

Researchers discover new markers for breast cancer that may aid diagnoses, point to new therapies

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Fluorescence micrograph of breast cancer cells. Credit: Lutz Langbein, Deutsches Krebsforschungszentrum

(Medical Xpress)—The fight against breast cancer is never-ending, but Virginia Tech researchers in the Medical Informatics and Systems Division at the Virginia Bioinformatics Institute have found additional diagnostic markers that may aid clinicians to better forecast and prevent the disease.

Since one in eight women are likely to develop breast cancer in their lifetimes, finding better predictive markers is more important than ever.

The study was published in *Breast Cancer Research and Treatment*.

By using breast cancer germline (blood) samples from The Cancer Genome Atlas Project and comparing them with samples from cancer-free individuals whose genomes are found in the 1000 Genomes Project, the researchers pinpointed several novel markers that may not only reveal risks for breast cancer, but may yield therapeutic benefits, as well.

The research team specifically looked at microsatellite variation, which can be a predictor of cancer. Microsatellites are small regions of repeating DNA in the genome that can affect proper cellular function in a variety of ways.

"There is still a lot we can learn from looking at the human genome and how it can be affected in ways that may be associated with disease," said Natalie Fonville, a geneticist on the research team."This study is the first of many in which we are engaged that identify subtle genomic changes which together may add up to <u>cancer risk</u>."

Often referred to as "junk DNA" in the past, microsatellites are currently thought to be important in a number of diseases and genetic mutations.

Generally, mutations that occur are cleaned up by DNA repair mechanisms during replication, but not all of them are. Some of these mutations may lead to cancer.

Next-generation sequencing technology and new analytic tools now allow scientists to probe more deeply into the dark matter of DNA. This could yield markers that are much more reliable than those used at present, as well as possible genetic therapies in the future.

The technology described in the study has been licensed by Virginia Tech Intellectual Properties to Genomeon LLC.

"The use of microsatellite variations as diagnostics has the potential to transform the way cancer and



other heritable diseases are diagnosed and treated," Michael B. Waitzkin, Genomeon's chief executive officer. This technology is a very exciting example of the possibilities for translating academic discoveries into clinical use," he observed.

More information: "Microsatellite genotyping reveals a signature in breast cancer exomes." L. J. McIver, N. C. Fonville, E. Karunasena, and H. R. Garner. *Breast Cancer Res Treat.* 2014; 145: 791–798. Published online May 17, 2014. doi: 10.1007/s10549-014-2908-8

Provided by Virginia Tech

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