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GENOMIC MEDICINE

Presymptomatic Testing of Minors

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Your first patient of the day, June, is a bright and mature 16-year-old who prematurely lost her favorite maternal aunt to metastatic breast cancer. Before her death, June's aunt informed June that her cancer was related to a mutation in the BRCA1 gene. June wants to be tested, as she has learned through the Internet that this mutation may be passed through the family and that mutation carriers can reduce their risk by enhanced screening. June's mother voices concerns about the testing of either herself or her daughter, for if June were found to have a mutation, her mother would learn that she harbors the mutation as well.

Uncomfortable though it may be, this scenario is not unrealistic. Presymptomatic genetic testing is becoming increasingly available for a wide range of disorders. The availability of these tests raises a number of questions surrounding the who, what, when, and why of test delivery. Of great interest is the question of whether or not presymptomatic tests should be offered to those under the age of legal consent.

Genetic test results can empower individuals to make informed decisions re-

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garding their elevated risk of developing a disease. For some disorders, proven risk-reducing choices include enhanced screening, behavioral modifications, chemoprophylaxis, or surgery. In June's case, at least some of these options might become available to her if she were known to have a BRCA1 mutation.

For other genetic tests, a result conferring risk might come with few recommendations proven to prevent the onset or progression of disease—a particularly controversial example of this is Alzheimer's disease predisposition testing. Without available risk-reducing measures, the utility of genetic testing is questionable, although some feel that testing allows better preparations for the future. Others fear that discovering a genetic predisposition for a disorder with no means to reduce risk could provoke excessive worry about the onset of disease. A number of additional concerns have been raised regarding presymptomatic genetic testing of minors. First, depending on the age of the minor in question, it may be difficult for them to make a fully informed, autonomous decision. June, her parents, and you as her health care provider cannot look into the future and predict whether she will want the genetic information as an adult. Second, some worry that minors could be particularly vulnerable to the fears and anxieties that can be associated with genetic testing. Testing may cause them to feel stigmatized or be treated differently by their families.

A number of practice guidelines for testing of minors have been issued by the genetics community, bioethics advisory boards, as well as by medical and pediatric professional associations (Clin. Genet. 2006;70:374-81). The accepted justification for presymptomatic genetic testing of minors is for an immediate health benefit that would be lost if testing is delayed. Typically, this situation involves serious conditions with a childhood age of onset; an example would be some forms of familial colon cancer. In this setting, genetic testing may help to determine if recommended invasive screening measures are warranted, and death or serious disease might be avoided in the short term.

Sometimes, the available guidelines are difficult to apply. Not uncommonly, the immediate and even long-term health benefits of early intervention following genetic testing are unclear. In other cases, diseases for which testing may be done may have a childhood age of onset, but limited prevention strategies. In the latter instance, the genetic test may offer benefit to the family, allowing them to prepare emotionally and perhaps guiding decisions about having more children. However, it may also confer emotional risks to both the family and the child.

Presymptomatic genetic testing can enhance efforts directed at the prevention and early detection of a wide range of conditions. A well-informed, compassionate health care professional willing to collaborate with patients and their families can be an invaluable resource for patients like June. ■

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