Breast cancer patients to be evaluated for genetic testing

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According to a statement on behalf of the American College of Medical Genetics and Genomics (ACMG) published Dec. 13 in the organization's official journal, *Genetics in Medicine*, there is insufficient evidence to

recommend universal genetic testing for BRCA1/2 alone or in combination with multi-gene panels for all breast cancer patients.

The guidance from the ACMG differs from a consensus guideline issued in February by the American Society of Breast Surgeons, which recommended genetic testing for all newly diagnosed patients with breast cancer. The ACMG recommends evaluations before genetic testing.

"What we are saying is that all women with breast cancer should be evaluated for the need for genetic testing based on existing clinical criteria," said one of the lead authors, Tuya Pal, MD, associate director of Cancer Health Disparities at Vanderbilt-Ingram Cancer Center.

The group wrote the statement on behalf of the ACMG Professional Practice and Guidelines Committee.

"We expect that the evidence to support testing may evolve at different rates for different genes, and we expect that therapeutic indications will play a major role in the incorporation of genes to multi-gene panels," Pal and co-authors stated in the paper. "Consequently, as guidelines for testing are developed, it is critical to ensure they are supported by evidence and resources supporting strategies that include screening, medical and/or surgical care as indicated. Ideally, professional societies should work together to weigh data, formulate and harmonize evidence-based recommendations and seek to reduce barriers to care."

The ACMC document stressed the importance of genetic testing and said all breast cancer patients should be evaluated to determine whether germline genetic testing for hereditary breast cancer is warranted. They noted that only a small proportion of the at-risk population for hereditary breast cancers has been tested, with one estimate indicating that less than 10% of adults with BRCA1/2 pathogenic or likely pathogenic variants in the U.S. have been identified. Testing rates are disproportionately lower

among racial and ethnic minority populations.

"As genetic testing now has the potential to guide cancer care, it has become imperative to ensure that all populations may benefit from these tremendous advances and that existing disparities in testing do not widen," Pal said. "In order to ensure this, we need to be intentional in developing and disseminating efforts such that improved outcomes based on genetic testing are experienced across populations."

The ACMG document provided the following guidance for clinicians to consider:

- Genetic testing for breast cancer patients is indicated based on patient characteristics, including age at diagnosis, family cancer history and expression of estrogen progesterone receptors and HER2 expression.
- In discussions with patients, clinicians should be aware of the current insufficient evidence to support genetic testing for all patients with breast cancer.
- After identification of a pathogenic or likely pathogenic mutation in moderately penetrant breast cancer genes, clinicians should recognize that guidance is based on consensus recommendations and that enhanced screening, to date, has not been associated with enhanced survival or earlier stage diagnosis.
- Whenever genetic testing is performed on a clinical basis, the testing should include full gene sequencing and be conducted in a lab certified or accredited by either the College of American Pathologists or Clinical Laboratory Improvement Amendments.
- Patients should be counseled about the implications of genetic testing by trained genetics professionals or health care providers with special expertise in cancer genetics principles.
- Patients who have a pathogenic or likely pathogenic variant in an established breast cancer associated gene should be educated

about the importance of cascade testing of family members.

More information: et al, 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 (ACMG), *Genetics in Medicine* (2019). DOI: 10.1038/s41436-019-0712-x , dx.doi.org/10.1038/s41436-019-0712-x

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