MedicalXpress

Possible new heritable marker for retinoblastoma

January 26 2010

Retinoblastoma is a pediatric eye cancer initiated by the loss or mutation of both copies of the retinoblastoma gene. Current evidence suggests that additional genetic alterations are required for retinoblastoma to become fully malignant.

Researchers working at the Sbarro Institute for Cancer Research and Molecular Medicine at Temple University in Philadelphia, PA and at the University of Siena in Siena, Italy, have shed light on the possible role of inactivation of the 16INK4A gene in the progression of retinoblastoma. Their study results appear in the latest edition of the *Journal of Cellular Physiology*.

"The finding that the expression of p16INK4A was reduced both in patients and their parents in our samples suggests that this alteration could be a novel marker of an inheritable susceptibility to retinoblastoma in young patients," said Antonio Giordano, M.D Ph.D., director of the Sbarro Institute and the Center for Biotechnology at Temple, the lead author of the study.

Researchers chose to investigate the 16INK4A gene because of its suspected role in the development of retinoblastoma, as well as its involvement in a predisposition to familial cancer.

The study examined blood samples taken from 29 patients and their parents. They found low to moderate 16INK4A protein expression in 5 of 11 (45%) retinoblastoma tumor specimens. They also found reduced

p16INK4a RNA expression in blood, correlated with the demethylation, or reduction, of the p16INK4a gene, in 16 of 29 (55%) of retinoblastoma patients relative to normal controls.

"Intriguingly, the researchers also found reduced expression in at least one parent among 9 of the 16 (56%) patients with reduced p16INK4a RNA expression. Dr. Giordano and his coworkers suggest that this finding could represent a marker for retinoblastoma susceptibility," said Joan O'Brien, Chair of the Department of Ophthalmology at the University of Pennsylvania and Director of the Scheie Eye Institute. "Confirmation of this hypothesis by future studies would enhance our understanding of genetic and epigenetic events contributing to this disease."

Provided by Sbarro Health Research Organization