Prospective clinical evaluation of inherited retinal diseases

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Ethical review	Approved WMO
Status	Recruiting
Health condition type	Retina, choroid and vitreous haemorrhages and vascular disorders
Study type	Observational non invasive

Summary

ID

NL-OMON48655

Source
ToetsingOnline

Brief title
IRD prospective

Condition

- Retina, choroid and vitreous haemorrhages and vascular disorders

Synonym
Inherited retinal disease, Stargardt disease

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum
Source(s) of monetary or material Support: Foundation Fighting Blindness USA

Intervention

Keyword: ABCA4, Inherited retinal diseases, Natural history, Stargardt disease

Outcome measures

Primary outcome

Best corrected visual acuity; visual field sensitivity and area measured by static perimetry; fundus autofluorescence intensity data, mean retinal sensitivity as measured by fundus-guided microperimetry; ellipsoid zone area as measured by SD-OCT; retinal function using full-field ERG amplitudes and timing in response to rod- and cone-specific stimuli.

Secondary outcome

Quality of life and patient reported outcomes.

Study description

Background summary

Inherited retinal diseases (IRDs) often cause progressive retinal degeneration due to mutations in one of many genes expressed in retinal cells. Up to now, the vast majority of IRDs are not treatable and therefore patients generally have not been examined at short intervals, and prospective studies on the course of the diseases are lacking. Trials on therapies for IRDs are upcoming, and to assess effectiveness of such therapies we need detailed knowledge on the natural course of these diseases as well as identification of clinical significant biomarkers to highlight disease progression. Visual acuity does not seem to be the optimal biomarker to use in therapeutic trials. The use of structural biomarkers (e.g. fundus autofluorescence imaging, optical coherence tomography or a combination thereof) may show a much more gradual progression that is indicative of functional vision loss at a later stage.

Study objective

The goal of this study is to characterize the natural course of IRDs that can potentially be modulated by future therapy. Second, this study aims to understand the relationship between various structural and functional

biomarkers in potentially therapy-eligible IRD cases which could ultimately lead to the acceptance of structural biomarkers as clinical endpoints.

Study design

Longitudinal, prospective natural history study of IRD cases, with a 6-monthly follow up for a total of 3 years.

Study burden and risks

Participants do not benefit from this study and risks are considered negligible. Procedures are non-invasive and take about 5 hours extra time from patient (and parent) per visit, twice a year. It is anticipated that, in the future, patients with retinal dystrophies will benefit from newly developed therapeutic strategies.

Contacts

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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

Study Participant Inclusion Criteria
Participants must meet the following:
1. Clinical diagnosis of STGD and at least two pathogenic or likely pathogenic mutations in trans in the ABCA4 gene of which at least one therapy-eligible mutation
2. Age >= 16 years
3. Willing and able to complete the informed consent
4. Ability to return for all study visits over 36 months;Ocular Inclusion Criteria
At least one eye of participants must meet the following:
1. Baseline visual acuity ETDRS letter score of 54 or more (approximate Snellen equivalent 20/80 or better)
2. Stable fixation and ability to perform perimetry reliably
3. Clear ocular media and adequate pupil dilation to permit good quality imaging

Exclusion criteria

Study Participant Exclusion Criteria
1. Mutations in genes that cause autosomal dominant or X-linked retinal dystrophy, or presence of biallelic mutations in autosomal recessive retinal dystrophy genes other than the gene studied in the patient cohort.;Ocular Exclusion Criteria
If both eyes have any of the following, the patient is not eligible:
1. Current vitreous hemorrhage
2. Current or any history of rhegmatogenous retinal detachment
3. Current or any history of (e.g., prior to cataract or refractive surgery) spherical equivalent of the refractive error worse than -8 Diopters of myopia
4. History of intraocular surgery (e.g., cataract surgery, vitrectomy, penetrating keratoplasty, or LASIK) within the last 3 months
5. Current or any history of retinal vascular occlusion or proliferative diabetic retinopathy
6. Expected to have cataract removal surgery during the study
7. History or current evidence of ocular disease that, in the opinion of the investigator, may confound assessment of visual function

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):	07-12-2018
Enrollment:	40
Type:	Actual

Medical products/devices used

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

Ethics review

Approved WMO Date:	26-07-2018
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	28-02-2019
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	03-04-2019
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

ID: 20528
Source: NTR
Title:

In other registers

Register	ID
CCMO	NL65175.091.18
OMON	NL-OMON20528