Trial readiness and fitness for congenital myopathies: A 2-year prospective natural history study including a cross-sectional study on muscle fatigability

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Ethical review	Approved WMO
Status	Recruiting
Health condition type	Neurological disorders congenital
Study type	Interventional

Summary

ID

NL-OMON56064

Source
ToetsingOnline

Brief title
READYCOM

Condition

- Neurological disorders congenital
- Neuromuscular disorders

Synonym
'Congenital myopathies' and 'inherited muscle disorders'

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum
Source(s) of monetary or material Support: Prinses Beatrix Spierfonds

Intervention

Keyword: Congenital myopathies, Muscle fatigability, Natural history, Trial readiness

Outcome measures

Primary outcome

-Motor function measure

-Endurance shuttle test

Secondary outcome

-Manual muscle testing

-Isometric dynamometry

-Range of motion

-Respiratory muscle function measured by spirometry

-Graded and timed rise from floor

-6 MWT

-7-day accelerometry

-Vignos and Brooke scale

-Laboratory tests

-Muscle ultrasound

-Muscle MRI

-DEXA scan

-Questionnaires

-Isokinetic dynamometry

-Repetitive nerve stimulation

Study description

Background summary

Congenital myopathies comprise a clinical, histopathological, and genetic heterogeneous group of early-onset muscle diseases. Several preclinical studies are moving towards clinical trials and for CNM the first trials are taking place. In parallel, many patients report severe muscle fatigability and positive effects of drugs improving the function of the NMJ. However, this has only been studied in small case series.[1,2]

Study objective

Part 1: Assess the natural disease course of three congenital myopathies during 24 months. This will enable us to obtain a detailed assessment of the phenotype and genotype, evaluate and optimize the current care, determine the rate of disease progression and select the most sensitive outcome measures.

Part 2: Assess the severity of muscle fatigability in four congenital myopathies.

Study design

Part 1: A 2-year prospective natural history study with internationally set clinical outcome measures and biomarkers in core myopathies, nemaline myopathies en centronuclear myopathies, with bi-annual follow-up (five visits; n=45). We will assess muscle strength and function, physical activity, experienced fatigue and sleep, pain, quality of life, daily functioning and social participation. Biomarkers will include laboratory tests, muscle ultrasound, muscle MRI, and bone densitometry.

Part 2: A cross-sectional study on muscle fatigability consisting of clinical and electrophysiological endurance tests in CCD/MmD, NEM, CNM (one visit; n=75). Muscle fatigability will be assessed during standardized functional endurance tasks and isolated muscle endurance tests using isokinetic dynamometry combined with surface EMG. Neuromuscular transmission (NMJ function) will be assessed with repetitive nerve stimulation. This will be combined with one a the visits from part 1. Everyone from part 1 that adheres to the inclusion criteria of part 2 will be able to participate in part 2.

Intervention

Non therapeutical intervention

Study burden and risks

The measurement days will be burdensome due to the long duration and it can cause sore muscles the next day. However, since we use all functional test using movements to which most patients are familiar (i.e. walking, transfers, etc.) the patient will be able to estimate his/her own risk. We don*t include tests in which we push patients to their physical limits.We will be gathering a lot of beneficial data in order to get a clear overview of this group of patients en the outcomes can be used in future research towards treatment options. Therefore we see it fit to conduct this research in minors.

Contacts

Public
Radboud Universitair Medisch Centrum

Geert Grooteplein Zuid 10
Nijmegen 6525 GA
NL
Scientific
Radboud Universitair Medisch Centrum

Geert Grooteplein Zuid 10
Nijmegen 6525 GA
NL

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

Genetically-confirmed congenital myopathy (core myopathy, nemaline myopathy, and centronuclear myopathy)

Exclusion criteria

Other neuromuscular, psychiatric or neurological disorders

Study design

Design

Study type: Interventional

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	28-03-2024
Enrollment:	100
Type:	Actual

Medical products/devices used

Registration:	No

Ethics review

Approved WMO

Date:	07-11-2023

Application type:	First submission
Review commission:	CCMO: Centrale Commissie Mensgebonden Onderzoek (Den Haag)
Approved WMO Date:	25-03-2024
Application type:	Amendment
Review commission:	CCMO: Centrale Commissie Mensgebonden Onderzoek (Den Haag)

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	NL83069.000.23