Creatine transporter defect in females.

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1. Define the clinical spectrum in female carriers of creatine transporter defect. 2. Correlate the clinical phenotype with X-inactivation studies.

Ethical review	Approved WMO
Status	Pending
Health condition type	Neurological disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON31374

Source
ToetsingOnline

Brief title
CRTR defect in females.

Condition

- Neurological disorders congenital
- Inborn errors of metabolism
- Mental impairment disorders

Synonym
creatine transporter defect

Research involving
Human

Sponsors and support

Primary sponsor: Vrije Universiteit Medisch Centrum
Source(s) of monetary or material Support: Vidi creatine transporter deficiency;Gajja Salomons.

Intervention

Keyword: creatine transporter defect, female carriers, phenotype, X-inactivation

Outcome measures

Primary outcome

The investigations will consist of 1.medical and family history, 2.physical examination, 3. laboratory analysis which will included biochemical analysis of urine and blood, X-inactivation studies in blood, hairs and saliva, RNA analysis of the creatine transporter gene and creatine uptake studies in fibroblasts and EBV-transformed lymphocytes, 4.1H-MRS of brain and 5. a neuropsychologic evaluation.

Secondary outcome

not appicable

Study description

Background summary

The creatine transporter defect is a X-linked cause of mental retardation that has recently been discovered at the metabolic unit of the department of clinical chemistry of the VUMC in collaboration with the Cincinnati Children*s Hospital Medical Center in the USA. Few clinical details have been reported about the phenotype in female carriers that, due to variable X-inactivation, can be expected to vary between asymptomatic and severe presentation similar to affected male patients.

Study objective

1. Define the clinical spectrum in female carriers of creatine transporter defect. 2. Correlate the clinical phenotype with X-inactivation studies.

Study design

cross-sectional observational study.

Study burden and risks

The burden of the study consists of two invasive procedures (one peripheral blood sampling and one skin biopsy), the pulling out of 20 hairs, the fact that it will take up time (two visits of about 3-4 hours) and the possible psychological burden of being confronted with the carrier status. The results of the test might be of importance to the subjects as it may give a prediction of the possible effectiveness of treatment with creatine and its biosynthesis percursors.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

proven carrier of pathogenic mutation in creatine transporter gene.

Exclusion criteria

no informed consent.

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	15-06-2007
Enrollment:	15
Type:	Anticipated

Ethics review

Approved WMO

Application type:	First submission
Review commission:	METC Amsterdam UMC

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	NL18019.029.07