

**FAST FACTS AND CONCEPTS #206  
GENETIC SCREENING AND DNA BANKING AT THE END OF LIFE**

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**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  $\geq 6$  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.

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