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# OCA. Occulocutaneous Albinism

NIHR BioResource – Rare Diseases study project

**Lead Investigators: Dr Helena Lee & Dr Jay Self**

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## Summary

Currently, there are no treatments for the eye problems seen in albinism. The average vision in albinism at 20/80, is below UK driving standards, which has implications for school, work and social life. This is why finding a treatment that can improve eyesight in albinism, was named as a priority by the Sight Loss and Vision Priority Setting Partnership in 2013.

We know that the brain has the amazing ability to change and adapt in children. We also know that we make use of the brain's ability to rewire itself, when we improve eyesight in lazy eyes using glasses and patching. In oculocutaneous albinism (OCA), a chemical called L-DOPA is missing from the eye and this causes problems with eye development. This is why eyesight is so poor in albinism. However, the eye is still able to change and develop in young children with albinism. Similar to the treatment of lazy eyes, we can target this flexibility in albinism. Potentially, replacing L-DOPA in albinism at a young age, will improve eye development and prevent sight loss in albinism.

Prompt diagnosis will be important for optimal treatment of this condition with L-DOPA. However, this is challenging due to the varying presentations of albinism and even a collective of clinical signs and genetic testing may not prove conclusive. We want to collect more information about the condition to help to improve the diagnosis and treatment of people with OCA.

## Recruitment Criteria

### Inclusion

Diagnosis of oculocutaneous albinism (OCA) based on previously established diagnostic criteria (see table below) as follows:

- 3 major criteria OR
- 2 major and 2 minor criteria for the diagnosis of albinism OR
- In the presence of a molecular diagnosis, 1 major criterion or 2 minor criteria for the diagnosis of albinism will be sufficient.

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| Major criteria | ○ Foveal hypoplasia grade 2 or more<br>○ Optic nerve misrouting on visual evoked potential (VEP) testing<br>○ Ocular hypopigmentation, either Iris transillumination defects or fundus hypopigmentation grade 2 or more |
| Minor criteria | ○ Infantile nystagmus<br>○ Cutaneous hypo-pigmentation<br>○ Grade 1 fundus hypopigmentation |
| Molecular diagnosis | ○ A genotype consisting of 1 previously published pathogenic variant in a known OCA gene or 1 novel variant deemed 'highly likely' to be pathogenic & either<br>○ A 2nd known or novel 'highly likely' pathogenic variant in the same gene or<br>○ A 2nd common variant in the same gene, known to be associated with an albino phenotype |

## Exclusion

- Diagnosis of ocular albinism (OA)
- Ocular abnormalities other than those associated with OCA