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Is There Evidence to Support *BRCA1/2* and Other Inherited Breast Cancer Genetic Testing for All Patients with Breast Cancer?

BETHESDA, MD – December 13, 2019 | Should germline genetic testing be offered to all patients with breast cancer? The American College of Medical Genetics and Genomics (ACMG) addresses this important question in a new statement published in *Genetics in Medicine*, "[Points to Consider: Is There Evidence to Support *BRCA1/2* and Other Inherited Breast Cancer Genetic Testing for All Breast Cancer Patients? A Statement of the American College of Medical Genetics and Genomics.](#)"

Of all cancers that develop in women in the United States (US), breast cancer has the highest incidence, regardless of race or ethnicity. Approximately 5-10% of breast cancers are estimated to result from hereditary causes, the majority of which are attributed to pathogenic or likely pathogenic (P/LP) variants in the *BRCA1* and *BRCA2* (*BRCA1/2*) genes, although variants in other genes such as *PALB2*, *TP53*, *PTEN*, *CDH1*, *CHEK2* and *ATM* also contribute.

Identification of inherited cancer risk empowers individuals and their families to prevent cancers or detect them early. Furthermore, incorporating genetic testing results into patients' care plans has the potential to guide treatment and improve outcomes. But testing alone will not improve outcomes. Implementation of appropriate care following testing is required and data are needed to generate evidence that informs clinical practice.

As progress in precision medicine continues, it is important that patients receive accurate information to ensure the implementation of risk reducing strategies and evidence-based cancer genomics best practices. The purpose of this new ACMG points-to-consider document is to outline the rationale for ongoing support of existing evidence-based guidelines built on a risk stratification approach while data related to broader testing strategies continues to emerge.

"Medical geneticists play an important role in facilitating the best care and practices of patients with cancer or a predisposition to develop cancer," said ACMG President Anthony R. Gregg, MD, MBA, FACOG, FACMG. "This Points to Consider document acknowledges the complexity of professional organization guidelines in the cancer space. Medical geneticists are uniquely qualified to analyze the literature that informs professional organizations and their guidelines. Implementation of cancer genetic testing guidelines is best when carried out with input and in many cases under the direction of a medical geneticist with cancer expertise."

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The new ACMG document provides points for clinicians to consider in the context of testing breast cancer patients for inherited cancer predisposition, including:

1. All patients with breast cancer should be evaluated to determine the need for germline genetic testing for hereditary breast cancer. According to national practice guidelines, genetic testing for breast cancer patients is indicated based on patient characteristics including age at diagnosis, family cancer history, expression of estrogen progesterone receptors and *HER2* expression and disease stage.
2. When discussing genetic testing for breast cancer patients, clinicians should be aware of the current insufficient evidence to support genetic testing for all patients with breast cancer, especially with multi-gene panels that include genes without evidence to support follow-up care.
3. When discussing management after identification of a P/LP in moderately penetrant breast cancer genes:
 - a. recognize that guidance is based on consensus recommendations; and
 - b. enhanced screening has not, to date, been associated with enhanced survival or identification of disease at an earlier stage.
4. Whenever genetic testing for inherited breast cancer is performed on a clinical basis, testing should include full gene sequencing, deletion/duplication analysis and detection of known P/LP intronic variants in a Clinical Laboratory Improvement Amendments (CLIA)-certified and/or College of American Pathologists (CAP)-accredited genetic testing laboratory.
5. The implications of genetic testing should be reviewed with patients in the context of genetic counseling as genetic testing is ordered. This counseling should include the expertise of a trained genetics professional or healthcare provider with special expertise in cancer genetics principles.
6. Patients who have a P/LP variant in an established breast cancer-associated gene in which evidenced-based follow-up recommendations exist should be educated about the importance of cascade testing of family members.

The points-to-consider document concludes by stating, “With the advances in sequencing technologies and increasing access to and expanding indications for genetic testing, it remains critical to ensure that implementation of testing is based

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on evidence. Currently, there is insufficient evidence to recommend genetic testing for *BRCA1/2* alone or in combination with multi-gene panels for all breast cancer patients.... Ideally, professional societies should work together to weigh data, formulate and harmonize evidence-based recommendations, and seek to reduce barriers to care... Moreover...the implementation of precision medicine approaches across oncology must also consider a means by which the promise of genetic testing for inherited cancer predisposition may be realized by all populations, regardless of race, ethnicity and ability to pay.”

About the American College of Medical Genetics and Genomics (ACMG) and ACMG Foundation

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical society dedicated to improving health through the clinical practice of medical genetics and genomics and the only medical specialty society in the US that represents the full spectrum of medical genetics disciplines in a single organization. The ACMG is the largest membership organization specifically for medical geneticists, providing education, resources and a voice for more than 2,300 clinical and laboratory geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. ACMG’s mission is to improve personal and public health through the clinical and laboratory practice of medical genetics as well as through advocacy, education and clinical research, and to guide the safe and effective integration of genetics and genomics into all of medicine and healthcare. Four overarching strategies guide ACMG’s work: 1) to reinforce and expand ACMG’s position as the leader and prominent authority in the field of medical genetics and genomics, including clinical research, while educating the medical community on the significant role that genetics and genomics will continue to play in understanding, preventing, treating and curing disease; 2) to secure and expand the professional workforce for medical genetics and genomics; 3) to advocate for the specialty; and 4) to provide best-in-class education to members and nonmembers. *Genetics in Medicine*, published monthly, is the official ACMG journal. ACMG’s website (www.acmg.net) offers resources including policy statements, practice guidelines, educational programs and a ‘Find a Genetic Service’ tool. The educational and public health programs of the ACMG are dependent upon charitable gifts from corporations, foundations and individuals through the ACMG Foundation for Genetic and Genomic Medicine.

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