

Children's cancer test may produce false positives

January 24 2014, by Josh Barney

(Medical Xpress)—A test used to detect a type of cancer in children may produce false positives because it is based on a faulty assumption, researchers at the University of Virginia School of Medicine have determined.

The test works by detecting a [gene fusion](#) thought to be unique to a rare form of cancer known as alveolar rhabdomyosarcoma. But the new U.Va. research shows that the fusion actually occurs during normal cellular development as well. The fusion of genes lasts only a brief time during normal development, but a diagnostic test could potentially detect it and indicate the presence of cancer where there is none, the researchers believe.

"Cancer does express this fusion. But it's not totally unique now. It's not black and white now. It's shades of gray," said researcher Hui Li of the School of Medicine's Department of Pathology and the U.Va. Cancer Center. "In normal development, it's only briefly expressed, so if we can understand the normal process a little better and know when exactly in development you see this, maybe we can develop a better assay to rule out this particular potential for false positives."

The findings serve as a word of caution for doctors evaluating children for alveolar rhabdomyosarcoma, a type of cancer of the muscle tissue that occurs most commonly among teenagers. While the new findings raise the possibility that a test could possibly detect a false positive, the test continues to have value, as doctors have multiple methods they can

use to diagnose and confirm the presence of rhabdomyosarcoma. The most common [soft tissue cancer](#) in children, rhabdomyosarcoma accounts for about 3 percent of all childhood cancers and about 350 new cases each year in the United States, according to the American Cancer Society.

Li and his team of researchers suspected the gene fusion might occur during normal muscle development, but knew that they faced a challenge in demonstrating that was the case.

"The general belief that we had in the lab is that [cancer](#) doesn't usually have something totally unique," Li said. "It doesn't come up with something totally new – it hijacks a normal process."

But how were they to find a gene fusion that might exist only fleetingly, if it occurred at all?

"Unless you look at the right cell at the right time, you never find these sort of things," Li said. "Without knowing when, we started from stem cells and just harvested time points along the muscle differentiation process. And lo and behold, we see the same fusion that everyone thinks is unique in this particular children's muscle tumor also shows up during muscle cell formation."

The research findings have been published by *Cancer Discovery*, a journal of the American Association for Cancer Research.

Provided by University of Virginia

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