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More Accurate Diagnosis for Leading Cancer Killer in Children May Be Possible

SALT LAKE CITY, January 12, 2010 — 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.”

Schiffman is an assistant professor in the Division of Pediatric Hematology/Oncology, Department of Oncological Sciences at the University of Utah. He is also affiliated with Primary Children’s Medical Center in Salt Lake City.
The mission of Huntsman Cancer Institute (HCI) at The University of Utah is to understand cancer from its beginnings, to use that knowledge in the creation and improvement of cancer treatments, to relieve the suffering of cancer patients, and to provide education about cancer risk, prevention, and care. HCI is a National Cancer Institute-designated cancer center, which means that it meets the highest national standards for cancer care and research and receives support for its scientific endeavors. HCI is also a member of the National Comprehensive Cancer Network (NCCN) a not-for-profit alliance of the world’s leading cancer centers, which is dedicated to improving the quality and effectiveness of care provided to patients with cancer. For more information about HCI, please visit www.huntsmancancer.org.

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