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Study Shows Consumers Have Few Negative Reactions to the Results of Genetic Testing for Cancer Mutations

Information Prompts Individuals to Consult Physician, Inform Relatives

A 23andMe study of consumers' reactions to genetic testing found that even when the tests revealed high-risk mutations in individuals, those individuals had few negative reactions to the news. Instead of inducing serious anxiety, the test results prompted people to take positive steps, including follow-up visits with a doctor and discussions with family members who could also be at risk.

The study, titled "Dealing with the unexpected: Consumer responses to direct-access BRCA mutation testing" published today as part of the launch of PeerJ, a new peer reviewed open access journal in which all articles are freely available to everyone (<https://PeerJ.com>).

"The paper addresses one of the most urgent questions in the field of genetics and genomics, namely the impact of receiving unexpected information about high genetic risk for a life-threatening disease," said the lead author of the paper and 23andMe's Senior Medical Director Dr. Uta Francke.

The study looked at how people reacted when they learned for the first time that they carried a mutation in either the BRCA1 or BRCA2 gene that put them at higher risk for breast and ovarian cancer. The study included interviews conducted with 32 individuals found to be mutation carriers and 31 individuals found to be non-carriers. Five to ten percent of breast cancers occur in women with a genetic predisposition for the disease, usually due to mutations in either the BRCA1 or BRCA2 gene. The mutations at the center of this study are responsible for a substantial number of hereditary breast and ovarian cancers among women with Ashkenazi Jewish ancestry.

Those who acquired the potentially life-saving information not only took appropriate actions on their own behalf, but also notified relatives who might share that risk. In what the study described as a "cascade effect," a number of relatives who were subsequently tested discovered they too had one of the mutations.

The findings are important given that a frequent concern regarding direct-to-consumer testing is based on the assumption that it causes either serious emotional distress or triggers deleterious actions on the part of consumers. Individuals who learned they had the mutation said they did not suffer serious emotional distress, and did not take inappropriate actions. All but one of the 32 mutation-positive participants appreciated learning their BRCA mutation status. None of the 31 mutation-negative individuals misinterpreted their result to think they are free from all risks and safe to abandon routine cancer screening.

The study provides important preliminary data that suggest some of the bioethical concerns may be overstated, at least for the self-selected pool of individuals who seek DTC personal genomics information. The authors suggested that broader screening of Ashkenazi Jewish women for these three BRCA mutations should be considered.

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Link to the Published Version of the article: <https://peerj.com/articles/8>

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About 23andMe

23andMe, Inc. is the leading personal genetics company dedicated to helping individuals understand their own genetic information through DNA analysis technologies and web-based interactive tools. The company's Personal Genome Service® enables individuals to gain deeper insights into their ancestry and inherited traits. The vision for 23andMe is to personalize healthcare by making and supporting meaningful discoveries through genetic research. 23andMe, Inc., was founded in 2006, and the

company is advised by a group of renowned experts in the fields of human genetics, bioinformatics and computer science. More information is available at www.23andme.com.

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Abstract (from the article)

Background. *Inherited BRCA gene mutations convey a high risk for breast and ovarian cancer, but current guidelines limit BRCA mutation testing to women with early-onset cancer and relatives of mutation-positive cases. Benefits and risks of providing this information directly to consumers are unknown.*

Methods. *To assess and quantify emotional and behavioral reactions of consumers to their 23andMe Personal Genome Service report of three BRCA mutations that are common in Ashkenazi Jews, we invited all 136 BRCA1 and BRCA2 mutation-positive individuals in the 23andMe customer database who had chosen to view their BRCA reports to participate in this IRB-approved study. We also invited 160 mutation-negative customers who were matched for age, sex and ancestry. Semi-structured phone interviews were completed for 32 mutation carriers, 16 women and 16 men, and 31 non-carriers. Questions addressed personal and family history of cancer, decision and timing of viewing the BRCA report, recollection of the result, emotional responses, perception of personal cancer risk, information sharing, and actions taken or planned.*

Results. *Eleven women and 14 men had received the unexpected result that they are carriers of a BRCA1 185delAG or 5382insC, or BRCA2 6174delT mutation. None of them reported extreme anxiety and four experienced moderate anxiety that was transitory. Remarkably, five women and six men described their response as neutral. Most carrier women sought medical advice and four underwent risk-reducing procedures after confirmatory mutation testing. Male carriers realized that their test results implied genetic risk for female relatives, and several of them felt considerably burdened by this fact. Sharing mutation information with family members led to screening of at least 30 relatives and identification of 13 additional carriers. Non-carriers did not report inappropriate actions, such as foregoing cancer screening. All but one of the 32 mutation-positive participants appreciated learning their BRCA mutation status.*

Conclusions. *Direct access to BRCA mutation tests, considered a model for high-risk actionable genetic tests of proven clinical utility, provided clear benefits to participants. The unexpected information demonstrated a cascade effect as relatives of newly identified carriers also sought testing and more*

mutation carriers were identified. Given the absence of evidence for serious emotional distress or inappropriate actions in this subset of mutation-positive customers who agreed to be interviewed for this study, broader screening of Ashkenazi Jewish women for these three BRCA mutations should be considered.