Towards a clinical trial for oculopharyngeal muscular dystrophy (OPMD) - natural history and outcome measures.

Published: 08-12-2015
Last updated: 19-04-2024

The primary objective is to identify clinical outcome measures in OPMD patients to support the future development of treatment strategies for OPMD. The secondary objectives are:• To identify molecular biomarkers for OPMD in biofluids to monitor…

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Neurological disorders congenital
Study type	Observational non invasive

Summary

ID

NL-OMON42807

Source
ToetsingOnline

Brief title
OPMD Forte

Condition

- Neurological disorders congenital

Synonym
oculopharyngeal muscular dystrophy, OPMD

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum

Source(s) of monetary or material Support: AFM

Intervention

Keyword: dysphagia, dystrophy, OPMD, outcomes

Outcome measures

Primary outcome

Any regression on the following functions at t=0 and t=18 months will be collected.

- Degree and extent of muscle weakness. Measured with MRC (Medical Research Council) scores, dynamometry, and assessment of the maximum bite force (using the Bite Force Gauge) and the maximum tongue strength (using Iowa Oral Performance Instrument (IOPI)).

- Fat fraction and degree of inflammation in muscles of legs and pharyngeal muscles. MRI-scans and muscle ultrasound will be performed.

- Swallow, chewing and speech capacity will be assessed and compared. Measurements of swallow capacity contains the maximum swallowing speed and maximum swallowing volume. The chewing capacity will be measured with the chewing test. Different parts of the standardized Dutch Dysartria Assessment (NDO-V) will be used to measure speech capacity.

- A structured interview about swallowing and speech function is included. And an interview on medical history and family history on OPMD.

- The functional motor abilities will be collected and measured with the Motor Function Measure (MFM) and the 10 steps stairs test.

- Scores on several questionnaires will be analyzed.

- Blood and saliva samples will be collected for DNA- and RNA analysis.

Secondary outcome

Not applicable

Study description

Background summary

Oculopharyngeal muscular dystrophy (OPMD) is a rare late onset, usually autosomal dominant, progressive muscle dystrophy. The dystrophy is characterized by ptosis, dysphagia, and limb weakness. At the moment natural history of OPMD is poorly understood and there are no reliable outcome measures for treatment effects. There is a need for tools to quantify symtpoms of OPMD for future clinical trials. To investigate and improve assessment of the muscle function and morphology of orofacial muscles and upper and lower extremity muscles further research is required. Also swallow capacity, chewing capacity and speech capacity in OPMD have to be further explored.

Study objective

The primary objective is to identify clinical outcome measures in OPMD patients to support the future development of treatment strategies for OPMD.
The secondary objectives are:
• To identify molecular biomarkers for OPMD in biofluids to monitor disease progression and better understand variety in disease severity.
• To describe the natural history of OPMD over 18 months.

Study design

observational study (longitudinal, cohort study)

Study burden and risks

Participants will be asked for two visits to the outpatient clinic at the department of neurology/rehabilitation. Their medical history will be taken, they will undergo a clinical and instrumental examination. Blood samples will be collected for DNA- and RNA analysis, and for storage at the Radboudumc biobank. It is possible to get a hematoma from blood venipuncture, this will recover by itself. Also, participants will undergo a MRI-scan and radiologic swallow assessment. Complications of MRI -scans or radiologic swallow

assessments are very uncommon. For the radiologic assessment, participants are a short time exposed to radiation, a restricted number of six swallows is determined. Participants are given the opportunity to object against any measurement. The risks of this study are negligible.

Contacts

Public
Radboud Universitair Medisch Centrum

Reinier Postlaan 2
Nijmegen 6525 GC
NL
Scientific
Radboud Universitair Medisch Centrum

Reinier Postlaan 2
Nijmegen 6525 GC
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

- Participants of 18 years and older who have a genetically confirmed diagnosis of OPMD with or without symptoms
- Participants of 18 years and older with a clinically diagnosed OPMD, who give permission for genetic testing for OPMD

Exclusion criteria

- Incapacitated persons will not be included in this study.
Participants can be excluded for parts of the study:
Participants with contra-indications for MRI-scan will not undergo MRI;
Participants who are claustrophobic will not undergo MRI, unless the participant approves;
Participants who are unable to swallow, hence are dependent of non-oral feeding or have no oral intake at all will not undergo swallowing tests.

Study design

Design

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

Primary purpose: Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	04-02-2016
Enrollment:	80
Type:	Actual

Ethics review

Approved WMO

Date:	08-12-2015
Application type:	First submission
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	NL54606.091.15