European Spinocerebellar Ataxia Type 3/Machado*Joseph Disease Initiative

Published: 23-03-2017 Last updated: 15-04-2024

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

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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

Public

Selecteer

Reinier Postlaan 4 Nijmegen 6525 GC NL

Scientific

Selecteer

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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