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

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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...

Ethische beoordeling	Positief advies
Status	Werving gestart
Type aandoening	-
Onderzoekstype	Observationeel onderzoek, zonder invasieve metingen

Samenvatting

ID

NL-OMON25784

Bron
NTR

Verkorte titel
Exercise and lcFAOD

Aandoening

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)

Ondersteuning

Primaire sponsor: Metakids
Overige ondersteuning: Metakids

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

(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)

Toelichting onderzoek

Achtergrond van het onderzoek

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.

Doel van het onderzoek

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.

Onderzoeksopzet

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.

Contactpersonen

Publiek

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Wetenschappelijk

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Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

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.

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

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

Onderzoeksopzet

Opzet

Type:	Observationeel onderzoek, zonder invasieve metingen
Onderzoeksmodel:	Anders
Toewijzing:	N.v.t. / één studie arm
Blindering:	Open / niet geblindeerd
Controle:	N.v.t. / onbekend

Deelname

Nederland

Status:	Werving gestart
(Verwachte) startdatum:	01-03-2021
Aantal proefpersonen:	25
Type:	Verwachte startdatum

Voornemen beschikbaar stellen Individuele Patiënten Data (IPD)

Wordt de data na het onderzoek gedeeld: Nog niet bepaald

Ethische beoordeling

Positief advies

Datum:	10-02-2021
Soort:	Eerste indiening

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

Geen registraties gevonden.

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

Register	ID
NTR-new	NL9257
Ander register	METC Utrecht : METC20-335/C

Resultaten