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

Published: 21-09-2023
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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...

Ethical review	Not approved
Status	Will not start
Health condition type	Metabolic and nutritional disorders congenital
Study type	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

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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 >= 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
- Porphyrias
- 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