

More accurate diagnosis for leading cancer killer in children may be possible

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Brain cancer is the leading cause of cancer death in children. Now a more accurate diagnosis of childhood brain cancers may soon be possible, according to researchers at Huntsman Cancer Institute (HCI) at the University of Utah. The information is published online today in the journal *Cancer Research*.

"Researchers already know cancerous tumors often lack certain copies of [genes](#). They also know certain cancer-promoting genes are mutated in cancer patients," says Joshua Schiffman, M.D., an HCI investigator. "But what's significant about this new research is that we've shown these two events occur simultaneously in a unique set of pediatric [brain tumors](#) — a finding previously unknown in these pediatric patients."

Schiffman, along with researchers from several other institutions including Stanford, Washington University, and UCSF, studied the tumors of pediatric [brain cancer](#) patients. Brain tumors are typically classified into grades 1-4 at the time of diagnosis based on their appearance under the microscope. Schiffman and colleagues studied samples from each brain tumor grade using new technology to detect either missing copies or extra copies of DNA. They also looked for mutations, or changes, in the DNA from the same brain tumor samples that can cause improper functioning of genes resulting in cancer. Researchers discovered genetic differences in the different tumor grades that may help explain tumor development and could lead to more accurate diagnosis and categorization of patients. While more research is needed, Schiffman believes these findings can eventually lead to more

targeted and individualized treatments.

The research focuses on BRAF, a gene known to be commonly affected in low-grade brain tumors called astrocytomas. Researchers studied more than 40 of these pediatric astrocytomas — the most common form of brain cancer in children — and found that five out of seven grade 2-4 astrocytomas with BRAF mutations occurred in combination with a deletion in CDKN2A, another gene associated with cancer. The findings suggest these combined alterations define a subset of pediatric malignant astrocytomas.

According to the Pediatric Brain Tumor Foundation, every day, nine children in the U.S. are diagnosed with a brain tumor. Brain tumors are the leading cause of cancer death from childhood cancer, accounting for 24 percent of cancer-related deaths. Pediatric brain tumors are different from those in adults and are often treated differently.

"A lot of progress has been made in our understanding of adult brain cancers, but we don't know as much about the genetics of pediatric brain cancers, which are the number one cancer killer of children," says Schiffman. "This information sheds new light in an area where little information was known. The ability to recognize unique subsets of tumors based on their genetic make-up could someday lead the way to more individualized treatments for pediatric brain cancers."

Provided by University of Utah Health Sciences

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