

# Researchers discover gene mutation that causes eye cancer

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A University of British Columbia geneticist has discovered a gene mutation that can cause the most common eye cancer - uveal melanoma.

Catherine Van Raamsdonk, an assistant professor of medical genetics in the UBC Faculty of Medicine and a team of researchers, have discovered a genetic mutation in a gene called GNAQ that could be responsible for 45 per cent of the cases of uveal melanoma.

The findings, published today in *Nature*, will allow researchers to develop therapeutic interventions against some melanomas.

"We discovered that GNAQ regulates melanocyte survival," says Van Raamsdonk. "When the GNAQ gene is mutated it leads to unregulated growth of melanocytes. Since cancer is a disease of unregulated cell growth, our findings led us to the discovery that a genetic mutation of the GNAQ gene causes uveal melanoma."

Uveal melanoma is a cancer arising from melanocytes located in the uveal tract. The uveal tract is one of the three layers that make up the wall of the eye. A melanoma is unregulated growth of melanocytes. Melanocytes are also found in the skin and are cells linked to a life-threatening form of skin cancer.

The mutation to GNAQ leads to the activation of a signaling pathway that has previously been implicated in many other types of melanoma. The researchers also found that this mutation is a key factor in the

development of a type of benign skin mole - blue naevi.

"Prior to our work, the mutations responsible for uveal melanoma were completely unknown," says Van Raamsdonk. "No other research looked at mutations in GNAQ. The next step is to develop an effective treatment by targeting the specific biological processes that this mutated gene controls."

Uveal melanoma, the most common eye cancer, affects one in 13,000 people. It is a highly aggressive cancer without any effective treatment options once it metastasizes. Although it only accounts for approximately five per cent of all melanomas, it represents the most common eye cancer in the United States.

Source: University of British Columbia

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