A systematic metabolic approach to the evaluation of nutrition in Rett syndrome according to the cardiorespiratory phenotype in Dutch Rett girls.

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The aims of this pilot study are to describe the nutritional status of a well-defined group of Dutch RTT girls with complete clinical, molecular and neurophysiological work-up (full description is presented in the section *study population*) and to...

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

Summary

ID

NL-OMON34012

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: Academisch Ziekenhuis Maastricht

Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Metabolic, Nutrition, Rett syndrome

Outcome measures

Primary outcome

To ascertain the nutritional status, a complete nutrition assessment and measurement of body composition will be carried out. To examine the consequences of an altered carbon dioxide metabolism, blood as well as urine samples will be collected concerning biochemical screening for metabolites from multiple pathways. Secondary, metabolites indicating possibly impaired mitochondrial function will also be investigated (full description is presented on page 21 and 22).

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. Irregular breathing is a common clinical feature, reflecting the immaturity of the brainstem in RTT. The primary pathophysiology is a defective control mechanism of carbon dioxide exhalation that leads to chronic respiratory alkalosis or acidosis. We assume that chronic respiratory acidosis or alkalosis causes derangement of the metabolic equilibrium in RTT females with important nutritional consequences.

Study objective

The aims of this pilot study are to describe the nutritional status of a well-defined group of Dutch RTT girls with complete clinical, molecular and neurophysiological work-up (full description is presented in the section *study population*) and to examine the consequences of a chronic respiratory acidosis or alkalosis on metabolic processes.

Primary objectives of the study are:
1. What is the nutritional status of the RTT girls?
2. Can metabolic alterations caused by chronic respiratory acidosis or alkalosis be detected?

Understanding the nutritional and cardiorespiratory requirements of these patients is important in order for them to receive appropriate and effective treatment. This treatment is of interest for general health and the quality of daily life of the whole family.

Study design

Observational pilot study.

Study burden and risks

Blood and urine samples will be collected once, in addition to a regular blood withdrawal. These analysis will require 13 ml extra blood. Blood withdrawal will be done by dr. E.E.J. Smeets. A person who is familiar with the person with RTT will attend the blood withdrawal. This study is carried out in girls with RTT, who are incapacitated persons. The study is group related; it is only possible to extent the knowledge of RTT using this group of persons. The risks include taking a blood and urine sample once, which are negligible and the burden of participation to the study is minimal. Regarding the parents time investment concerning filling in a three-day nutritional diary and the interview will be about 45 minutes.

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

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):	20-05-2009
Enrollment:	12
Type:	Actual

Ethics review

Approved WMO Date:	11-03-2009
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
Other	00786071
CCMO	NL25356.068.08