Examining brain phenotypes in carriers of (common) mutations in Polymerase gamma (PolG) using 7T MRI imaging.

Published: 15-03-2017
Last updated: 17-01-2025

I) To apply MRI techniques to identify anatomical and functional POLG related cerebral biomarkers. II) Investigate differences in said biomarkers between subjects and healthy controls.

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational non invasive

Summary

ID

NL-OMON55367

Source
ToetsingOnline

Brief title
7T fMRI in PolG carriers

Condition

- Chromosomal abnormalities, gene alterations and gene variants

Synonym
Brain structure, Mitochondrial disease

Research involving
Human

Sponsors and support

Primary sponsor: Universiteit Maastricht
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Imaging, PolG

Outcome measures

Primary outcome

Biomarkers in non-symptomatic POLG subjects, including anatomical and/or

functional differences.

Secondary outcome

NVT

Study description

Background summary

In this study, carriers of POLG mutation(s), displaying an asymptomatic phenotype, will be examined to determine possible anatomical and functional (MRI) deviations. Our goal is to examine both structural MRI differences between a mildly impaired and a healthy system, and the functional changes in a mildly impaired compared to healthy system.

Study objective

I) To apply MRI techniques to identify anatomical and functional POLG related cerebral biomarkers.
II) Investigate differences in said biomarkers between subjects and healthy controls.

Study design

Observational study

Study burden and risks

Including the informed consent procedure, the burden to participants is limited to approximately four contact hour on one day. All measurements are non-invasive. Participants with contraindications for MRI will be excluded. Therefore the risks associated with participating in this study are negligible.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)

Inclusion criteria

-Carrier of a POLG mutation
-asymptomatic phenotype

Exclusion criteria

-contra-indications for MRI
-severe phenotype

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	10-04-2018
Enrollment:	20
Type:	Actual

Medical products/devices used

Registration:	No

Ethics review

Approved WMO Date:	15-03-2017
Application type:	First submission
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)
Approved WMO Date:	19-03-2021
Application type:	Amendment
Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)

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	NL56881.068.16