

Study finds genetic testing motivates behavior changes in families at risk for melanoma

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Lisa G Aspinwall, Ph.D., Huntsman Cancer Institute at the University of Utah; Sancy Leachman, MD, Ph.D., Knight Cancer Institute; Tammy K. Stump, Ph.D., Oregon Health and Science University Credit: Huntsman Cancer Institute; Northwestern University; Oregon Health and Science University

Skin cancer is the most commonly diagnosed cancer in the United States, and melanoma is the most severe type of skin cancer. The National Cancer Institute estimates more than 96,000 new cases will be diagnosed

this year, and the disease will cause more than 7,000 deaths. Utah has a particularly high melanoma rate. A new study led by researchers at Huntsman Cancer Institute (HCI) at the University of Utah (U of U) and collaborators at Northwestern University (NW) and Oregon Health and Science University (OHSU) investigated whether genetic testing would motivate people at risk of developing melanoma to alter their behavior in order to reduce their risk. The study was published today in *Genetics in Medicine*.

"We are trying to understand whether a genetic test result adds value over and above what can be achieved by patient counseling alone," said study co-author Lisa G. Aspinwall, Ph.D., HCI researcher and professor of psychology at the U of U. "A genetic test result is concrete and highly personalized. We thought this would be more motivating for difficult behavior change than information about risk based on [family history](#) alone."

The Utah Behavior, Risk Information, Genealogy, and Health Trial (BRIGHT) study focused on families with a high risk of [melanoma](#). Individuals enrolling in the study had three or more [family members](#) diagnosed with melanoma. Participants between the ages of 16-70 were recruited from melanoma-prone families of two types: families with a known cancer-causing gene called CDKN2A and families with comparably high rates of melanoma but no identified CDKN2A mutation. Researchers at the U of U previously discovered that individuals who carry an inherited mutation in the CDKN2A gene are rare, but those individuals have a risk of up to 76 percent of developing melanoma in their lifetime. Co-author Sancy Leachman, MD, Ph.D., director of the melanoma research program at Knight Cancer Institute and professor of dermatology at OHSU, explains, "All melanoma has a strong genetic component, but individuals with a strong [family](#) history are at extremely high risk. They are ideal candidates for prevention and early detection measures. Making a few relatively simple changes could

save their lives."

Each participant received individual counseling from a licensed genetic counselor at HCI. These sessions consisted of a review of family medical history and education about melanoma risk factors, including exposure to environmental ultraviolet radiation (UVR) and genetic predisposition. Participants also received pretest counseling and basic information about melanoma and [genetic testing](#). Members of families known to carry the CDKN2A mutation then were assessed through clinical genetic testing, while subjects from families with no known CDKN2A mutations received information about risk based on family history alone. All participants received identical recommendations for reducing sun exposure.

The multidisciplinary BRIGHT research team included genetic counselors, psychologists, a dermatologist, photobiologists, and an atmospheric scientist. The team examined changes in sun exposure following genetic counseling and test reporting. They used objective measures to track participants, including a special wristwatch-like device to measure UVR and a laser that measures skin color to assess the degree of tanning.

The BRIGHT study results showed genetic counseling about highly elevated melanoma risk, both with and without test reporting, led to sustained reductions in UVR exposure. Additionally, the results provide evidence of a unique benefit to participants who received genetic testing—those who learned they carry the CDKN2A mutation showed reduced exposure to daily UVR the month following genetic counseling, and they showed lighter skin pigmentation one year later.

"The results support the use of melanoma genetic testing to motivate people to adopt risk-reducing behaviors," Aspinwall concluded.

"Previously, it was thought genetic testing wouldn't matter because

members of high-risk families already knew about their risk and were already being advised to reduce their sun exposure. Our study shows that genetic testing, paired with counseling about familial risk and its management, can be a useful tool to motivate cancer prevention behavior."

Co-author Tammy K. Stump, Ph.D., NW researcher in the department of preventive medicine, added, "We are especially confident in these results because we were able to use state-of-the-art objective measures of sun exposure. It's not simply that those with the CDKN2A mutation wished to limit sun exposure—information about personal risk resulted in significant reductions in the levels of [sun exposure](#) during the year following genetic counseling."

Researchers say their next goal is to better understand how knowledge of having a high risk for cancer leads to health-promoting changes in behavior rather than leading to debilitating responses. They also seek to improve education so the public understands a suntan is a sign of skin and DNA damage that can lead to melanoma.

Findings from this study and future work will help guide the development of counseling techniques, including optimal follow-up and integration into a patient's routine healthcare program. Dr. Stump noted, "Genetic testing is becoming more common in healthcare, but we still don't know a lot about how and when to deliver this information in a manner that promotes healthy behavior changes. This study is an example of the type of research needed to guide decisions about genetic test disclosure in clinical settings."

Provided by Huntsman Cancer Institute

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