

# What is the effect of newborn screening on the clinical outcome of VLCADD patients

No registrations found.

|                              |                            |
|------------------------------|----------------------------|
| <b>Ethical review</b>        | Positive opinion           |
| <b>Status</b>                | Recruiting                 |
| <b>Health condition type</b> | -                          |
| <b>Study type</b>            | Observational non invasive |

## Summary

### ID

NL-OMON25272

### Source

Nationaal Trial Register

### Health condition

Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD) (OMIM 201475)  
In Nederland wordt dit aangeduid als VLCADD of VLCAD deficiëntie

## Sponsors and support

**Primary sponsor:** University Medical Center Utrecht

**Source(s) of monetary or material Support:** ZonMw (financial)  
Metakids (financial)

## Intervention

## Outcome measures

### Primary outcome

Hypoglycemia

Cardiomyopathy and/ or arrhythmia

Myopathy

## **Secondary outcome**

Ability to attend school/ keep a job

Admissions

CK values

## **Study description**

### **Background summary**

Very long chain acyl-CoA dehydrogenase deficiency (VLCADD) is an autosomal recessive inherited disorder of mitochondrial long-chain fatty acid beta-oxidation. In The Netherlands, VLCADD was included in the newborn screening (NBS) panel since 2007. The aim of this study is to evaluate the effect of NBS on clinical outcome in VLCADD. Therefore, comparison will take place of genetic, biochemical, and clinical characteristics of all VLCADD patients diagnosed before NBS and by NBS that are registered in the Dutch Database Registry of Metabolic Diseases (DDRMD). Simultaneously prospective clinical evaluation of these patients will be performed by a standardized protocol in the Dutch Expertise Center for Fatty Acid Oxidation Disorders.

### **Study objective**

With the introduction of very long chain acyl-CoA dehydrogenase deficiency (VLCADD) in newborn screening (NBS) programs, the number of diagnosed patients has rapidly increased. Most infants are asymptomatic at time of diagnosis and remain so. The lingering question is whether this is an effect of prompt diagnosis or of diagnosing asymptomatic individuals.

### **Study design**

1-2 years

### **Intervention**

Diagnosis by newborn screening yes/no

## **Contacts**

**Public**

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## Eligibility criteria

### Inclusion criteria

All patient have a confirmed diagnosis based on deficient VLCAD enzymatic activity in lymphocytes and/or cultured fibroblasts and the presence of biallelic mutations in the ACADVL gene (OMIM 609575).

### Exclusion criteria

No confirmed diagnosis of VLCADD (see inclusion criteria)

## Study design

### Design

|                     |                                 |
|---------------------|---------------------------------|
| Study type:         | Observational non invasive      |
| Intervention model: | Parallel                        |
| Allocation:         | Non-randomized controlled trial |

|          |                         |
|----------|-------------------------|
| Masking: | Open (masking not used) |
| Control: | N/A , unknown           |

## Recruitment

|                           |             |
|---------------------------|-------------|
| NL                        |             |
| Recruitment status:       | Recruiting  |
| Start date (anticipated): | 01-01-2011  |
| Enrollment:               | 50          |
| Type:                     | Anticipated |

## Ethics review

|                   |                  |
|-------------------|------------------|
| Positive opinion  |                  |
| Date:             | 20-06-2017       |
| 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  | NL6407                              |
| NTR-old  | NTR6582                             |
| Other    | METC UMC Utrecht : 10/430 en 15/582 |

## Study results