

A treatment for one form of albinism?

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Individuals with oculocutaneous albinism, type 1 (OCA1) have white hair, very pale skin, and light-colored irises because they have none, or very little, of the pigment melanin in their skin, hair, and eyes. Affected individuals have impaired eyesight and a substantially increased risk of skin cancer. Current treatment options are limited to attempts to correct eyesight and counseling to promote the use of sun protective measures. A team of researchers, led by Brian Brooks, at the National Eye Institute, Bethesda, has now generated data in mice that provide hope for a new treatment for a subset of patients with OCA1.

OCA1 is caused by mutations in the gene *Tyr* that result in either complete loss of activity of the protein tyrosinase (which is key to the generation of melanin) or the generation of a tyrosinase protein with reduced activity. Brooks and colleagues found that treating mice that model OCA1 caused by mutations that generate a tyrosinase protein with reduced activity (OCA-1B) with nitisinone, which is already FDA-approved for treating a blood condition known as hereditary tyrosinemia type 1, increases their eye and hair pigmentation. They therefore suggest that nitisinone could improve pigmentation in patients with OCA-1B and potentially ameliorate their [vision loss](#).

In an accompanying commentary, Seth Orlow and Prashiela Manga, at New York University School of Medicine, New York, discuss the inspired study of Brooks and colleagues but caution that there are issues that might limit the use of nitisinone as a treatment for OCA-1B.

More information: [www.jci.org/articles/view/5937 ...](http://www.jci.org/articles/view/5937...)

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