Every child should be able to exercise! (Dutch: Ieder kind moet kunnen sporten!)

No registrations found.

Ethical review	Positive opinion
Status	Recruiting
Health condition type	-
Study type	Observational non invasive

Summary

ID

NL-OMON25784

Source
NTR

Brief title
Exercise and lcFAOD

Health condition

Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD), Carnitine Palmitoyl Transferase Type 2 Deficiency (CPT2D), Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHADD), General Mitochondrial Trifunctional Protein Deficiency (MTPD)

Sponsors and support

Primary sponsor: Metakids
Source(s) of monetary or material Support: Metakids

Intervention

Outcome measures

Primary outcome

(The course of) exercise tolerance and myopathic symptoms based on the results of maximal

and endurance exercise stress tests (muscle, pulmonary and cardiac function, and laboratory results (including CK, NT-proBNP, troponine, acylcarnitines) up until 24 hours after exercise)

Secondary outcome

- Reported muscle complaints, exercise tolerance, daily life activity levels, dietary regimen and metabolic exacerbations due to uncontrolled excessive exercise
- Quality of life (measured with the Pediatric Quality of Life Inventory (PEDSQL))

Study description

Background summary

Long-chain fatty acid oxidation deficiencies (lcFAOD) represent a group of inborn errors of metabolism characterized by hepatic, muscular and cardiac symptoms. Some patients may also develop neuropathy and/or retinopathy. Implementation of lcFAOD in the newborn screening (NBS) program in the Netherlands in 2007 allowed registration and follow-up of all newly identified patients. Up until now, most of the patients identified by NBS are asymptomatic.

Some of the main signs and symptoms in patients with lcFAOD diagnosed before the implementation of NBS (pre-NBS) are exercise-induced muscle pain and rhabdomyolysis. It is still unknown to what extent the currently asymptomatic lcFAOD patients identified by NBS will develop myopathic symptoms during adolescence, the age when in pre-NBS lcFAOD populations the incidence of myopathy peaks. The first lcFAOD patients identified by NBS in the Netherlands are now approaching this vulnerable age in which the risks and the social and physical benefits of exercise need to be careful balanced.

In this study, we aim to gain insight in the individual risk/benefit balance of exercise in patients with lcFAOD identified by NBS and improve this balance with safe exercise advice. To provide a personalized advice regarding safe exercise, exercise tests using cycling ergometry under safe laboratory conditions will be implemented in the standard follow-up of Dutch lcFAOD patients from 10 years of age. Muscle complaints, exercise tolerance, daily life activity levels and dietary regimen will be recorded with regular interviews and questionnaires. This standardized follow-up protocol enables prospective observation and evaluation of (the course of) exercise tolerance and myopathic symptoms, the individual risks and consequences of exercise, and the effects of safe exercise advice in lcFAOD patients identified by NBS.

Study objective

1) Personalized advice regarding safe exercise may improve physical condition, decrease uncontrolled excessive exercise and metabolic exacerbations, decrease insecurities regarding exercise, enhance participation in social life and as such provide a better quality of

life in general for the patients of lcFAOD patient.

2) Standardized exercise tests and follow-up will increase insight in (the course of) exercise tolerance and individual consequences of exercise of lcFAOD patients diagnosed by NBS. This will lead to identification of subgroups of patients with lcFAOD that require stratified exercise advice and/or treatment strategies and may allow evaluation of specific characteristics (genetic variations, lcFAO-fluxes, metabolites or their ratios) that predict reaction to exercise and will enable specific exercise recommendations for patients with lcFAOD, also beyond our study cohort.

Study design

Patients will perform the standardized exercise test every one or two years during follow-up, from the age of 10 years old. The intensity of the exercise test frequency depends on the severity of signs and symptoms in individual patients. More severe patients will be tested more frequently. After the first exercise test, patients will be interviewed every four weeks to monitor muscle complaints, exercise tolerance, daily life activities and dietary regimen. Questionnaires regarding quality of life will be filled out before the first exercise tests and yearly thereafter.

Contacts

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Eligibility criteria

Inclusion criteria

Children (>10 years old) with an lcFAOD, as confirmed by DNA analysis, enzyme activity measurement in lymphocytes, and in most cases also by lcFAO-flux analysis in fibroblasts.

Exclusion criteria

Severe cardiomyopathy endangering safe exercise.
For other symptomatic patients, exercise tests will be adapted to individual safe possibilities.

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non controlled trial
Masking:	Open (masking not used)
Control:	N/A , unknown

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-03-2021
Enrollment:	25
Type:	Anticipated

IPD sharing statement

Plan to share IPD: Undecided

Ethics review

Positive opinion

Date:	10-02-2021
Application type:	First submission

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
NTR-new	NL9257
Other	METC Utrecht : METC20-335/C

Study results