

## TREATMENT OF OCULOCUTANEOUS/OCULAR ALBINISM AND FOR INCREASING PIGMENTATION

### SUMMARY

The National Eye Institute's Ophthalmic Genetics and Visual Function Branch seeks interested parties to co-develop the use of nitisinone (NTBC) for oculocutaneous albinism or as a treatment for increasing pigmentation in the eyes, hair and/or skin of patients.

### REFERENCE NUMBER

E-113-2010

### PRODUCT TYPE

- Therapeutics

### KEYWORDS

- Ocular disorders
- Albinism
- Pigmentation
- Nitisinone
- achromia, achromasia, achromatosis

### COLLABORATION OPPORTUNITY

This invention is available for licensing and co-development.

### CONTACT

Alan Hubbs

NEI - The National Eye Institute

240-276-5532

[Alan.Hubbs@nih.gov](mailto:Alan.Hubbs@nih.gov)

### DESCRIPTION OF TECHNOLOGY

Albinism (also called achromia, achromasia, or achromatosis) is a congenital disorder characterized by the complete or partial absence of pigment in the skin, hair and eyes due to absence or defect in any one of a number of proteins involved in the production of melanin. Certain forms of albinism are known to be due to mutations in tyrosine metabolism. In oculocutaneous albinism (OCA), pigment is lacking in the eyes, skin and hair. In ocular albinism, only the eyes lack pigment. Patients with albinism experience varying degrees of vision loss associated with foveal hypoplasia, nystagmus, photophobia and/or glare sensitivity, refractive errors, and abnormal decussation of ganglion cell axons at the optic chiasm. Current treatment options for vision problems caused by albinism are limited to correction of refractive

errors and amblyopia, low vision aids, and (in some cases) extraocular muscle surgery.

Nitisinone (NTBC) is an FDA-approved drug used in the treatment of tyrosinemia, type 1. The drug blocks the normal degradation pathway of tyrosine thus allowing greater circulating plasma levels of tyrosine. NEI investigators have identified that administration of NTBC to subjects (e.g., mice or humans) with certain forms of albinism, can result in increased circulating tyrosine levels, an increase in tyrosinase activity, and, subsequently, increased pigmentation.

#### **INVENTOR(S)**

- [Brian P. Brooks](#) (NEI), [William A Gahl](#) (NHGRI), David R Adams (NHGRI)

#### **DEVELOPMENT STAGE**

- Pre-clinical (in vivo)

#### **PATENT STATUS**

- **U.S. Issued:** US 8,822,540
- **Foreign Filed:** WO 02011106655

#### **THERAPEUTIC AREA**

- Eye and Ear