GENE RESPONSIBLE FOR MANY SPONTANEOUS BREAST CANCERS IDENTIFIED

Research published in the journal GENETICS links NF1, a known oncogene driver in other cancers, with more than 25% of breast cancers—an important finding during National Breast Cancer Awareness Month

BETHESDA, MD – October 2, 2012 -- Cancerous tumors contain hundreds of mutations, and finding these mutations that result in uncontrollable cell growth is like finding the proverbial needle in a haystack. As difficult as this task is, it’s exactly what a team of scientists from Cornell University, the University of North Carolina, and Memorial Sloan-Kettering Cancer Center in New York have done for one type of breast cancer. In a report appearing in the journal GENETICS (www.genetics.org), researchers show that mutations in a gene called NF1 are prevalent in more than one-fourth of all noninheritable or spontaneous breast cancers.

In mice, NF1 mutations are associated with hyper-activation of a known cancer-driving protein called Ras. While researchers have found earlier evidence that NF1 plays a role in other types of cancer, this latest finding implicates it in breast cancer. This suggests that the drugs that inhibit Ras activity might prove useful against breast cancers with NF1 mutations.

“As we enter the era of personalized medicine, genomic technologies will be able to determine the molecular causes of a woman’s breast cancer,” said John Schimenti, Ph.D., a researcher involved in the work from the Center for Vertebrate Genomics at Cornell University College of Veterinary Medicine in Ithaca, New York. “Our results indicate that attention should be paid to NF1 status in breast cancer patients, and that drug treatment be adjusted accordingly both to reduce the cancer and to avoid less effective treatments.”

To make this discovery, scientists analyzed the genome of mammary tumors that arise in a mouse strain prone to genetic instability -- whose activity closely resembles the activity in human breast cancer cells -- looking for common mutations that drive tumors. The gene NF1 was missing in 59 out of 60 tumors, with most missing both copies. NF1 is a suppressor of the oncogene Ras, and Ras activity was extremely elevated in these tumors as a consequence of the missing NF1 gene. Researchers then examined The Cancer Genome Atlas (TCGA) data, finding that NF1 was missing in more than 25 percent of all human breast cancers, and this was associated with a decrease in NF1 gene product levels, which in turn is known to increase Ras activity.

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“This research is compelling because it helps us understand why some breast cancers are more likely to respond to only certain types of treatment,” said Mark Johnston, Editor-in-Chief of the journal *GENETICS*. “The findings reported in this article may guide clinicians to better treatments specific to the needs of each patient.”

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*Comparative Oncogenomics Implicates the Neurofibromin 1 Gene (NF1) as a Breast Cancer Driver*  
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**ABOUT GENETICS:** Since 1916, *GENETICS* (http://www.genetics.org/) has covered high quality, original research on a range of topics bearing on inheritance, including population and evolutionary genetics, complex traits, developmental and behavioral genetics, cellular genetics, gene expression, genome integrity and transmission, and genome and systems biology. *GENETICS*, a peer-reviewed, peer-edited journal of the Genetics Society of America is one of the world's most cited journals in genetics and heredity.

**ABOUT GSA:** Founded in 1931, the Genetics Society of America (GSA) is the professional membership organization for scientific researchers, educators, bioengineers, bioinformaticians and others interested in the field of genetics. Its nearly 5,000 members work to advance knowledge in the basic mechanisms of inheritance, from the molecular to the population level. The GSA is dedicated to promoting research in genetics and to facilitating communication among geneticists worldwide through its conferences, including the biennial conference on Model Organisms to Human Biology, an interdisciplinary meeting on current and cutting edge topics in genetics research, as well as annual and biennial meetings that focus on the genetics of particular organisms, including *C. elegans*, Drosophila, fungi, mice, yeast, and zebrafish. GSA publishes *GENETICS*, a leading journal in the field and an online, open-access journal, *G3: Genes|Genomes|Genetics*. For more information about GSA, please visit [www.genetics-gsa.org](http://www.genetics-gsa.org). Also follow GSA on Facebook at [facebook.com/GeneticsGSA](http://facebook.com/GeneticsGSA) and on Twitter [@GeneticsGSA].

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