Forward to a Colleague

March 17, 2018

ACMG Responds to FDA's Approval for Direct-to-Consumer Testing for Three BRCA Gene Mutations

ACMG has long been an outspoken advocate on the need for knowledgeable health professionals to be involved in interpreting the clinical implications of the results of genetic testing for a specific individual, and the equally important need for professionals be involved in informing people of which test is most useful for them, if any. ACMG's first Statement on Direct to Consumer Genetic Testing was published in 2004, followed by statements in 2008 and 2015, as the field evolved.

The public deserves access to different types of medical information about themselves. However, consumers are better off when this type of information is provided through a health care system where informed consent is obtained and the privacy of individual health information is protected and only made available secondarily with explicit permission from those to whom the data relates. Consumers are best served when testing is done with ready access to unconflicted experts able to both order the most appropriate tests for that individual and then interpret those tests whether the test result is positive or negative.

With the U.S. Food and Drug Administration's March 6, 2018 approval of the marketing of the 23andMe Personal Genome Service Genetic Health Risk Report for BRCA1/2 (Selected Variants), consumers will obtain a qualitative, general report of increased risk based on only three out of more than 1,000 *BRCA1* and *BRCA2* mutations known to correspond to increased cancer risk, rather than a quantitative measure of that risk which is specific to their situation, thereby making it incomplete. Such nonspecific results will require follow-up in the health care system to individualize the risk. ACMG believes that a test result has maximal value when the information it provides is interpreted in the context of a particular individual as well as our current knowledge and understanding of the test's limitations.

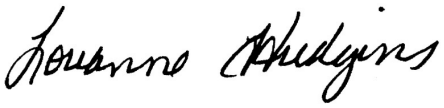
Consumers who consider participating in direct to consumer (DTC) genetic testing must be aware of the limited results that they will receive and the types of questions that they will have to anticipate in the

follow-up of the results. Appropriate counseling can be provided by medical geneticists and genetic counselors and also may be done by other professionals who have acquired experience and training related to the particular genetic risks related to conditions in their area of specialty.

It is important to note that this test does not provide information on which a person should make medical decisions. For those identified with a mutation, the result will have to be put into the context of that person's family history and medical history to provide individualized interpretation of the risk associated with this finding.

False reassurance after a negative result can cause harm. Even in someone who is Ashkenazi Jewish, one needs to take into account personal and family medical history in the interpretation. A negative result that leads to false reassurance of risk could reduce the chances that an individual pursues the best tests and health practices in the future (including for example, mammograms) for their own risk indicators. In this situation, it is important for consumers to know that a negative (normal) result does not rule out an increased cancer risk or the presence of a different *BRCA1* or *BRCA2* pathogenic variant.

As always, we encourage individuals who have questions about genetic testing to find a board-certified genetics professional in their area by visiting www.acmg.net.



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