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Magnetic Resonance Spectroscopy (MRS)-analysis of muscle energy metabolism in medium-chain acyl-CoA dehydrogenase (MCAD) deficiency

Submission date
19/12/2013

Recruitment status
No longer recruiting

Registration date
05/03/2014

Overall study status
Completed

Last Edited
05/03/2014

Condition category
Nutritional, Metabolic, Endocrine

[ ] Prospectively registered
[ ] Protocol
[ ] Statistical analysis plan
[ ] Results
[ ] Individual participant data
[ ] Record updated in last year

Plain English summary of protocol

Background and study aims
Fatty acid oxidation is an important source of energy during prolonged fasting. Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency is the most common inborn error of fatty acid oxidation. Patients experience may vary, ranging from sudden infant death to remaining without any symptoms throughout life. The reason for this has not been fully understood. Treatment includes prevention of prolonged fasting and an emergency regimen during catabolic stress (breakdown of nutrients). Patients also have difficulty in exercising continuously. During moderate-intensity exercise, energy supply is met by fatty acid oxidation of the fat stored in muscle cells. In this study we aim to analyse the effect of MCAD deficiency on exercise tolerance.

Who can participate?
Adult patients with MCAD deficiency can participate in this study.

What does the study involve?
All patients are invited to take part in two sessions of exercise on a bicycle ergometer. Blood samples will be drawn from a thin tube that will be inserted into the vein. Participants will be asked to refrain from food intake three hours before both study sessions. A food diary will be kept for 7 days prior to session 2.

What are the possible benefits and risks of participating?
No side effects of this study are expected, as the measurements are non-invasive. You may be hesitant towards the small space in the MRI scanner, but there will be sufficient time to get acquainted with the small space. Blood samples will be taken once and are without risk.

Where is the study run from?
Session 1 takes place at the paediatric test ward at the University Medical Centre Groningen (Netherlands). Session 2 takes place at the Neuroimaging Centre (NIC), Groningen (Netherlands).

When is study starting and how long is it expected to run for?
The study started in October 2012 and ended in December 2013.

Who is funding the study?
The study is funded by RVVZ Foundation, Netherlands.

Who is the main contact?
Prof. G.P.A. Smit
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Contact information

Type(s)
Scientific

Contact name
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Contact details
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9700 RB

Additional identifiers

EudraCT/CTIS number

IRAS number

ClinicalTrials.gov number

Secondary identifying numbers
N/A

Study information

Scientific Title
In vivo analysis by 1H- and 31P-MRS of the effect of moderate-intensity exercise on intramyocellular lipid content and mitochondrial function in patients with MCAD deficiency

Study objectives
Patients with MCAD deficiency are unable to oxidize sufficient intramyocellular fatty acids during exercise to meet the energy demands, resulting in intramuscular energy deficiency and muscle complaints.

Ethics approval required
Old ethics approval format

Ethics approval(s)

Medical Ethics Committee of the University Medical Centre Groningen, 01/10/2012, ref: METc 2012/262

Study design
Interventional case-control study

Primary study design
Interventional

Secondary study design
Case-control study

Study setting(s)
Hospital

Study type(s)
Diagnostic

Participant information sheet
Not available in web format, please use the contact details below to request a patient information sheet

Health condition(s) or problem(s) studied
MCAD deficiency

Interventions
All participants undergo the two described study sessions as follows.
Incremental test on bicycle ergometer for determination of VO2max (session 1). Exercise for 80 minutes on bicycle ergometer and MRS analyses before, during and after exercise (session 2). Exercise during MRS will be performed on an MR-compatible bicycle. Venipuncture during session 2 (four small blood samples will be drawn from a cannula that will be inserted upon venipuncture for analysis of metabolites that play a role in muscle metabolism). For matters of standardization, participants will be asked to abstain from food intake three hours before both study sessions. A food diary will be kept for 7 days prior to session 2.

Intervention Type
Other

Phase
Not Applicable

Primary outcome measure
Intramyocellular lipid content is measured by means of 1H-MRS. We obtain spectra at baseline and after prolonged exercise, and have professional help for analysis of the spectra.

Secondary outcome measures
Concentrations of phosphocreatine, adenosine 5-triphosphate, inorganic phosphate, and intracellular pH.
These are measured by 31P-MRS. We obtain spectra before, during, and after exercise, and have professional help for analysis of the spectra.

Overall study start date
01/10/2012

Completion date
15/12/2013

Eligibility

Key inclusion criteria
Patient group: Patients with MCAD deficiency who are over 18 years of age
Control group: Healthy volunteers over 18 years of age, matched for age and gender to the patient group

Participant type(s)
Healthy volunteer

Age group
Adult

Lower age limit
18 Years

Sex
Both

Target number of participants
16

Key exclusion criteria
1. Disease other than MCAD deficiency
2. Contra-indications for MR analysis (e.g. the presence of metals in the body)

Date of first enrolment
01/10/2012

Date of final enrolment
15/12/2013

Locations

Countries of recruitment
Netherlands

Study participating centre
Hanzeplein 1
Groningen
Netherlands
9700 RB

Sponsor information

Organisation
University Medical Centre Groningen (Netherlands)

Sponsor details
Hanzeplein 1
Groningen
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Sponsor type
Hospital/treatment centre

ROR
https://ror.org/03cv38k47

Funder(s)

Funder type
Research organisation

Funder Name
RVVZ Foundation (Reserves Voormalige VrijwilligeZiekenfonds-verzekeringen) (Netherlands)

Results and Publications

Publication and dissemination plan
Not provided at time of registration

Intention to publish date

Individual participant data (IPD) sharing plan

IPD sharing plan summary
Not provided at time of registration