

Genetic variants for skin color in African Americans linked to vitamin D deficiency

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One day, physicians may be able to look at an African American's skin color, and with the help of other determinants, know if prescribing vitamin D supplements would lower that person's risk of getting cancers of the prostate, colon, rectum or breast.

"We should not shy from this new study looking at the genetics of skin color and its effects on vitamin D deficiency because being 'colorblind' is what has led to the widespread health disparities that we as a society are now trying to address," said Rick Kittles, Ph.D., director of the Division of Health Equities at Beckman Research Institute of City of Hope, a world-renowned independent research and treatment center for cancer, diabetes and other life-threatening diseases.

"Skin color has strong social and biological significance—social because of race and racism and biological because over 70% of African Americans are vitamin D deficient, resulting in increased risk for cancer and cardiovascular disease," Kittles added. Notably, the difference in cancer death rates between African Americans and in the sun than people with lighter skin.

whites is 14%.

Researchers in the City of Hope-led data study, published Feb. 18 in PLOS Genetics, conducted a genome-wide association study using the data of 1,076 African Americans to analyze the genetics of skin pigmentation in this group, replicate results and test if the identified genetic variants are linked to vitamin D deficiency in African Americans.

This was the first genome-wide association study of skin pigmentation in African Americans, Kittles said. Study participants self-identified as African American. Blood samples for DNA analysis and vitamin D levels were collected at recruitment, and scientists measured the sun-protected area of the skin in the inner upper arm of participants using a digital reflectometer.

Although skin pigmentation was measured in an area of the body unexposed to the sun, various factors such as aging, outdoor activities and consistent UV exposure over the years may influence skin pigmentation and the association between skin pigmentation and vitamin D levels. Understandably, researchers found that skin pigmentation gene variants, rather than skin pigmentation, measured using a reflectometer were associated with serum vitamin D levels.

Scientists found three regions (SLC24A5, SLC45A2 and OCA2) in the genes of African Americans with strong links to skin color and severe vitamin D deficiency. The genetic variant rs2675345, which is near a region in the gene called SLC24A5, showed the strongest association with skin pigmentation and vitamin D deficiency.

Studies have shown that individuals with darker skin pigmentation require longer or more intense ultraviolet radiation exposure to synthesize sufficient levels of vitamin D. In other words, if you have darker skin, you tend to make less vitamin D



Kittles and his colleagues are exploring how they can one day leverage their newly identified risk score in doctors' offices—potentially creating a precision medicine tool. For example, depending on skin tone, occupation and lifestyle, doctors can better prescribe the correct dose of vitamin D supplementation.

"This study is an example of the interplay of race and skin color on health and how if we ignore things such as the color of a person's skin, we may be ignoring potential medical issues, thus contributing to health care disparities," Kittles said. "Our study provides new knowledge about an easily modifiable factor such as vitamin D supplementation and inherited genetic factors affecting vitamin D deficiency in African Americans. With more research, in the future doctors could offer patients of color with an inexpensive way to reduce their risk of vitamin deficiency, which ultimately could help protect against certain cancers."

"Genetic loci associated with skin pigmentation in African Americans and their effects on vitamin D deficiency" was published in *PLOS Genetics*.

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