

# Study of the natural history of patients with rare inherited metabolic diseases, optimal treatment and complications

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(also see: C1. Protocol, page 9, paragraph 2)Primary: Creating a metabolic bio-database with body material of patients with inherited metabolic diseases, combined with clinical data.Secondary: Achieve better participation in national and...

|                              |  |
|------------------------------|--|
| <b>Ethical review</b>        | Not approved                                   |
| <b>Status</b>                | Will not start                                 |
| <b>Health condition type</b> | Metabolic and nutritional disorders congenital |
| <b>Study type</b>            | Observational invasive                         |

## Summary

### ID

NL-OMON53541

### Source

ToetsingOnline

### Brief title

Life with an inherited metabolic disease

### Condition

- Metabolic and nutritional disorders congenital
- Hepatic and hepatobiliary disorders
- Inborn errors of metabolism

### Synonym

(layman's term not used by patient groups, metabolic disease, not applicable)

### Research involving

Human

### Sponsors and support

**Primary sponsor:** Erasmus MC, Universitair Medisch Centrum Rotterdam

**Source(s) of monetary or material Support:** Niet van toepassing; het is 'investigator driven'.

## Intervention

**Keyword:** disabilities, metabolic disease, natural course, quality of life

## Outcome measures

### Primary outcome

(also see: C1. Protocol, page 11, paragraph 5.1.1)

A metabolic bio-database, with DNA, blood samples, urine samples, faeces samples and hair samples from patients with inherited metabolic diseases, combined with the clinical and physical data.

### Secondary outcome

(also see: C1. Protocol, page 11 and 12, paragraph 5.1.2)

Clinical data: medication, history and symptoms, radiology data, other medical information such as endoscopies and surgery reports, and ECG.

Disease specific or general questionnaires.

Physical data: physical disabilities, neurological disorders, anthropometry, blood pressure, eye-hand coordination, activity level and pain level, and fundus.

## Study description

### Background summary

Little is known about the natural history of most rare inherited metabolic diseases. The mortality in childhood is still significantly high. For many diagnoses limited research has been done on the treatment of rare hereditary metabolic diseases in adulthood. It is important to create a biobank with linked patient characteristics. It is easier and faster than to let patients

participate in (inter) national research, and to conduct prospective research into the natural course of a disease.

Additional measurement of quality of life, eye-hand coordination, activity and pain level in patients with inherited metabolic diseases and in the healthy population by contrast will increase our knowledge of inherited metabolic diseases.

## **Study objective**

(also see: C1. Protocol, page 9, paragraph 2)

Primary:

Creating a metabolic bio-database with body material of patients with inherited metabolic diseases, combined with clinical data.

Secondary:

Achieve better participation in national and international studies.

Study quality of life, eye-hand coordination, pain and activity in cases and controls.

## **Study design**

(also see: C1. Protocol, page 10, paragraph 3)

This prospective case control study will create a metabolic bio-database and will compare the quality of life, eye-hand coordination, activity and pain level in patients of rare inherited metabolic diseases to the general healthy population.

## **Study burden and risks**

(also see: C1. Protocol, page 16, paragraph 8.3)

Participants will be asked to have a lock of hair cut off, fill in questionnaires, wear an activity meter, enter pain scores into the watch, and keep a diary. A hematoma and a small risk of infection may occur after venapunction.

There is no direct benefit for cases and controls. We aim to increase our knowledge about inherited metabolic diseases.

## **Contacts**

### **Public**

Erasmus MC, Universitair Medisch Centrum Rotterdam

Westzeedijk 353

Rotterdam 3015 AA

NL

## Scientific

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## Trial sites

### Listed location countries

Netherlands

## Eligibility criteria

### Age

Adolescents (16-17 years)  
Adults (18-64 years)  
Elderly (65 years and older)

### Inclusion criteria

(see Protocol C1. page 10-11, paragraph 4.2 Inclusion criteria)

- Age  $\geq 16$  years.
- Written informed consent.
- Cases: capacitated and incapacitated patients with a rare inherited metabolic disease, who are in care at UMCs and satellite expert centers.
- Controls: healthy siblings and/or unrelated neighbors/ friends/ partners of the patients selected via the cases.

List of diagnosis groups based on current patient population in the UMCs:

- Urea cycle defects
- Disorders of amino acid metabolism
- Phenylketonuria
- Homocystinuria
- Glycogen overloading diseases
- Mitochondropathy
- Lysosomal overloading diseases
- Fatty acid oxidation disorders
- Disorders of glycosylation
- Galactosemia

- Peroxisomal disorders
- Congenital hyperinsulinism
- Porphyrrias
- Lipoprotein deficiencies
- Disorders of vitamin metabolism
- Disorders of glycosylation (CDG)
- Rest of rare inherited metabolic diseases

## Exclusion criteria

(see Protocol C1. page 11)

No signed 'informed consent' present.

## Study design

### Design

|                     |                                 |
|---------------------|---------------------------------|
| Study type:         | Observational invasive          |
| Intervention model: | Other                           |
| Allocation:         | Non-randomized controlled trial |
| Masking:            | Open (masking not used)         |
| Control:            | Active                          |
| Primary purpose:    | Basic science                   |

### Recruitment

|                     |                |
|---------------------|----------------|
| NL                  |                |
| Recruitment status: | Will not start |
| Enrollment:         | 2000           |
| Type:               | Anticipated    |

## Ethics review

|                   |                  |
|-------------------|------------------|
| Not approved      |                  |
| Date:             | 21-09-2023       |
| Application type: | First submission |

Review commission:

METC Erasmus MC, Universitair Medisch Centrum Rotterdam  
(Rotterdam)

## 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             |
|----------|----------------|
| CCMO     | NL83254.078.22 |