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Direct-to-consumer genetic testing: Access and marketing

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The American College of Medical Genetics (ACMG), in their "Statement on Direct-to-Consumer Genetic Testing"[1] (in this issue of *Genetics in Medicine*), argues for the involvement of appropriately qualified health care professionals in the ordering and interpretation of genetic tests, and the counseling of individuals and families regarding the meaning and significance of the test results. They point out the potential harms that may result if such health professionals are not involved, including misused tests, misinterpreted results, and misguided follow-up. We feel that this statement from the ACMG will not only educate the public and professionals about this issue, but will also lead to further discourse. In this commentary, we will provide a brief background for these discussions.

One argument in favor of direct-to-consumer testing relies on respect for patient autonomy. Such reliance, however, ignores the need for information if autonomy and decision-making are to be meaningful. We acknowledge and cherish the autonomy of individuals to make decisions regarding their own health care. Geneticists show respect for and uphold autonomy through nondirective counseling. We agree with the ACMG, however, that decisions regarding whether and how an individual wishes to use a genetic test, and the information derived from it, should be informed by discussions with a knowledgeable health professional.

Others argue that direct access to genetic testing reduces health care costs by eliminating the need for consultation with trained professionals. However, this argument fails to account for the costs likely to result from the uninformed and unnecessary uses of genetic tests and from the adverse consequences of inappropriate responses to test results, whether positive or negative, valid or invalid. Such consequences can include underuse and overuse of health care resources.

Concerns regarding direct access to genetic testing are closely tied to concerns about direct-to-consumer advertising and marketing of genetic testing. Arguments have been made for direct-to-consumer marketing of genetic tests based on experience with direct-to-consumer advertisement of prescription drugs.[2] Benefits cited by advocates include increased compliance and facilitation of patient-physician communication (based on the required "talk with your doctor" phrase). In the context of genetic tests, benefits of direct-to-consumer advertising may include an increased awareness of the importance of family history, the relation between risk and family history, the role of genetics in disease, and the value of genetic counseling.

When advertisements for genetic tests are presented on the Internet or in the media by a commercial entity, however, significant clinical information may be missing. For example, there is frequently no information provided regarding the clinical validity and utility of the test. Consumers are not advised, for example, whether a test will provide the answers they are seeking regarding a particular disorder. An individual consumer is unlikely to know the positive predictive value (probability that a positive test result indicates the person will develop the disorder) or the negative predictive value (probability that a person will not develop a disorder if the test does not find a mutation) of the test. The consumer also is unlikely to know the performance characteristics of that test in the specific context in which they are requesting the test result; e.g., test performance may be very different in the presence or absence of a positive family history for the disease, or in the presence or absence of a known mutation.

A consumer also may not be able to determine whether a particular genetic test is appropriate for them in the absence of consultation with a trained health professional.[2] For example, a woman with a family history of breast cancer who is concerned about her risk for breast cancer (consultand) would do better to have a sample from an affected relative who meets the criteria for familial breast cancer (proband) tested for mutations in *BRCA1* and *BRCA2* than to have her own sample tested. If the proband did not have a *BRCA1* or *BRCA2* mutation, then the consultand would be at no different risk for breast cancer before or after she was tested. An appropriately trained health professional can help the consultand identify the best proband(s) for initial testing in order for the consultand to receive the best and most useful information from their genetic testing.[2,3] Without this kind of health professional involvement and counseling, direct-to-consumer advertising may reinforce an erroneous deterministic interrelationship between genotype and phenotype.[4]

An appropriately trained health professional also would be able to advise the consultand regarding the actual information that may be available from the test, as well as its potential utility. The difficulties encountered in the interpretation of genetic test results are well-recognized.[3] The public generally expects to receive a definitive, yes/no answer from a medical test. Results from genetic tests often inform only the estimation of the probability of developing a disease or the predictive risk of

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disease. Many direct-to-consumer advertisements are vague or misleading regarding how the results could influence the individual's predictive risk[3] and may erroneously reinforce a deterministic interrelationship between genotype and phenotype.[4] A correct estimation of predictive risk must be based, not only on genetic testing results, but also on family history and personal medical history and, therefore, should be individualized for each person being tested. Appropriate counseling is required for a full explanation, and a full understanding, of an individual's predictive risk.[2,3]

Without information from a trained health professional, an individual also would not know how his or her medical options may be influenced by the results of the test, or even what those options might be.[2,3] For example, if a woman had a mutation in *BRCA2*, one option would be to have an oophorectomy. If she planned to have children in the future, however, she might not want to have her ovaries removed until after child-bearing plans are over. In fact, she also might not have wanted to have the genetic test performed until she was in a position to act on the information from the test. An appropriately trained health professional can help such an individual determine whether and when testing might be appropriate for her personally, including medical, psychological, and cultural considerations.

The person considering a genetic test also must understand the implications of the test results for their health insurance.[5] Because of concerns about genetic discrimination, these days, individuals often have genetic tests performed under a pseudonym so that their health insurer will not know that a test was performed and will not know the results. These individuals later may be surprised to learn that the "small print" on their policy may render it null and void if they have, or subsequently receive, additional pertinent information that they do not report to their insurer.

In addition to those noted above, other concerns that have been raised about direct-to-consumer marketing of genetic tests include misinformation, risk exaggeration, and lack of an evidence-base for clinical decision-making.[2–4] Further, direct-to-consumer marketing may target vulnerable individuals, as when a testing company included an advertisement for *BRCA1* and *BRCA2* mutation analysis in the printed program for a play describing the painful death of a woman with ovarian cancer.[6] Finally, direct-to-consumer advertisements for genetic tests are, at the present time, considered to be less "well-regulated" than those for prescription drugs by the U.S. Food and Drug Administration (FDA) and Federal Trade Commission (FTC).[2]

As a final note, we believe that ACMG statements such as the "Statement on Direct-to-Consumer Genetic Testing" engender valuable discussion and, therefore, can be quite powerful. The ACMG statement of concerns regarding policies on gene patents and licenses[7] contributed to a broad discussion of these issues[5,8,9] that resulted in a significant change in gene patent policies by the U.S. Patent and Trademark Office.[10] We would hope that the ACMG statement on direct-to-consumer genetic testing will inform health professionals and the public about this issue, and encourage further discussion of this important topic and the closely intertwined issues associated with direct-to-consumer marketing of genetic tests.

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