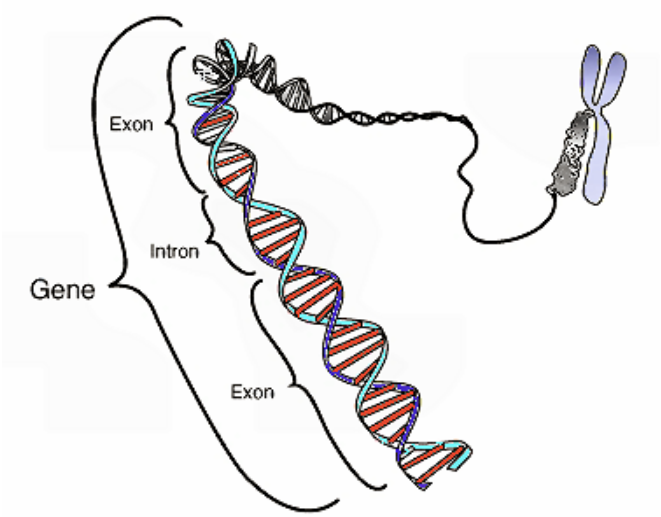
Pale melanomas masked by albino gene

23 September 2020



This image shows the coding region in a segment of eukaryotic DNA. Credit: National Human Genome Research Institute

People with pale colored melanomas are more likely to have a gene mutation associated with albinism, University of Queensland research has found.

Study lead author Dr. Jenna Rayner said albinism, a rare genetic disorder affecting one in 10,000 people, prevented brown pigment from being synthesized in the body and led to fair hair and extremely pale skin that was easily sunburned and prone to skin cancers.

"Albinism develops when there are two mutated genes, so people with one mutation usually don't know they have it," Dr. Rayner said.

"These people may be more prone to developing pale colored melanomas, called amelanotic, because tumors accumulate new mutations, and they already have a mutated albinism gene."

The researchers studied DNA samples from more than 380 volunteers using whole exome sequencing, while looking for rare genetic mutations that cause albinism.

Queensland has the highest rate of melanoma in the world and more than 14,000 cases are diagnosed in Australia each year.

UQ Dermatology Research Centre Associate Professor Rick Sturm said up to eight percent of melanomas could be amelanotic, making them difficult to diagnose and easily mistaken for non-cancerous conditions like warts or scars.

"Amelanotic melanomas are normally diagnosed in advanced stage, compared with darker melanomas, causing patients to often miss out on early treatment and their best chance of a cure," he said.

When funding becomes available, researchers plan to collect amelanotic melanoma samples to compare their genotype with that of the patient.

Dr. Rayner said it could lead to personalized medicine—where doctors would be alerted to monitor potential amelanotic melanomas in people with one albinism gene mutation.

"This could optimize early intervention and consequently improve patient outcomes," she said.

The paper was published in *PLOS ONE*.

More information: *PLOS ONE* (2020). DOI: 10.1371/journal.pone.0238529

Provided by University of Queensland