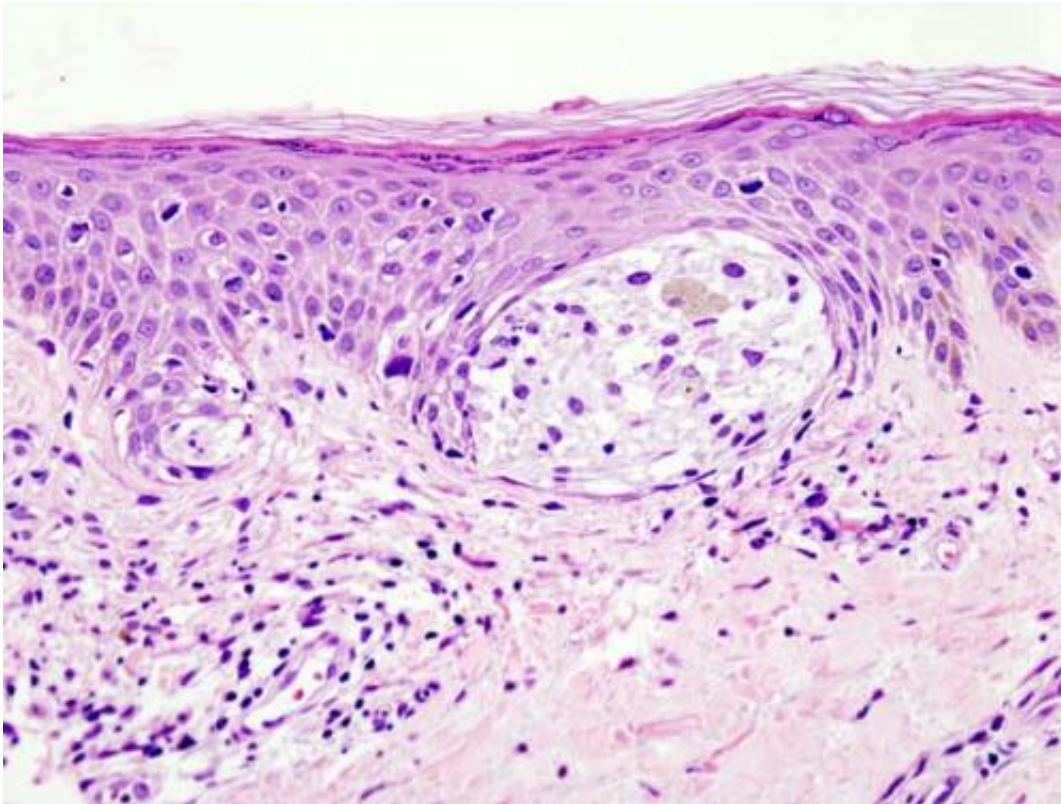


Scientists double understanding of genetic risk of melanoma

April 27 2020



Melanoma in skin biopsy with H&E stain—this case may represent superficial spreading melanoma. Credit: Wikipedia/CC BY-SA 3.0

A global collaboration of scientists has more than doubled the known number of regions on the human genome that influence the risk of developing melanoma.

The research, involving the University of Leeds in the UK, QIMR Berghofer in Australia, and the National Cancer Institute in the US, has been published today in the journal *Nature Genetics*.

Melanoma is a sometimes-deadly skin cancer, with an estimated 350,000 cases worldwide in 2015, resulting in nearly 60,000 deaths.

The joint study leader and QIMR Berghofer statistical geneticist, Associate Professor Matthew Law, said the researchers identified 33 new regions of the genome and confirmed another 21 previously reported regions that are linked to a person's risk of developing [melanoma](#) of the skin.

"By finding new regions we can now narrow in on the specific underlying genes and better understand the pathways that lead to melanoma," Associate Professor Law said.

"Two of the new regions we've discovered that are linked to melanoma have previously been linked to autoimmune disorders. This provides further evidence that the immune system plays an important role in a person developing melanoma.

"We also found an association between melanoma and common genetic variants in the gene TP53, which is a gene critical in controlling DNA repair when cells divide, and in suppressing cancer."

The UK based co-lead author, Dr. Mark Iles from the University of Leeds' Institute for Data Analytics, said the researchers examined DNA from 37,000 people who had been diagnosed with melanoma and compared their genetic information to that of nearly 400,000 people with no history of the disease.

"The large population sample made it possible to recognise which

regions of the genome were active in people with melanoma," Dr. Iles said.

"The population sample we used is three times larger than any previous genetic study on melanoma risk and gives us strong confidence that the new regions we've discovered all play a role in the disease.

"It's a product of power in numbers. The only way to discover these things is by having such a large study population that spans across the globe, and we'd urge more people to sign up for these large melanoma research projects."

Melanoma begins in melanocytes, cells in the skin responsible for making the pigment melanin that gives colour to the skin.

Melanin can block some of the harmful effects of UV radiation, which is why people with pale skin are at a higher risk of skin cancer, but the protection is not complete.

Moles also develop from melanocytes and having a high number of moles is a risk factor for melanoma.

Dr. Maria Teresa Landi, co-lead author on the study and senior investigator at the US National Cancer Institute, part of the National Institutes of Health, said the research also uncovered other important clues to the genetic causes of melanoma.

"We used the relationship between moles, pigmentation, and melanoma to identify 31 additional gene regions that potentially influence melanoma risk. For example, one of the regions we identified is involved in melanocyte growth," Dr. Landi said.

"Moreover, we also included people from Mediterranean populations

involved in the MelaNostrum Consortium. Most studies of melanoma use people with northern or western European ancestry (e.g. British) and by expanding our analysis to include Mediterranean populations, we will gain a greater understanding of the genetics of melanoma in this highly sun-exposed group.

"Larger datasets are needed to find further genes that influence melanoma risk, and in parallel we need functional studies to work out the exact mechanisms that lead to these genes causing melanoma."

The researchers said the study reaffirmed the importance of protecting skin from the sun by avoiding UV exposure between the hours of 10 AM and 4 PM, seeking shade, covering up exposed [skin](#), and using sunscreen.

More information: Genome-wide association meta-analyses combining multiple risk phenotypes provide insights into the genetic architecture of cutaneous melanoma susceptibility , *Nature Genetics* (2020). [DOI: 10.1038/s41588-020-0611-8](https://doi.org/10.1038/s41588-020-0611-8) , www.nature.com/articles/s41588-020-0611-8

Provided by University of Leeds

Citation: Scientists double understanding of genetic risk of melanoma (2020, April 27) retrieved 24 April 2024 from <https://medicalxpress.com/news/2020-04-scientists-genetic-melanoma.html>

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