Selecting Outcome measures for MITOchondrial disease in children: an explorative study

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To study feasibility, reliability and validity of a specific set of outcome measures in children with mitochondrial myopathy.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Metabolism disorders NEC
Study type	Observational non invasive

Summary

ID

NL-OMON43301

Source
ToetsingOnline

Brief title
SO-MITO study

Condition

- Metabolism disorders NEC

Synonym
energy metabolism disorders, mitochondriopathy

Research involving
Human

Sponsors and support

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

Intervention

Keyword: mitochondrial disease, myopathy, outcome measures

Outcome measures

Primary outcome

Feasibility (primary endpoint; % of patients who were able to complete the test), reliability (inter-rater reliability, intra-rater reliability, test-retest reliability), and validity (correlation with predefined anchors and between parameters measuring the same construct) of all instruments tested in this study, per study group.

Secondary outcome

NA

Study description

Background summary

For clinical trials, it is of great importance to select clinically relevant, feasible, reliable and valid instruments to measure disease progression. To date, there is no experience or information on which instruments to use in children with mitochondrial disorders. Previously, we selected 33 clinically relevant and scientifically robust instruments based on an extensive literature search. Besides, several instruments for paediatric mitochondrial disease have been developed. In this study we aim to obtain more knowledge about the value of these instruments in measuring disease severity and progression in children with mitochondrial disease with the final aim to provide more information on these instruments to be used in future drug intervention clinical trial.

Study objective

To study feasibility, reliability and validity of a specific set of outcome measures in children with mitochondrial myopathy.

Study design

The study is a longitudinal observational study, in which patients are seen at time x, time x + 2 weeks (retest) and time x * 2 weeks (training). The patients will be seen by two experienced assessors, who will both keep their own CRF. The examinations will be videotaped and, where possible, be scored again after 4 weeks to determine inter-rater reliability. Since mitochondrial disorders, are rare, we use a multi-centre design (for recruitment only).

Study burden and risks

This protocol was supported by three different patients organizations, underlining the need for reliable outcome measures also from a patient perspective. Patients will be seen at the outpatient clinic of the Rehabilitation department three times. This means that they will not be able to attend day-care, kindergarden, or school during that time. They will be asked to perform several tests, which are regularly used to follow the functional capacities of handicapped children. Most children will be familiar with these kinds of test from their daily (rehabilitation) care. All tests measure daily functional capacities, none of the tests alone will be too tiring, though since these patients have a mitochondrial disorder and are easily tired, it is possible that the total day of testing will be tiring. To minimize this, we created a balanced programme with regular pauses, which in our previous experiences is not too burdensome to the patient. The programme was checked by a child with mitochondrial myopathy and estimated to be feasible. Also, it is known that some patients with mitochondrial disease may suffer from muscle pain after light exercise. The risks of a serious complication of the individual tests are however, both in literature and in our experience, negligible.
Since we aim to test paediatric measurement instruments, this study cannot be performed in adults. We think this study will be relevant not only for children with mitochondrial myopathies, but also for other paediatric and adult mitochondrial diseases, since using feasible, reliable and valid instruments increases the likelihood that relevant and valid conclusions will be drawn on the effect of future treatments.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Children (2-11 years)

Inclusion criteria

-17 years old at the inclusion date
* Mitochondrial myopathy
o - Signs of myopathy on examination (muscle weakness, hypotonia) or testing (exercise intolerance).
o - No signs or history of encephalopathy (IQ < 80*, epilepsy, classical migraine, encephalopathy, psychiatric disorders**, autism**, spasticity, ataxia, extrapyrimidal signs, brainstem failure or abnormalities on MRI)
* tested only when struggling in main school, tests from *5 years accepted
** diagnosed by a paediatric psychiatrist
* A confirmed pathogenic mutation

Exclusion criteria

* Insufficient knowledge of the Dutch language
* It is expected that the studies will be too burdensome for the patient or the family
* End-of-life expected within 3 months from the initiation of the study
* Other disabling disease
* Gluten allergy (for TOMASS cracker test only)

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	27-09-2017
Enrollment:	10
Type:	Actual

Ethics review

Approved WMO

Date:	14-11-2016
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	NL59491.091.16