National survey of people carrying the mt.3243A>G mutation of mitochondrial DNA.

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The purpose of this study concerns at a standardized way identifying the phenotype of patients with mt.3243A>G mutation, and relatives in the maternal line of patients with the mt.3243A>G mutation

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Metabolic and nutritional disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON34556

Source
ToetsingOnline

Brief title
mt.3243A> G inventory

Condition

- Metabolic and nutritional disorders congenital
- Neuromuscular disorders

Synonym
energiestofwisselingsziekte, niet-vasculaire beroertes

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Sint Radboud
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: inventory, MELAS, Mitochondrial disease, mt.3243A>G

Outcome measures

Primary outcome

The Newcastle Mitochondrial Disease Scales give a score which may be continued

in time and gives a measure of the seriousness of the mitochondrial disorder

Secondary outcome

Identifying a minimally invasive alternative to demonstrate the presence of the

mt.3243A> G mutation in patients and/or carriers.

Study description

Background summary

Mitochondrial dysfunction can be caused by a defect in the oxidative fosforyleringssysteem (OXPHOS). The mutation mt.3243A>G leads to a defect in the OXPHOS and is responsible for 80% of patients with MELAS syndrome (Mitochondrial myopathy, Encephalopatie, lactate acidosis and stroke-like episodes), however, the mt3243A>G mutation has a much larger clinical variability, which has never been reviewed.
For valid measurement of the degree of heteroplasmy in a patient, an invasive test is necessary. By comparison of different types of material, a minimally invasive alternative could to be identified.

Study objective

The purpose of this study concerns at a standardized way identifying the phenotype of patients with mt.3243A>G mutation, and relatives in the maternal line of patients with the mt.3243A>G mutation

Study design

With all patients participating, the Newcastle Mitochondrial Disease Adult Scale or the Newcastle Pediatric Mitochondrial Disease Scale is taken during a single outpatient visit to Nijmeegs Centre of Mitochondrial Disease. The Newcastle Mitochondrial Disease Scales are a validated way to determine the

severity of mitochondrial disease and follow the severity of disease in time.

Study burden and risks

Patients are asked to come to the outpatient clinic. Here, the Newcastle Mitochondrial Disease Adult Scale or the Newcastle Pediatric Mitochondrial Disease Scale is taken. This will take about 45 minutes to complete. In addition, a portion of patients urine is collected, saliva is taken by a buccal scrape and blood is taken to determine the degree of heteroplasmy in these materials.

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)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

patients carrying the mt.3243A>G mutation and relatives in the maternal line

Exclusion criteria

Absence of mt.3243A> G mutation

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-10-2010
Enrollment:	100
Type:	Actual

Ethics review

Approved WMO Date:	21-09-2010
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	16-03-2015
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

Approved WMO Date:	13-10-2015
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
CCMO	NL32683.091.10