Visual Function in Albinism: contribution of ocular abnormalities and pigmentation.

Published: 25-08-2015
Last updated: 19-04-2024

1. To investigate the relative contributions of different ocular abnormalities to the visual function.2. To investigate the precise relationship between ocular pigmentation and the development of the visual system abnormalities.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Eye disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON42506

Source
ToetsingOnline

Brief title
ViFA

Condition

- Eye disorders congenital
- Pigmentation disorders

Synonym
Albinism, hair and skin, Hypopigmentation of the eyes only or the eyes

Research involving
Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum
Source(s) of monetary or material Support: ODAS Stichting;Landelijke Stichting voor

Blinden en Slechtzienden (LSBS);Vereniging Bartiméus Sonneheerdt

Intervention

Keyword: Albinism, Misrouting, Opticopathy, Visual acuity

Outcome measures

Primary outcome

The presence and severity of opticopathy in patients with albinism with relatively poor BCVA and relatively good BCVA, i.e. less than 0.3 logMar and more than 0.8 logMar, respectively.

Secondary outcome

The difference in clinical and electrophysiological profile between patients with albinism with relatively poor and good BCVA. Furthermore, the difference in clinical and electrophysiological profile of patients with albinism, FHONDA and IIN. Clinical profile includes BCVA, refraction, strabismus, stereopsis, amblyopia, nystagmus, photophobia, iris transillumination, and macula and optic nerve characteristics. Electrophysiological profile includes VEP misrouting and VEP opticopathy (pattern VEP) analysis in detail.

Study description

Background summary

Albinism is a pigmentation disorder that can be restricted to the eyes (ocular albinism) or can involve eyes, hair and skin (oculocutaneous albinism). The ocular abnormalities in albinism comprise nystagmus, iris translucency, foveal hypoplasia, optic nerve abnormalities and misrouting, i.e. an abnormal projection of the visual input in the cerebral cortex. Usually the disorder results in a non-progressive reduced visual acuity. A visual acuity of 0.7 logMAR to 1.0 logMAR is common (0.2 -0.1 Snellen). (Summers, 1996). However, albinism shows a great variety in clinical presentation and patients with a

near normal visual acuity are known. The relative contributions of the different ocular abnormalities to the visual disability are still unkown. To investigate this we will compare patients with albinism on both ends of the spectrum, with relatively poor and relatively good visual acuity.
The ocular abnormalities were considered to be secondary to the lack of pigmentation. However, we question this hypothesis because of the recently discovered FHONDA: Foveal Hypoplasia Optic Nerve Decussation defects and Anterior segments dysgenesis. Patients with this disorder present with the same ocular abnormalities as seen in albinism, but without a lack of pigmentation. All patients with FHONDA have severely reduced visual acuity. Thus, it seems unlikely that the ocular abnormalities in albinism are caused by the lack of pigmentation. In order to give an accurate prognosis for young children with albinism, it is necessary to gain more insight in the precise relationship between ocular pigmentation and the development of the visual system. To this end we will compare patients with albinism with patients with similar ocular abnormalities and normal pigmentation, i.e. patients with FHONDA or Idiopathic Infantile Nystagmus (IIN).

Recently we started a retrospective study of patients with albinism, FHONDA and IIN. We collected clinical, genetic and electrophysiological data from the records of patients with albinism seen at Bartiméus and the Leiden University Medical Center (LUMC) and patients with FHONDA and IIN seen at Bartiméus. Until now approximately 480 patients have been included. Evaluation of these data confirm the large variety in signs and symptoms in patients with albinism. These data will be used in this prospective observational case series as well.

Study objective

1. To investigate the relative contributions of different ocular abnormalities to the visual function.
2. To investigate the precise relationship between ocular pigmentation and the development of the visual system abnormalities.

Study design

The study is a prospective observational case series. We will select patients of 12 years or older from the retrospective study. Selection criteria for the albinism group are a BCVA of more than 0.8 logMAR (less than 0.16 Snellen) or BCVA of less than 0.3 logMAR (more than 0.5 Snellen).
We will invite these patients for a repeat clinical examination, to obtain a complete dataset including VEP for misrouting and an eye-tracker test to quantify nystagmus. If possible, this repeat examination will replace a regular control visit.
We will then compare the clinical data of patients with relatively good BCVA to those with relatively poor BCVA. We will furthermore compare patients with albinism to patients with an overlap in symptoms, but without hypopigmentation,

i.e. patients with FHONDA or IIN. Of the latter patients, most information will be available from patients* records. However, if data are missing or are of insufficient quality, we will invite these patients to Bartiméus as well. All tests will be performed during one visit. Duration of the visit will be approximately 3 hours including VEP testing, or, 1 hour if VEP is not necessary. All investigations will be performed in Bartiméus, because most of the patients are known in Bartiméus and uniformity of the used equipment will lead to better interpretation of the results.
We will invite patients to participate by written invitation in which the procedures and research goals are explained. In case patients visit the clinic for other purposes, we will ask these patients to participate after oral explanation of the research and provide them with additional information in writing. Subjects will only participate after informed consent, of the patients themselves, or, in case of minors, also of their parents or legal guardians.

Study burden and risks

There is no risk to the health of the participants. The burden involving participation in this investigation will be minimal: approximately three hours at Bartiméus for ophthalmological and electrophysiological examination, including a VEP (visual evoked potentials) test, an OCT (optical coherence tomography), and fundoscopic imaging. These investigations are standard clinical procedure for all new patients referred to Bartiméus with a possible diagnosis of albinism. We will only perform electrophysiological tests, OCT and fundoscopic images if they were not obtained earlier or are of insufficient quality for the purpose of this study. The patients will be asked if they want us to share the obtained data with their own ophthalmologist. This means that the visit to Bartiméus may replace a regular control visit. No mydriatic eyedrops will be administered, since dilatation of the pupils won*t be necessary. Patients will be compensated for their travel expenses based on ¤0.19 per kilometre, or public transportation fare based on 2nd class train tickets.

Contacts

Public
Leids Universitair Medisch Centrum

Albinusdreef 2
Leiden 2333 ZA
NL
Scientific
Leids Universitair Medisch Centrum

Albinusdreef 2
Leiden 2333 ZA
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

Patients diagnosed with albinism, Idiopathic Infantile Nystagmus (IIN) or FHONDA from the LUMC and/or Bartiméus.
Patients must be older than 12 years of age.

Exclusion criteria

Patients with another condition besides albinism, FHONDA or IIN that could cause reduced visual function.

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled

Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	05-08-2016
Enrollment:	95
Type:	Actual

Ethics review

Approved WMO

Date:	25-08-2015
Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden) metc-ldd@lumc.nl

Study registrations

Followed up by the following (possibly more current) registration

No registrations found.

Other (possibly less up-to-date) registrations in this register

No registrations found.

In other registers

Register	ID
CCMO	NL53735.058.15

Study results

Date completed:	01-09-2018
Actual enrolment:	50

Summary results
Trial is onging in other countries