European Spinocerebellar Ataxia Type 3/Machado*Joseph Disease Initiative

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To set up a trial*ready cohort by bringing together 7 European cohorts and 1 US cohortand include new subjects, which together comprise more than 800 subjects with the developmentand validation of innovative assessment instruments and disease...

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Movement disorders (incl parkinsonism)
Study type	Observational invasive

Summary

ID

NL-OMON55616

Source
ToetsingOnline

Brief title
ESMI

Condition

- Movement disorders (incl parkinsonism)

Synonym
cerebellar dysfunction, disorder of coordination

Research involving
Human

Sponsors and support

Primary sponsor: Neurologie
Source(s) of monetary or material Support: Joint Program Neurodegenerative Disease Research

Intervention

Keyword: Ataxia, SCA3, trial ready cohort

Outcome measures

Primary outcome

Main study parameters/endpoints:

A large cohort of preclinical and mildly ataxic SCA3 mutation carriers, which

includes a model of

disease evolution in SCA3 and new clinical, motor, imaging and biochemical

markers.

Secondary outcome

na

Study description

Background summary

SCA3 is the most common dominant form of cerebellar ataxia. As there is an advanced understanding
of the molecular mechanisms underlying SCA3, new therapeutic approaches are being developed,
and the SCA3 field is entering a phase of intense trial activity. To enable interventional trials,
availability of large cohorts that consist of preclinical mutation carriers and mildly affected patients is
mandatory.

Study objective

To set up a trial*ready cohort by bringing together 7 European cohorts and 1 US cohort
and include new subjects, which together comprise more than 800 subjects with the development
and validation of innovative assessment instruments and disease markers, including a new highly

sensitive motor test battery, ambulatory sensor*based activity measurement, automated MRI
volumetric evaluation, diffusion tensor imaging (DTI), and blood as well as CSF markers based on
transcript profiling and disease protein (ataxin*3) measurement.

Study design

Study design: An observational cohort study

Study burden and risks

Patients will visit the local partner once a year for three consecutive years. These visits includes clinical assessment which includes validated ataxia* scales and tests to assess cognitive function, mood, activities of daily living and quality of life. Blood samples will be acquired at each visit in a subset of patients. This results in a total of 3 blood samples for 15 patients during the study. In a subgroup of 60 SCA3 mutation carriers, and a subset thereof (estimated 15*20 subjects) in the Netherlands, detailed quantitative motor assessment will be performed. At about half of these visits new MRI scan will be acquired according to that needs about 35 minutes to be completed. Controls will only receive clinical assessment and blood sampling. Optionally, patients will be asked to undergo a lumbar puncture.

Contacts

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Scientific
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Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

Patients and carriers with SCA3
18 and older

Exclusion criteria

Other neurological disorders
Claustrofobia
Metal prosthesis or other metal objects in body
Implanted electronic devices such as a pacemaker

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)

Primary purpose: Other

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-05-2017

Enrollment:	35
Type:	Actual

Ethics review

Approved WMO Date:	23-03-2017
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	08-11-2017
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	20-03-2019
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	14-09-2020
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	24-08-2021
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
CCMO	NL58267.091.16