

# Optimizing therapeutic strategies in VLCADD

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<b>Registration date</b> 22/06/2017	<b>Overall study status</b> Completed	<input type="checkbox"/> Protocol
<b>Last Edited</b> 25/09/2018	<b>Condition category</b> Nutritional, Metabolic, Endocrine	<input type="checkbox"/> Statistical analysis plan
		<input checked="" type="checkbox"/> Results
		<input type="checkbox"/> Individual participant data

## Plain English summary of protocol

### Background and study aims

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD) is an inherited disorder of that causes the body to be unable to break down certain fats. Patients are unable to use fat as an energy source and can develop symptoms during fasting, fever and extensive exercise. To prevent symptoms many countries screen for VLCADD in their newborn screening (NBS) programs over the last years. This resulted in the detection of patients at an early age, often before the development of symptoms. The only treatment is a strict low-fat diet with regular feedings, but this is not necessary for every patients. Some will never develop symptoms, even without a strict diet. A recent study showed that measurement of fat oxidation in skin cells is currently the best marker to predict if a patient will develop symptoms. The aim of this study is to use this test as a tool to choose the best diet for VLCADD patients.

### Who can participate?

Anyone aged 10 to 99 years old who have a confirmed diagnosis of VLCADD

### What does the study involve?

Participants attend regular visits (about once every one to two years) where they are examined by a multidisciplinary team consisting of a metabolic specialist, a research dietician, a neurologist, a physical therapists and a cardiologist. Before the study visits, participants are asked to fill in a three day food diary. During this visit an extensive medical questionnaire, neurological examination, and undergo heart tests. If necessary, patient records from the local metabolic center of the participant are collected (only after permission by the participant).

### What are the possible benefits and risks of participating?

There are no benefits or risks with participating.

### Where is the study run from?

Dutch Fatty Acid Oxidation Expertise Center, University Medical Center Utrecht (Netherlands)

### When is the study starting and how long is it expected to run for?

October 2010 to June 2017

### Who is funding the study?

Metakids (Netherlands)

Who is the main contact?

Dr Gepke Visser  
gvisser4@umcutrecht.nl

## Contact information

### Type(s)

Scientific

### Contact name

Dr Gepke Visser

### Contact details

Department of Metabolic Diseases  
Dutch Fatty Acid Oxidation expertise center  
Wilhelmina Children's Hospital  
KE 04.306.0,  
PO Box 85090  
Utrecht  
Netherlands  
3508 AB  
+31 887555555  
gvisser4@umcutrecht.nl

## Additional identifiers

### Protocol serial number

METC 10-430

## Study information

### Scientific Title

Observational study on the dietary habits and clinical outcome of Dutch VLCADD patients

### Study objectives

LC-FAO flux (beta-oxidation (LC-FAO) flux score (rate of oleate oxidation)) can be used as a tool to select an individualized dietary strategy.

### Ethics approval required

Old ethics approval format

### Ethics approval(s)

Medical Ethics Committee of the University Medical Centre Utrecht, 07/12/2010, ref: METC 10-430/C

### Study design

Retrospective observational cohort study

### Primary study design

Observational

## **Study type(s)**

Diagnostic

## **Health condition(s) or problem(s) studied**

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD) is an autosomal recessive inherited disorder of mitochondrial long-chain fatty acid beta-oxidation (OMIM 201475)

## **Interventions**

For this observational study, all registered Dutch Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD) patients diagnosed before 2007 (when VLCADD was included in the Dutch newborn screening programmes (NBS) panel) are asked to visit the Dutch Fatty Acid Oxidation Expertise Center in the University Medical Center in Utrecht on a regular basis (around once every one to two years). Before the visit, participants are asked to fill in a three day food diary.

During visits, participants are interviewed and examined by a team of medical experts (metabolic specialists, a research dietician, a neurologist, a physical therapist and a cardiologist). Routine cardiac ultrasound and EKG is also performed during the visits. Information on previous symptoms and/ or lab results was collected from patient records from their local metabolic center (if this was mentioned during the interview). Participant's long-term dietary histories and their beta-oxidation (LC-FAO) flux score (rate of oleate oxidation) in cultured skin fibroblasts are evaluated in relation to clinical outcomes. This was in addition to their regular follow-up by their local metabolic specialist. Available data on diagnostic markers (including LC-FAO flux measured in fibroblasts) are collected and evaluated in relation to clinical outcome.

## **Intervention Type**

Behavioural

## **Primary outcome(s)**

Clinical outcomes are measured using the Clinical Severity Score (CSS) during visits to the Dutch FAO (around everyone to two years).

## **Key secondary outcome(s)**

Ability to work/ attend a regular school as reported by the patient or parents is measured using a standardized questionnaire at each visit to the Dutch FAO expertise center at the University Medical Center Utrecht.

## **Completion date**

01/06/2017

# **Eligibility**

## **Key inclusion criteria**

1. Confirmed diagnosis based on deficient VLCADD enzymatic activity in lymphocytes and/or cultured fibroblasts
2. Presence of biallelic mutations in the ACADVL gene (OMIM 609575). The diagnosis should not be based on newborn screening.
3. Aged between 10 to 99 years old

**Participant type(s)**

Patient

**Healthy volunteers allowed**

No

**Age group**

All

**Sex**

All

**Key exclusion criteria**

Diagnosis based on results of Newborn Screening

**Date of first enrolment**

01/01/2011

**Date of final enrolment**

01/01/2017

**Locations****Countries of recruitment**

Netherlands

**Study participating centre**

**Dutch Fatty Acid Oxidation Expertise Center**

Wilhelmina Children's Hospital

University Medical Center Utrecht

Lundlaan 6

Utrecht

Netherlands

3584 EA

**Sponsor information****Organisation**

University Medical Center Utrecht

**ROR**

<https://ror.org/0575yy874>

# Funder(s)

## Funder type

Charity

## Funder Name

Metakids

# Results and Publications

## Individual participant data (IPD) sharing plan

The datasets generated during and/or analysed during the current study are/will be available upon request from Dr. Gepke Visser (gvisser4@umcutrecht.nl). Requests for participant data can also be made at the official fatty acid oxidation disorders registry [www.fattyacidoxidation.org](http://www.fattyacidoxidation.org)

## IPD sharing plan summary

Available on request

## Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?
<a href="#">Results article</a>	results	01/01/2019		Yes	No
<a href="#">Participant information sheet</a>	Participant information sheet	11/11/2025	11/11/2025	No	Yes
<a href="#">Study website</a>	Study website	11/11/2025	11/11/2025	No	Yes