

UPMC-developed test rapidly, accurately profiles genetics and treatment of brain tumors

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Brain tumors can be rapidly and accurately profiled with a next-generation, gene-sequencing test developed at UPMC and the University of Pittsburgh School of Medicine.

The test, called Glioseq, is now being used by UPMC oncologists to help guide treatment planning of brain cancers, said senior investigator Marina Nikiforova, M.D., professor of [pathology](#), Pitt School of Medicine, and director of UPMC's Molecular & Genomic Pathology Laboratory. Her team's findings about the test were recently published in *Neuro-Oncology*.

"The diagnosis of [brain tumors](#) has been based primarily on cellular features seen under the microscope," Dr. Nikiforova said. "However, patients with tumors that look identical may experience different clinical outcomes and responses to treatment because the underlying genetic characteristics of their tumors differ. We designed this panel to quickly identify those traits from very small biopsies of the brain lesion."

For the study, the researchers used Glioseq to test 54 adult and pediatric brain tumor samples for genetic abnormalities, including point mutations, gene fusions, and small gene insertions and deletions that had already been characterized by other means. They used next-generation sequencing to simultaneously identify all previously known alterations, as well as many additional genetic markers in these tumors. This provided important information on classification of these tumors, and on possible new targets for therapy.

"This can help guide the physician and the patient in planning treatment, since the molecular information allows us to more precisely characterize tumors and more confidently predict survival and response to therapy. In addition,

Glioseq facilitates the identification of clinical trial options with the appropriate molecular targets, as well as cases in which molecularly targeted drugs are available," said co-investigator Frank Lieberman, M.D., professor of neurology, neurosurgery and medical oncology at Pitt and director of the Adult Neuro-Oncology Program at UPMC CancerCenter, partner of the University of Pittsburgh Cancer Institute.

"Using Glioseq helps us to understand in detail the genetic profile of brain tumors, and takes us one step closer to personalized management of our patients," Dr. Nikiforova said. "We are also working on further improving this test to include additional, recently discovered molecular alterations."

Other research team members include Abigail I. Wald, Ph.D., Melissa A. Melan, Ph.D., Somak Roy, M.D., Shan Zhong, Ph.D., Ronald L. Hamilton, M.D., Jan Drappatz, M.D., Nduka M. Amankulor, M.D., Ian F. Pollack, M.D., and Yuri E. Nikiforov, M.D., Ph.D., all of UPMC and Pitt; and Craig Horbinski, M.D., Ph.D., of Northwestern University.

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Dr. Nikiforova was part of the team that developed ThyroSeq, a next-generation sequencing [test](#) that identifies certain gene mutations that are indicative of an increased likelihood of thyroid cancer.

Provided by University of Pittsburgh Schools of the Health Sciences

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