary colon cancer, multiple endocrine neoplasia types 2A and 2B, and von Hippel–Lindau syndrome. A positive test would signal the need to begin screening or monitoring, and possibly treating, at a young age.20, 21

The Essential Nursing Competencies and Curricula Guidelines for Genetics and Genomics established by a nursing consensus panel,22, 23 and the Genetics–Genomics Nursing: Scope and Standards of Practice,24 developed by the International Society of Nurses in Genetics and the American Nurses Association, provide direction to nurses for delivering competent nursing care regarding genetic and genomic issues. Nurses and other health care professionals have opportunities to work with patients and their families to understand what is involved in genetic disclosure and to provide them with accurate, appropriate, and current information, resources, and referrals to facilitate disclosure decisions. (See *Resources for Families.*)

Acknowledgments

The research reported in this article is supported by the National Institutes of Health (NIH), National Human Genome Research Institute (NHGRI), Ethical, Legal and Social Implications Research Program, Grant no. R01 HG02036. Manuscript preparation was supported by the Center for Reducing Risks in Vulnerable Populations, Grant no. P30 NR009014, National Institute of Nursing Research, NIH.

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