Multifactorial eye diseases

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The purpose of this protocol amendment is to meet the current and future needs of the MacTel Project’s collaborating investigators and continue meeting initial objectives. In addition, a third objective is to understand the mechanisms involved in...

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

Summary

ID

NL-OMON57496

Source
Onderzoeksportaal

Brief title
Multifactorial eye diseases

Condition

- Retina, choroid and vitreous haemorrhages and vascular disorders

Synonym
macular telangiectasia type 2

Research involving
Human

Sponsors and support

Primary sponsor: Lowy Medical Research Institute
Source(s) of monetary or material Support: Lowy medical research institute

Intervention

- No intervention

Explanation

N.a.

Outcome measures

Primary outcome

Not applicable

Secondary outcome

Not applicable

Study description

Background summary

MacTel is a complex genetic disease, with no single gene responsible for the disease. The genetic variants identified through the MacTel Project sequencing efforts to date have been associated with increased risk of developing MacTel. These variants exhibit incomplete penetrance or are not the single factor causing disease. Because of this complex genetic landscape, no individual results can or will be provided to the site investigator or an individual participant regarding a person’s MacTel genetics.

Study objective

The purpose of this protocol amendment is to meet the current and future needs of the MacTel Project’s collaborating investigators and continue meeting initial objectives. In addition, a third objective is to understand the mechanisms involved in the disease etiology. Together, these three objectives work toward the overarching goal of finding possible causes, novel therapeutic treatments, preventions, or a definitive cure for MacTel Type 2

Study design

To accomplish these objectives, the single in-clinic study visit format will be retained. Data will be collected through updated and more detailed medical, ocular and medication histories. Imaging will be done, including additional advanced imaging procedures at sites that have the capability. Genetic sequencing will continue.

Intervention

Not applicable

Study burden and risks

neglectable risk (natural history)

Contacts

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Trial sites

Trial sites in the Netherlands

Radboud Universitair Medisch Centrum
Target size: 300

Listed location countries

United States, Switzerland, Australia, United Kingdom, France, Germany, Netherlands

Eligibility criteria

Age
Adults (18-64 years)

Inclusion criteria

1. Ability to review and understand the informed consent document and agree to the form’s contents. (In cases with significant visual impairment, the informed consent may be read to the participant); 2. Stated willingness to comply with all study procedures; 3. Male or female, aged >18; and 4. Diagnosed with or suspected to be affected by MacTel Type 2; OR an immediate family member of a NHOR participant with MacTel; OR a healthy volunteer (control) or a volunteer (control) affected with a disorder thought to be related to MacTel Type 2.

Exclusion criteria

1. Inability to provide informed consent or undergo required procedures; and 2. Confounding (excluding diabetic retinopathy) ocular disorder that impacts the ability of the Reading Center to analyze images.

Study design

Design

Study phase:	N/A
Study type:	Observational non invasive
Intervention model:	Single
Allocation:	Non controlled trial
Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Aetiology

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-04-2025
Enrollment:	300
Duration:	1 months (per patient)
Type:	Anticipated

WORLD

Recruitment status:	Pending
Start date (anticipated):	01-01-2015
Enrollment:	2000
Type:	Anticipated

Medical products/devices used

Product type:	N.a.

IPD sharing statement

Plan to share IPD: Undecided

Plan description
N.a.

Ethics review

Approved WMO Date:	19-03-2025
Application type:	First submission
Review commission:	METC Oost-Nederland

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
Research portal	NL-009551