Newly-identified gene mutation could help explain how breast cancer spreads

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A newly-identified genetic mutation could increase our understanding of how breast cancer spreads and potentially guide treatment options for women with the disease, according to a study from Magee-Womens Research Institute (MWRI) and the University of Pittsburgh Cancer Institute (UPCI) presented today at the 2014 San Antonio Breast Cancer Symposium.

This research represents the most comprehensive analysis to date of genomic changes that occur in breast cancer progression and indicate the extensive changes that happen during the spread of the disease.

Researchers from MWRI and UPCI sequenced frozen breast tumor samples from six patients, beginning with the primary tumor when the cancer was first diagnosed through the progression of metastatic disease. Using multiple sequencing techniques, the team identified a new gene created by two separate genes that fused together as a result of unstable DNA. This fusion gene was identified in a metastatic tumor sample and is believed to play a part in the spread of the original breast cancer.

"We applied all of our sequencing technologies to the tumors in order to understand the changes that occur between the first breast cancer occurrence and late-stage disease," said Ryan Hartmaier, a research instructor at the University of Pittsburgh and lead author of the study.

Since several types of breast cancer are fueled by the hormone estrogen, estrogen blocking treatment is often recommended to prevent the disease from spreading. However, the fusion gene identified did not respond to estrogen blocking treatment, contributing to the breast cancer's spread.

"This research helps us further understand the genomic landscape of metastatic breast cancer," said Adrian Lee, Ph.D., the study's senior author, director of the Women's Cancer Research Center and professor of pharmacology, chemical biology and human genetics Pitt.

"The new class of genetic changes identified take us another step further in personalized medicine and could change the way we treat certain patients if we are able to identify who will develop this genetic mutation."

Provided by University of Pittsburgh Medical Center