Dissecting Down Syndrome * Study on the Variability in Phenotype

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We aim to obtain more insight in the pathogenesis of DS and the potential role of methylation in this by a small initial study (proof of principle). If the study shows a potential role of disturbed methylation in DS pathogenesis we will subsequently...

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

Summary

ID

NL-OMON38454

Source
ToetsingOnline

Brief title
Dissecting Down Syndrome

Condition

- Chromosomal abnormalities, gene alterations and gene variants

Synonym
Down Syndrome, Trisomy 21

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: - Down syndrome, - Methylation, - Pathogenesis, - Phenotypic variability

Outcome measures

Primary outcome

Significant difference in methylation patterns between controls and DS

individuals.

Secondary outcome

None.

Study description

Background summary

Down syndrome (DS) is the most frequent genetic cause of intellectual disability. DS is explained by the presence of an additional chromosome 21 which serves as the basis of DS-pathogenesis. However, underlying cellular and molecular processes causing the DS-phenotype remain not well understood. Also, among DS individuals a wide variability of phenotypic features can be observed which can not be explained yet.

Several hypotheses exist regarding the implications and effects of the presence of an extra copy of chromosome 21. A disturbed methylation by the presence of an additional copy of chromosome 21 has not been explored to date. The location of chromosomes within the nucleus has become known to be important in the methylation process. Altered methylation may influence transcription of genes located on chromosome 21 and also genes elsewhere in the genome.

Study objective

We aim to obtain more insight in the pathogenesis of DS and the potential role of methylation in this by a small initial study (proof of principle). If the study shows a potential role of disturbed methylation in DS pathogenesis we will subsequently initiate a similar study in a large cohort.

Study design

Observational study (proof of principle) with invasive measurements.

Study burden and risks

The samples will be obtained when blood sampling is needed because of clinical care. For the control indivduals the samples will be obtained from the general lab anonymously ('restmateriaal').

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Children (2-11 years)

Inclusion criteria

- Individual with Down syndrome caused by an extra, not translocated copy of chromosome 21
- Age 0-1 yr

Exclusion criteria

Individual with Down syndrome with a major malformation

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:	Recruitment stopped
Start date (anticipated):	03-03-2014
Enrollment:	40
Type:	Actual

Ethics review

Approved WMO

Date:	17-09-2013
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	NL45496.018.13