New study validates activity of rare genetic variant in glioma

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Researchers at Moffitt Cancer Center working with colleagues at three other institutions have validated a link between a rare genetic variant and the risk of glioma, the most common and lethal type of brain tumor. The validation study also uncovered an association between the same rare genetic variant and improved rates of survival for patients with glioma.

The study, the first to confirm a rare susceptibility variant in glioma, appeared in a recent issue of the Journal of Medical Genetics, a journal published by the British Medical Association.

"Glioma is a poorly understood cancer with high morbidity and devastating outcomes," said study lead author Kathleen M. Egan, Sc.D., interim program leader of Cancer Epidemiology and vice chair of the Department of Cancer Epidemiology. "However, the discovery of the association of the TP53 genetic variant rs78378222 with glioma provides new insights into these tumors and offers better prospects for identifying people at risk."

According to the authors, their study "genotyped' the single nucleotide polymorphism (SNP, or "snip") rs78378222 in TP53, an important tumor suppressor gene. The researchers said the SNP disrupts the TP53 signal and, because of its activity, has been linked to a variety of cancers. This study linked the presence of the rare form of rs78378222 to deadly glioma.

The researchers conducted a large, clinic-based, case-control study of

individuals age 18 and older with a recent glioma diagnosis. A total of 566 glioma cases and 603 controls were genotyped for the rs78378222 variant.

Study results reveal that the odds of developing glioma were increased 3.5 times among the rare variant allele carriers. However, when researchers examined the impact of rs78378222 on survival, they found an approximately 50 percent reduction in death rates for those who were variant allele carriers.

"That the variant increased survival chances was an unexpected finding," Egan said. "It is tempting to speculate that the presence of the risk allele could direct tumor development into a less aggressive path."

The researchers concluded that their study results "may shed light on the etiology and progression of these tumors."

In addition to researchers from Moffitt, researchers from The University of Alabama at Birmingham, Emory School of Medicine and Vanderbilt University participated in the study and co-authored the paper.

Provided by H. Lee Moffitt Cancer Center & Research Institute