Metabolic evaluation of nutrition in Rett syndrome: creatine metabolism.

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Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON34479

Source
ToetsingOnline

Brief title
Metabolic evaluation of nutrition in Rett syndrome

Condition

- Chromosomal abnormalities, gene alterations and gene variants

Synonym
Rett syndrome

Research involving
Human

Sponsors and support

Primary sponsor: Medisch Universitair Ziekenhuis Maastricht
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Creatine, Metabolic, Nutrition, Rett syndrome

Outcome measures

Primary outcome

Blood as well as urine samples will be collected to confirm previous findings (plasma and urine creatine concentrations) and perform mutation analysis regarding the SLC6A8 gene. Secondary, a skin biopsy will be collected for functionality investigations regarding creatine transporter.

Secondary outcome

Not applicable.

Study description

Background summary

Rett syndrome (RTT) is an X-linked severe neurodevelopmental disorder. Despite their good appetite, many females with RTT meet the criteria for moderate to severe malnutrition. The pathological mechanism is barely understood. Although feeding difficulties may play a part in this, other constitutional factors as altered metabolic processes are suspected. Preliminary research showed elevated plasma creatine concentrations and increased urinary creatine/creatinine ratios in half of the RTT girls. Further investigations will be done in study. Blood and urine samples as well as a skin biopsy will be collected.

Study objective

The aim of this study is to confirm previous findings and examine the functionality of the creatine transporter in RTT girls, at which mutation analysis of the SLC6A8 gene (as a possible cause of an altered functionality of the creatine transporter) will be performed.

Primary objectives of the study are:
1. Can previous findings be confirmed?
2. Is the functionality of the creatine transporter altered in RTT girls?

3. Are mutations in the SLC6A8 gene present in RTT girls?

Study design

Observational study.

Study burden and risks

Blood and urine samples will be collected once, in addition to a regular blood withdrawal. These analysis will require 6 ml extra blood. Secondary, a skin biopsy will be collected under local anesthesia, using an Emla-plaster. The risks are negligible and the burden of participation to the study is minimal.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)

Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

Clinical diagnosis of Rett syndroom according to the diagnostic criteria (Hagberg et al, 2002)
MECP2-mutation
Complete neurophysiological work-up
Participant preliminary research (NL25356.068.08)

Exclusion criteria

Male gender

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	06-09-2010
Enrollment:	13
Type:	Actual

Ethics review

Approved WMO

Date:	12-08-2010
Application type:	First submission

Review commission:	METC academisch ziekenhuis Maastricht/Universiteit Maastricht, METC azM/UM (Maastricht)

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	NL32481.068.10