

Newly identified brain cancer mutation will aid drug development

June 1 2014

A collaborative effort between Duke Medicine researchers and neurosurgeons and scientists in China has produced new genetic insights into a rare and deadly form of childhood and young adult brain cancer called brainstem glioma.

The researchers identified a genetic mutation in the [tumor cells](#) that plays a role in both the growth and the death of a cell. Additionally, the mutation to the newly identified gene may also contribute to the tumor's resistance to radiation.

The findings, published online in the journal *Nature Genetics* on June 1, 2014, provide both immediate and long-term benefits. Knowing that this mutation may render radiation ineffective, patients could be spared that therapy. The mutation would also serve as a strong candidate for drug development.

The researchers conducted genetic tests and found that many of the tumor cells had a mutation in a gene called PPM1D, which causes cells to proliferate and avoid natural death. It is the first time this mutation has been found to be a major driving force in the development of brainstem gliomas; it is not evident in other brain tumors.

If tumors have this PPM1D mutation, they do not have another more common genetic mutation to the TP53 gene, a tumor suppressor that, when defective, is linked to half of all cancers.

"This finding has immediate clinical applications, because either mutation - PPM1D or TP53 – cause the tumor cells to be resistant to radiation," said senior author Hai Yan, M.D., Ph.D., a professor of pathology at Duke University School of Medicine. "Knowing that could spare patients from an ineffective treatment approach."

Additionally, the PPM1D genetic mutation is a strong candidate for new drug development.

"This finding gives us a clue as to why these particular tumors are growing inappropriately," said co-author Zachary Reitman, M.D., Ph.D., a research associate at Duke. "These clues may help us to design better treatments for this type of cancer."

Yan said his lab is working to identify new treatments that could target the PPM1D genetic mutation and shut down its cancer-growing capabilities.

"PPM1D is itself a target for drug development, because the [gene mutation](#) causes cells to avoid death and proliferate," Yan said. "In [drug development](#), it's easier to turn that growth function off than it is to switch on the cell's defective tumor suppression mechanism."

More information: Paper: Exome sequencing identifies somatic gain-of-function PPM1D mutations in brainstem gliomas, [DOI: 10.1038/ng.2995](#)

Provided by Duke University Medical Center

Citation: Newly identified brain cancer mutation will aid drug development (2014, June 1) retrieved 17 April 2024 from

<https://medicalxpress.com/news/2014-06-newly-brain-cancer-mutation-aid.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.