Rate of Progression in EYS Related Retinal Degeneration

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1. To report the natural history of retinal degeneration in patients with biallelic mutations in the EYS gene.2. To identify sensitive structural and functional outcome measures to use for future multicenter clinical trials in EYS-related retinal...

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

Summary

ID

NL-OMON49574

Source
ToetsingOnline

Brief title
Pro-EYS

Condition

- Eye disorders congenital
- Congenital eye disorders (excl glaucoma)

Synonym
Retinal dystrophy, retinitis pigmentosa

Research involving
Human

Sponsors and support

Primary sponsor: JAEB Center for Health Research
Source(s) of monetary or material Support: Foundation Fighting Blindness

Intervention

Keyword: EYS mutation, natural history, retinitis pigmentosa

Outcome measures

Primary outcome

Visual field sensitivity measured by static perimetry, best corrected visual acuity, mean retinal sensitivity as measured by fundus guided microperimetry, ellipsoid zone area as measured by spectral-domain optical coherence tomography, retinal function using full-field electroretinography amplitudes and timing in response to rod- and cone-specific stimuli.

Secondary outcome

not applicable

Study description

Background summary

Biallelic mutations in EYS represent a common cause of retinitis pigmentosa. There are at least four isoforms of the EYS gene that are expressed in the human retina. The exact role of EYS is unclear but it has been speculated to be involved in maintenance of the rods and cones. There has not been a strong establishment of correlation between the mutation in the EYS gene and the progression of the disease. The average age of onset has been reported to be approximately twenty years with loss of visual acuity starting around age thirty. Retrospective studies have shown the rate of vision loss in EYS to be more severe than that of mutations caused by USH2A. There are few data available on retinitis pigmentosa patients with mutations in the EYS gene. The Pro-EYS study will conduct a multicenter natural history study in patients with biallelic mutations in EYS with the purpose of better understanding disease progression as well as obtaining preliminary data for potential therapeutic trials in the future.

Study objective

1. To report the natural history of retinal degeneration in patients with biallelic mutations in the EYS gene.
2. To identify sensitive structural and functional outcome measures to use for future multicenter clinical trials in EYS-related retinal degeneration.
3. To identify well-defined subpopulations for future clinical trials of investigative treatments for EYS-related retinal degeneration

Study design

This study is designed as a multicenter longitudinal, prospective natural history study. Patients will be defined to 3 cohorts based on their visual acuity and kinetic visual field.

Study burden and risks

We anticipate that study enrollment will be representative of the population of patients with biallelic mutations in the EYS gene.
Participants do not benefit, risks are considered negligible, procedures are non-invasive and take 3 to 6 hours extra time from patient per visit, one visit per year. It is anticipated that, in the future, patients with EYS-related retinal degeneration will benefit from newly developed therapeutic strategies.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

1. Willing and able to complete the informed consent process
2. Ability to return for all study visits over 48 months if in the natural history study
3. Age >=18 years
4. Have retinal degeneration caused by mutations in the EYS gene, as defined by a clinically certified lab

Exclusion criteria

1. Have other mutations in your DNA that could cause retinal degeneration.
2. Be planning to enter a study testing treatments for retinal degeneration during the time of this study.
3. Have a history of treatment that could have affected the retina.
4. Have had certain eye surgeries that may affect the tests for this study.

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	09-08-2021
Enrollment:	6

Type:	Actual

Medical products/devices used

Generic name:	Octopus 900 Pro
Registration:	Yes - CE intended use

Ethics review

Approved WMO Date:	01-12-2020
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	12-07-2021
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

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
ClinicalTrials.gov	NCT04127006
CCMO	NL74105.091.20