The Importance of Family Health History
Your Patients' and Your Own

As much as we might anticipate that the most cutting-edge scientific approaches to health assessment would afford the most reliable indicators for guiding health care decisions, that isn't always the case. Researchers at the Genomic Medicine Institute's Center for Personalized Genetic Healthcare at the Cleveland Clinic recently reported that when an assessment of family health history was compared to evaluations from commercially available personal genome screening test kits, family health history consistently served as the superior tool for predicting an individual's risk of developing certain diseases.[1,2] More specifically, this study of 22 women with breast cancer and 22 men with prostate cancer was designed to determine the accuracy and concordance of risk assessment for 3 common forms of cancer (breast, prostate, colon) when risk was predicted by family history versus by the genomic screening kits marketed by one vendor (Navigenics, Foster City, California). Each assessment method was used to sort patients into one of 3 risk categories: general population, moderate risk, or high risk. Some of the more notable findings from this research include the following[2]:

- For all 3 types of cancer, family history and genetic testing risk assessments agreed on the subjects' risk categories an average of only 40% of the time.[1]
- Overall concordance between the 2 methods of risk assessment was low ($K < 0.15$) for all 3 types of cancer. Concordance between these methods of risk appraisal varied from 59% for breast cancer to 41% for prostate cancer and only 39% for colon cancer.
- Of the 22 women with breast cancer, 10 had high hereditary breast cancer risk based on family history, yet genetic screening only identified 1 as high risk ($K = 0.12$). Similarly for colon cancer, none of the 9 individuals with high hereditary risk identified via family history were categorized as high risk by genomic screening. Likewise for prostate cancer, none of the 3 subjects with high hereditary risk based on family history were designated as high risk by personal genomic screening. Overall, of the 22 patients with high hereditary risk as determined by family history, their personal genetic screening tests identified only 1 subject as high risk.[2] If commercially prepared genetic screening kits marketed directly to the public neglect to identify hereditary risks, individuals may be erroneously reassured by false negative results and conclude that they have no reason to seek further health care screening or follow-up.[3]
- Conversely, genomic testing classified 8 men at a moderate/high risk for prostate cancer, while their family history categorized them with risk no higher than the general population, suggesting that false positives may also be a concern with genomic screening.[2,3] The researchers' explanation for this finding is that the genomic screening products marketed to consumers typically examine a

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wide array of DNA variants, including some not universally recognized, yet may not include genetic mutations associated with specific disorders.[3]

The researchers concluded that a family history and personalized genomic screening may one day become complementary tools for gauging cancer risk,[2] but at least for now, family health history remains "the gold standard in personal disease risk assessment."[1(page 2)] That conclusion is important for both critical care nurses' assessments and counseling of critically ill patients as well as for monitoring our own personal and family health risks.

Definition of Family Health History

As *Critical Care Nurse* readers know, a family health history (also referred to as a family medical history, a family history, or medical family tree) is a compilation of relevant information about medical conditions affecting a patient and his/her close family members.[4,5] It represents an essential component of a patient's medical history, typically obtained at the time of admission to a health care facility as one component of a comprehensive patient assessment. Two features that distinguishes a family health history from a patient's medical history are that a family history extends beyond enumeration of the patient's major health problems to identify those experienced by each member of that patient's immediate family and its indication of the nature of the relationships among family members.[6]

Relevance

Critical care physicians, nurses, and many other health care professionals routinely record patients' family medical history to obtain a more inclusive depiction of who the patient is and the health issues most likely to affect him/her owing to genetic or familial influences. Because family members share not only patients' genetic material but often their environment, habits, and lifestyle choices, recording a family history may facilitate identification of patients at heightened risk for certain disorders because it reflects both that person's genes and other shared risk factors.[4] Following decades of public health education regarding the risk factors for a number of common chronic diseases, even school children now know that family history is one of the most important risk factors in the development of health problems such as heart disease and cancer. When research reveals that a family health history is more effective than genetic testing in predicting a patient's risk for a number of these disorders, then the relative value of this traditional mode of patient assessment needs to be illuminated: "Family history risk assessment has been shown to be one of the most effective tools for predicting what diseases an individual may be at risk for developing."[2(page 1)]

Advantages

In addition to its value as an effective and accurate marker of risk for developing a number of chronic health disorders, a family health history offers a number of other practical advantages for patients, family members, and their health care providers: the assessment device is simple to construct, easy to gather and update information for, inexpensive to prepare, readily accessible, immediately useful to family and care providers, and clinically applicable in actual patient care settings.[1]

Who to Include as "Family"

Although the term *family* may be narrowly or broadly interpreted with or without contemporary notions of what constitutes a family, within the context of a family health history, the scope of family members typically embraces 3 generations of relatives by birth[7]: you, your children, your siblings, your parents, your maternal and paternal grandparents, and your maternal and paternal aunts and uncles.

Information to Record

The information sought for inclusion in a family health history primarily relates to major medical disorders, diseases, and/or conditions associated with an hereditary or familial component and the person's age when the disorder was first diagnosed.[5,7] Examples of these disorders are listed in Table 1.

Other environmental and lifestyle factors in a person's history such as alcohol addiction, drug addiction, smoking, diet, exercise, and weight problems may also affect inherited risks.[3] The National Society of Genetic Counselors[8] also lists birth defects, learning problems, mental retardation, and vision or hearing losses at a young age as relevant data. For some families, intermixing of different racial or ethnic backgrounds may be pertinent as well as living in or travelling to foreign countries. For every family member who is deceased, important data points are their age at the time of death,

Table 1 Major medical diseases, disorders or conditions usually included in family health history[5,7]

Arthritis
Asthma
Cancer (especially primary site)
Congenital defects
Diabetes mellitus
Disabilities
Gastrointestinal disorders
Heart disease, including heart failure, rhythm, and conduction disorders
Hypertension
Hypercholesteremia
Mental/behavioral health problems
Miscarriage, stillbirth, birth defects, or infertility
Peripheral vascular disease
Pulmonary disorders
Renal disease
Stroke

Table 2 Applications of family health history information[5,7]

Distinguish among risks for different diseases
Identify patterns of disorders relevant to a specific patient's health
Determine which diagnostic tests are most appropriate to order
Distinguish whether a patient or his/her close relatives should have a specific genetic test
Plan the type and frequency of screening tests tailored to a patient's needs
Refer patients or family members for genetic testing and/or counseling
Identify other family members who may need to be notified regarding their risk of developing a specific disease
Assess the probability of a patient passing certain conditions on to their children
Diagnose a condition that might not otherwise have been considered
Recommend therapies or modifications in diet, exercise, weight, personal habits, or other lifestyle changes to diminish the risk of disease
Distinguish factors related to diet, exercise, weight, personal habits, or other lifestyle changes that may increase the risk of disease
Review with patient or family members to clarify or correct erroneous interpretations of risk based on family history[9]

the cause(s) of death, and, when known, the person's age at the onset of that cause.[7]

It is extremely important that these data be gathered from *both* sides of the family. In a recent study by Rubinstein et al,[9] 2500 healthy women indicated they knew much less about the health history of their paternal family and that they tended to discount the risk of breast cancer when its family history existed on their paternal versus maternal side. For unexplained reasons, it appeared as though these women perceived that their maternal health history was more relevant to their risk than their paternal pedigree.

How Information Is Used

Health care professionals may apply the information contained in a family health history in a number of important ways (Table 2). Charis Eng, MD, PhD, Founding Director and geneticist at the Genomic Medicine Institute's Center for Personalized Healthcare, relates that there are 3 red flags when reviewing a family health history[10]:

- Determining whether more than one family member has developed the same condition or disorder over the years
- Identifying how closely the affected individuals are related to each other
- Identifying the ages of onset when that disorder or condition arose in each affected individual

When a pattern in this information is evidenced, it may be time to speak with one's physician and, if warranted, to a genetic counselor. Relevant patterns might include early age for onset of cancer or heart disease (eg, an acute myocardial infarction in a 32-year-old), several generations of family members affected by the same disorder, or multiple primary cancers in the same individual versus a single primary site with metastasis.[4]

Findings from Rubinstein et al's[9] report suggest that another potential use for family history data is that it affords an opportunity for health care professionals to clarify or correct erroneous interpretations of risk related to family history. Study results found that beyond their parents, significantly more women reported cancer in their maternal relatives compared to their paternal lineage and that a family history of cancer on the paternal side was associated with a lower perceived risk for breast cancer than when the cancer history existed on the maternal side. Findings suggest these women may have a limited awareness and/or understanding that their paternal family history is equally relevant as their

Table 3 Strategies for soliciting family medical history[5]

- **Explain the purpose for requesting this information.** Clarify that you are creating a document that can help members of your family determine whether they have a history of certain diseases or health conditions that run in the family.
- **Offer to share copies of the medical history.** Be sure to relate that once the family medical history is prepared, it can be shared with other family members, who may then provide it to their personal health care providers.
- **Provide multiple alternatives for responding to questions.** Some family members may not be Internet- or even computer-savvy, so online or e-mailed inquiries may not produce replies. Others may prefer informal, personal touches via face-to-face conversations in their home or telephone calls at mutually convenient times rather than mailed questionnaires. Some of the most senior members of the family may need to be interviewed in person.
- **Pose questions clearly and directly.** Use brief and direct questions.
- **Maintain respect for confidentiality.** Some relatives may not wish to share any of this information, whereas others may offer limited amounts or restrictions on its use.
- **Listen well.** As relatives describe their health problems, be vigilant to listen without making judgments or offering personal commentary.

maternal side. In addition, this sample of women perceived the influence of age and parenthood in directions opposite to scientific evidence—eg, perceiving that risk of cancer decreases (rather than increases) with age.[9]

Other patterns that may suggest heightened risk for developing related disorders include diseases that do not usually affect persons of that gender (eg, a male who develops breast cancer) and specific combinations of disorders within the family (eg, diabetes and peripheral arterial disease or breast and ovarian cancers).[6] In families that demonstrate these patterns, family health history represents a significant risk factor for development of these conditions.

Approaches to Gather Information

The approaches most frequently suggested for soliciting family medical history information are by asking questions and talking with family members at natural family gatherings such as holidays, reunions, or even funerals.[7] Many families are fortunate to have members who enjoy tracking and researching family genealogy, who may have some of the demographic data already organized. Other families may include one or more members who assume the role of family historian and could serve as valuable resources for locating this information. Public records such as death certificates may also be used for this purpose.

Although some families may welcome the opportunity to participate in developing its medical history, others may not. In addition, individual family members' interest or willingness to contribute to this effort may vary. Attempts at gathering this information, then, may be met with enthusiasm, indifference, reluctance, or even outright refusal to discuss this information. Some family members may not see the value of this endeavor, others may consider these issues as highly private, and for yet others, discussions related to deceased family members may precipitate a cascade of sorrowful, painful memories laden with guilt, regret, shame, anger, or other emotions they would prefer remain in the past. Given these possibilities, some strategies recommended for soliciting this information are listed in Table 3.

Documenting Family History

Documentation of a family health history is important for establishing, updating, correcting, maintaining, accessing, and sharing this information among and between family members as well as with various health care professionals. Another form of documentation can be a family medical tree, drawn to visually summarize these findings in graphic form.[10] The US Surgeon General's "My Family Health Portrait" is an online tool developed specifically for these purposes; it is available at http://familyhistory.hhs.gov.[7] An alternate Web site (www.hhs.gov/familyhistory) provides background information on this tool. Free print versions are also available by calling the Health Resources and Services Administration Information Center at (888) ASK-HRSA.[7]

In a recent study[11] of 35 veterans (71% male, 78% older than 50 years), investigators assessed satisfaction with using this tool and found that all participants rated the tool as very useful and that most (88%) viewed its functionality, look, navigation, and performance favorably. The total time that participants needed to complete the tool averaged 25 minutes. The study also identified several barriers to use this tool: lack of knowledge regarding

Table 4 Family health history resources

American Society of Human Genetics "Talk Health History Campaign" offers a comprehensive array of family health history information, online tools, and resources for consumers and health care practitioners at **www.talkhealthhistory.org**.

CDC, Office of Genomics and Disease Prevention, Family History Public Health Initiative (**www.cdc.gov/genomics/famhistory/index.htm**), offers valuable information and FAQs about family health history and provides fact sheets, case studies, news articles, multimedia presentations, and disease-specific information.

CDC's Family Healthware software (**www.cdc.gov/genomics/famhistory/famhx.htm**), an Internet-based research tool that determines familial risk for 6 diseases (diabetes, coronary heart disease, stroke, breast, ovarian, and colorectal cancer), generates a personalized plan for prevention via recommended lifestyle changes and screening, and collects data about health-related behaviors such as smoking and exercise, screening tests such as mammography, and health history among one's first- and second-degree relatives.

Family History Tools—American Medical Association (**www.ama-assn.org/ama/pub/physician-resources/medical-science/genetics-molecular-medicine/family-history.shtml**) offers several free family history tools developed by the American Medical Association for genetic screening, a sample pedigree and an adult family history questionnaire.

Mayo Clinic—Compiling Your Family Medical History (**www.mayoclinic.com/health/medical-history/HQ01707**)

National Human Genome Research Institute (**genome.gov/health**) provides information about genetic testing, key issues in genetics and health, fact sheets, and a glossary of terms.

NIH's National Genome Research Institute's list of online tools for generating a family history (**www.genome.gov/11510372**)

Surgeon General's Family Health History Initiative. National family history day (**www.hhs.gov/familyhistory**)

the importance of familial risks in developing chronic diseases and privacy and confidentiality concerns related to entering personal data into an online database. In addition to the Surgeon General's online tool, a number of other resources are available to assist in preparing and maintaining a family health history. Some of these resources are identified in Table 4.

Closure

As health care increasingly progresses toward personalized approaches to detect and treat health problems, the American Society of Human Genetics characterizes a family health history as "the most important genetic test of all."[12] Amid the myriad technological advances for monitoring, diagnosing, and screening of our current health status and likely health challenges in decades ahead, it is important for critical care nurses to keep in mind that—at least for the present—the family medical history remains the most reliable and valid tool we currently have available to make these patient assessments and provide guidance to our patients.

Creating, updating, and sharing a family health history can empower individuals to be more proactive in their personal health and lifestyle surveillance, make more timely and informed health-related decisions, improve their own health outcomes, minimize development of serious complications, and offer peace of mind in place of anxiety or fear of the unknown.[4] Learning about and acting upon one's family's health history may help to ensure a longer, healthier future together with fewer family funerals and many more joyous family reunions to celebrate those years.

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References

1. American Society of Human Genetics. New research validates clinical use of family health history as the 'gold standard' for assessing personal disease risk. Press release October 22, 2010. http://www.ashg.org/pdf/PR_FamilyHealthHistory_110510.pdf. Accessed November 11, 2010.
2. Leach B, Eng C. Comparison of family health history to personal genomic screening for risk assessment of breast, colon and prostate cancer. Abstract presented at American Society of Human Genetics 60th Annual Meeting; November 2-6, 2010; Washington, DC. http://ashg.org/cgi-bin/2010/showdetail.pl?absno=20199. Accessed November 11, 2010.
3. Neergaard L. Family health history: 'Best kept secret' in care. Associated Press, Medical News, 2010. http://news.yahoo.com/s/ap/us_med_healthbeat_family_health. Accessed November 11, 2010.
4. Cleveland Clinic Genomic Medicine, Patient Resources-Family History. http://my.clevelandclinic.org/genomics-genetics/patient-resources/family-history.aspx. Accessed November 12, 2010.
5. Mayo Foundation for Medical Education and Research. Mayo Clinic. Medical History: Compiling Your Family Medical History. http://www.mayoclinic.com/health/medical-history/HQ01707. Accessed November 12, 2010.
6. US Department of Health & Human Services. Surgeon General's Family History Initiative: Resource Packet for Health Professionals. Frequently Asked Questions About Family Health History. http://www.hhs.gov/familyhistory/respachealth.html. Accessed November 12, 2010.
7. US Department of Health & Human Services. Surgeon General's Family History Initiative: Resource Packet for Health Professionals. Fact Sheet. http://www.hhs.gov/familyhistory/respachealth.html. Accessed November 12, 2010.

8. National Society of Genetic Counselors. Your Family History. http://www.nsgc.org /About/FamilyHistoryTool/tabid/226 /Default.aspx. Accessed November 12, 2010.
9. Rubinstein WS, O'Neill SM, Rothrock N, et al. Components of family history associated with women's disease perceptions for cancer: a report from the Family Healthware Impact Trial (FHITr). Abstract presented at American Society of Human Genetics 60th Annual Meeting; November 2-6, 2010; Washington, DC. http://www.ashg.org /cgi-bin/2010 /showdetail.pl?absno=20467 .pdf. Accessed November 11, 2010.
10. Grendzynski D. Talking Family History at Thanksgiving. Cleveland Clinic—About Us. http://myclevelandclinic.org/news_service /family_history.aspx. Accessed November 12, 2010.
11. Arar N. Veterans' Response to the Use of Family Health History Online Tool. Department of Medicine, Division of Nephrology, University of Texas Health Science Center, San Antonio, TX. Abstract presented at American Society of Human Genetics 60th Annual Meeting; November 2-6, 2010; Washington, DC. http://ashg.org/cgi-bin /2010/showdetail .pl?absno=21223.pdf. Accessed November 11, 2010.
12. American Society of Human Genetics. Know Your Family Health History: Information for Healthcare Providers. http://www .talkhealthhistory.org/healthcare/index .shtml. Accessed November 11, 2010.