Researchers discover hereditary predisposition of melanoma of the eye

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Ohio State University researchers have discovered a hereditary cancer syndrome that predisposes certain people to a melanoma of the eye, along with lung cancer, brain cancer and possibly other types of cancer.

The hereditary cancer syndrome is caused by an inherited mutation in a gene called BAP1, researchers say.

The findings suggest that BAP1 mutations cause the disease in a small subset of patients with hereditary uveal melanoma and other cancers.

Uveal melanoma is a cancer of the eye involving the iris, ciliary body, or choroid, which are collectively known as the uvea. These tumors arise from the pigment cells, also known as melanocytes that reside within the uvea giving color to the eye. This is the most common type of eye tumor in adults.

The findings are reported in the Journal of Medical Genetics.

"We are describing a new cancer genetic syndrome that could affect how patients are treated," said first author Dr. Mohamed H. Abdel-Rahman, researcher at the Ohio State University Comprehensive Cancer Center – Arthur G. James Cancer Hospital and Richard J. Solove Research Institute. "If we know that a patient has this particular gene mutation, we can be more proactive with increased cancer screenings to try to detect these other potential cancers when they are beginning to grow."

Study leader Dr. Frederick H. Davidorf, professor emeritus of ophthalmology at Ohio State University, explained that BAP1 seems to play an important role in regulating cell growth and proliferation, and that loss of the gene helps lead to cancer.

"If our results are verified, it would be good to monitor these patients to detect these cancers early when they are most treatable," said Davidorf, who treats ocular oncology patients at Ohio State along with researcher and physician Dr. Colleen Cebulla.

The study involved 53 unrelated uveal melanoma patients with high risk for hereditary cancer, along with additional family members of one of the study participants. Of the 53 patients in the study, researchers identified germline variants in BAP1 in three patients.

"We still don't know exactly the full pattern of cancers these patients are predisposed to, and more studies are needed," said Abdel-Rahman, also an assistant professor of ophthalmology and division of human genetics at Ohio State University College of Medicine.

"So far, we've identified about six families with this hereditary cancer syndrome. We are working with researchers at Nationwide Children's Hospital to develop a clinical test to screen for the BAP1 gene mutation," he said. "Families with this cancer syndrome should be screened for inherited mutations that increase their risk for developing several other cancers."

Provided by The Ohio State University