Mayo researchers recommend all women with breast cancer diagnosis under age 66 be offered genetic testing

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A study by researchers at Mayo Clinic published this week in the *Journal of Clinical Oncology* suggests that all women with a breast cancer

diagnosis under the age of 66 be offered germline genetic testing to determine if they have a gene mutation known to increase the risk of developing other cancers and cancers among blood relatives. Current guidelines from the National Comprehensive Cancer Network (NCCN) recommend germline testing for all women diagnosed with breast cancer under the age of 46 regardless of their family history and breast cancer subtype.

"There is considerable confusion regarding the best method for selecting who may benefit from hereditary cancer genetic testing from among all women diagnosed with breast cancer," says Fergus Couch, Ph.D., a breast cancer researcher at Mayo Clinic. "The NCCN has very specific guidelines for who may benefit from genetic testing based on the age of diagnosis and family history of certain cancers while the American Society of Breast Surgeons (ASBrS) recommends testing all women with breast cancer."

For their study, Dr. Couch and his colleagues evaluated all known breast cancer predisposition genes in a Mayo Clinic breast cancer registry and showed that NCCN guidelines overlooked approximately 30% of patients with genetic mutations known to increase the risk of developing breast cancer. Based on this information

Dr. Couch and his colleagues recommend increasing the age for genetic testing to all women diagnosed with breast cancer under the age of 66 irrespective of family history of cancer . "This change would help identify 98% of women with BRCA1 and BRCA2 mutations, and more than 90% of women with mutations in other predisposition genes, while avoiding testing of 20% of all breast cancers," says Dr. Couch.

He says this approach may also result in a reduced burden on the genetic services needed for women receiving testing.

"We were surprised to find that the NCCN guidelines missed approximately 30% of mutation carriers in breast cancer predisposition genes," says Siddhartha Yadav, M.B.B.S., a medical oncologist and first author of the study.

"A few recent studies have demonstrated that NCCN guidelines could miss a substantial number of mutation carriers. However, these studies included several genes that are not typically associated with breast cancer risk. Our study was appropriately restricted to nine breast cancer predisposition genes with clear management guidelines."

Dr. Couch says it was encouraging to note that by simply changing the age cutoff for germline genetic testing in women with breast cancer, rather than other more complicated approaches, it should be possible to identify the majority of mutation carriers. In women diagnosed with breast cancer over the age of 65, the study supports the use of NCCN guidelines to decide who should undergo germline genetic testing. The overall result is that many more women and their family members can benefit from knowing that they are at increased risk of cancer.

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