

Researchers close in on new melanoma gene

It has long been known that prolonged exposure to the sun's harmful UV rays can lead to melanoma, the deadliest form of skin cancer. An unanswered question, however, is why some people are more likely to develop melanoma than others. Despite years of research and clinical development, melanoma incidences continue to rise around the world. According to the National Cancer Institute, the percentage of people in the United States who develop melanoma each year has more than doubled in the past 30 years.

Results published today from a study led by researchers from The Translational Genomic Research Institute (TGen) in Phoenix, Arizona and The Queensland Institute of Medical Research (QIMR), Queensland, Australia, however, may yet change these statistics. The team is close to discovering a new gene that could help explain variation in melanoma risk.

In a report appearing in an Advance Online Publication (AOP) of the journal *Nature Genetics*, the researchers and their colleagues identify a region on chromosome 20 (20q11.22) that influences a person's risk of developing melanoma.

According to Dr Kevin Brown, TGen Investigator and the paper's co-first author, compared to other genetic research focusing on familial (or inherited) cases of melanoma, this finding holds implications for the general population

"We're closing in on genetic variants which cause 16 percent of the population to be at nearly double the increased risk of developing the disease. In public health terms, this finding is highly significant," Dr. Brown said.

The researchers narrowed the gene location through a genome-wide association study — a first in melanoma research. Genome-wide studies involve rapidly scanning DNA of many people to find genetic variations associated with a particular disease. After identifying new genetic associations, researchers can use the information to develop better strategies to detect, treat and prevent the disease.

"The aim of our work is to identify and understand the genetic factors influencing melanoma so we can better predict risk estimates," said QIMR's Dr. Stuart MacGregor, co-first author on the study. "This in turn, means people will be better informed and can take the right precautions to avoid developing this increasingly common cancer."

How The Study Works

A genome-wide association study involves scanning the DNA from two sets of individuals: those with a particular disease (cases) and those of similar individuals without the disease (controls). The DNA of each sample then undergoes examination for strategically selected markers of genetic variation, called single nucleotide polymorphisms, or SNPs. If certain genetic variations are found to be significantly more frequent in people with the disease compared to people without disease, those variations are said to be "associated" with the disease. The associated genetic variations can serve as roadmaps to the region of the human genome where the disease-causing gene resides.

The study — whose data collection began 20 years ago — involved more than 4,000 Australian samples (2,019 cases and 2,105 controls) and was a joint project between Australian, American and European research groups.

Source: The Translational Genomics Research Institute

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