



[Print](#) :: [Close](#)

FAST FACTS AND CONCEPTS #206

Author(s): John M Quillin PhD, Joann N Bodurtha MD, and Thomas J Smith MD

Background Many dying patients voice concern for the health of surviving family members (1,2). The most common causes of death can cluster in families, and this clustering can reflect shared family genes. About 5% to 10% of cancers are strongly hereditary (3) and a family history of heart disease is well established as a risk factor for the disease (4-6). Family members may benefit from knowing their genetic risk, and offering testing can be a generative act for a dying patient. Today's genetic tests can identify known markers of disease for only some families, so testing is most helpful if it includes the affected patient (otherwise 'negative' test results are less informative). In addition, more informative genetic tests will likely be available in the future. Once patients die, however, their DNA is no longer readily available for this future testing. On the other hand, decisions about genetic testing are complex and can have profound emotional, familial, and financial impacts on those affected and should not be pursued hastily.

Possible Genetic Conditions If a patient or family member asks if survivors could be affected by the patient's disease, consider recommending genetic testing or banking for known genetic disorders (e.g. hemochromatosis, cystic fibrosis) and in the following settings:

- Earlier-than-expected age at diagnosis (e.g. breast cancer before age 50)
- Multiple primary cancers (e.g. a history of both colon and ovarian cancer)
- A major birth defect (e.g. spina bifida, congenital heart defect) or multiple minor physical anomalies
- Profound hearing or vision loss without an environmental explanation
- Developmental disability or autism
- Disorders of sexual development
- Unusually tall or short stature compared to relatives
- Unusual skin pigmentation, such as ≥ 6 café-au-lait spots, or lumps (e.g., multiple lipomas)
- Congenital myopathy or muscular dystrophy
- Cardiomyopathy or arrhythmia without clear cut cause, or at an age earlier than expected
- Suspected connective tissue disease (e.g. hyperflexibility)
- Excessive bleeding or clotting tendencies not associated with medication or comorbidity
- Seizures without an identifiable etiology

Talking to Patients and Families There is no consensus on who should be approached for discussion of familial risk, and to date no studies on effective communication strategies for genetic screening at the end of life are available, despite acknowledgment of a practice gap (7-9). In practice, a straightforward invitation for discussion of the topic may be effective: "I'd like to talk with you about your family health history. Some health conditions tend to run in families and knowing your health history and seeing if it is connected to your illness could help others in your family to stay healthy. Most diseases are not strongly genetic, but sometimes it is helpful to have a genetic test or store a blood sample for testing later."

Genetic counselors, medical geneticists, and genetic nurses can facilitate comprehensive genetics evaluation, assist in test selection, provide informed consent, and educate patients and family members about indications for and costs of testing and banking. Genetic consultation is usually covered by major insurers and Medicare, especially for patients with active disease such as cancer.

DNA Banking Banking involves drawing blood for long-term storage at a DNA banking facility. DNA banking is

typically not covered by insurance; costs vary from one to a few hundred dollars. Facilities offering DNA banking vary in their informed consent requirements and documentation for ownership of samples (e.g. who is authorized to submit a sample to a laboratory for genetic testing). Banking can be particularly helpful considering turn-around-time for genetic test results can be long and more informative tests may become available in the future. In addition, given the complex and emotional decision making that can be involved in genetic testing, banking gives family members time to seek counseling and to carefully consider such decisions, without feeling pressure to pursue testing before a loved one dies.

Resources Clinical laboratories that currently offer DNA banking can be found at <http://www.genetests.org>. Providers can search for nearby genetic counselors and geneticists at <http://www.nsgc.org> and <http://www.acmg.net>. General resources for clinicians about genetic counseling and testing can be found in references 10-13.

References

1. Duggleby W, Wright K. Elderly palliative care cancer patients' descriptions of hope-fostering strategies. *Int J Palliat Nurs*. 2004; 10:352-359.
2. Skirton H, Frazier LQ, Calvin AO, Cohen MZ. A legacy for the children--attitudes of older adults in the united kingdom to genetic testing. *J Clin Nurs*. 2006; 15:565-573.
3. Offit K. *Clinical Cancer Genetics*. New York: Wiley-Liss, Inc.; 1998.
4. Hunt SC, Gwinn M, Adams TD. Family history assessment: Strategies for prevention of cardiovascular disease. *Am J Prev Med*. 2003; 24:136-142.
5. Murabito JM, Pencina MJ, Nam BH, et al. Sibling cardiovascular disease as a risk factor for cardiovascular disease in middle-aged adults. *JAMA*. 2005; 294:3117-3123.
6. Lloyd-Jones DM, Nam BH, D'Agostino RB S, et al. Parental cardiovascular disease as a risk factor for cardiovascular disease in middle-aged adults: A prospective study of parents and offspring. *JAMA*. 2004; 291:2204-2211.
7. Kirk J. The family history of cancer - a common concern in palliative care. *Progress in Palliative Care*. 2004; 12:59-65.
8. Lillie AK. Exploring cancer genetics and care of the family: An evolving challenge for palliative care. *Int J Palliat Nurs*. 2006; 12:70-74.
9. Quillin JM, Bodurtha JN, Smith TJ. Genetics assessment at the end of life: Suggestions for implementation in clinic and future research. *J Palliat Med*. 2008; 11:451-458.
10. Pletcher BA, Toriello HV, Noblin SJ, et al. Indications for genetic referral: A guide for healthcare providers. *Genet Med*. 2007; 9:385-389.
11. American Society of Clinical Oncology. American society of clinical oncology policy statement update: Genetic testing for cancer susceptibility. *J Clin Oncol*. 2003; 21:2397-2406.
12. Genetics and Your Practice. March of Dimes. Available at: <http://marchofdimes.com/gyponline/index.bm2>. Accessed July 10, 2008.
13. National Office of Public Health Genomics. Centers for Disease Control and Prevention. Available at: <http://www.cdc.gov/genomics/>. Accessed July 10, 2008.

Authors' Affiliation: Massey Cancer Center, Virginia Commonwealth University, Richmond, VA.

Fast Facts and Concepts are edited by Drew A Rosielle MD, Palliative Care Center, Medical College of Wisconsin. For more information write to: drosiell@mcw.edu. More information, as well as the complete set of Fast Facts, are available at EPERC: www.eperc.mcw.edu.

Version History: Originally published August 2008. Current version re-copy-edited in June 2009.

Copyright/Referencing Information: Users are free to download and distribute Fast Facts for educational purposes only. Quillin JM, Bodurtha JN, Smith TJ. Genetic Screening and DNA Banking at the End of Life. Fast Facts and Concepts. August 2008; 206. Available at: http://www.eperc.mcw.edu/fastfact/ff_206.htm.

Disclaimer: Fast Facts and Concepts provide educational information. This information is not medical advice.

Health care providers should exercise their own independent clinical judgment. Some Fast Facts cite the use of a product in a dosage, for an indication, or in a manner other than that recommended in the product labeling. Accordingly, the official prescribing information should be consulted before any such product is used.

ACGME Competencies: Medical Knowledge; Interpersonal and Communications Skills

Keywords: Ethics, Law, Policy Health Systems; Communication

© 2008 Medical College of Wisconsin

Medical College of Wisconsin

8701 Watertown Plank Road, Milwaukee, WI 53226

www.mcw.edu | 414.456.8296

[Print](#) :: [Close](#)