

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

<b>Submission date</b> 19/12/2013	<b>Recruitment status</b> No longer recruiting	<input type="checkbox"/> Prospectively registered
<b>Registration date</b> 05/03/2014	<b>Overall study status</b> Completed	<input type="checkbox"/> Protocol
<b>Last Edited</b> 05/03/2014	<b>Condition category</b> Nutritional, Metabolic, Endocrine	<input type="checkbox"/> Statistical analysis plan
		<input type="checkbox"/> Results
		<input type="checkbox"/> Individual participant data
		<input type="checkbox"/> 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  
g.p.a.smit@umcg.nl

## Contact information

**Type(s)**  
Scientific

**Contact name**  
Prof G. Peter A. Smit

**Contact details**  
Hanzeplein 1  
Groningen  
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9700 RB

## Additional identifiers

## Study information

**Scientific Title**  
In vivo analysis by <sup>1</sup>H- and <sup>31</sup>P-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

**Study type(s)**

Diagnostic

**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 VO<sub>2</sub>max (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(s)**

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.

**Key secondary outcome(s)**

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.

**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

**Healthy volunteers allowed**

No

**Age group**

Adult

**Lower age limit**

18 years

**Sex**

All

**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)

**ROR**

<https://ror.org/03cv38k47>

**Funder(s)****Funder type**

Research organisation

**Funder Name**

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

## **Results and Publications**

**Individual participant data (IPD) sharing plan**

**IPD sharing plan summary**

Not provided at time of registration