MedicalXpress

Research gives new hope to those with rare vascular cancer

August 31 2011

A specific genetic alteration has been discovered as a defining feature of epithelioid hemangioendothelioma (EHE), a rare but devastating vascular cancer. These findings have also been used to develop a new diagnostic test for this blood vessel disease.

An international research effort led by Brian Rubin, M.D., Ph.D., of Cleveland Clinic's Pathology and Laboratory Medicine Institute and Lerner Research Institute, devised an innovative approach to reveal the genetic alteration thought to cause EHE, which is considered uncommon: it comprises less than one percent of all cancers. There are approximately 100 new cases in the U.S. each year.

The genetic aberration was detected in 89 percent (42 of 47) of EHE tumor tissues examined; none of the non-EHE vascular tumors contained the anomaly. The research, published in the Aug. 31, 2011, issue of *Science Translational Medicine*, was done in collaboration with Cleveland Clinic's Taussig Cancer Institute.

The authors defined the genetic aberration as a "translocation" between chromosomes 1 and 3, where chromosomes 1 and 3 exchange DNA fragments in such a way that the DNA is "transposed" onto opposite chromosomes. The result is that the swapped DNA encodes a unique, fused gene that contains components from each chromosome. Since genes are translated into proteins, the result of this unique gene is a correspondingly unique protein, whose function is deduced to be oncogenic (cancer-causing).

"This finding is the beginning of a new era for patients with EHE," said Dr. Rubin. "We firmly believe that the characterization of this genetic translocation will lead to a cure for EHE patients."

Identification of translocations in cancers is critical to understanding the molecular pathways at work within cancer cells. Understanding these molecular pathways allows cancer researchers to target them with specific drugs to disrupt the cancer. Thus, finding this translocation in EHE is a gateway to curing this cancer; using the techniques reported here may likewise provide breakthroughs for other cancers as well.

Provided by Lerner Research Institute