Dissecting Down Syndrome

Published: 03-12-2015
Last updated: 19-04-2024

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Ethical review	Approved WMO
Status	Will not start
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON42548

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, Phenotype variability

Outcome measures

Primary outcome

Significant differences in DNA-methylation- and genetranscription profiles

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 extra chromosome 21 which serves as the fundament 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 hypothesis exist regarding the implications and effects of the presence of an extra copy of chromosome 21. A genome-wide disturbance in methylation by the presence of an extra copy of chromosome 21 was demonstrated by us in a previous study (2013-2014).

Study objective

We aim to obtain more insight in the pathogenesis of DS. This replication study will focus on molecular determinants suchs as methylation and gene transcription. In addition, we will collect phenotypic information and link this with information retreived by the molecular studies.

Study design

Observational study with invasive measurements and access to medical file.

Study burden and risks

The samples will be obtained when blood sampling is needed because of clinical care. Regarding the control individuals 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

Patients:
- Individuals with Down syndrome caused by an extra, not translocated copy of chromosome 21
- Age 0 - 1 yr
Controls
- Individuals without Down syndrome and a non-hereditary disorder fo which blood sampling is needed
- Age 01- 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:	Will not start
Enrollment:	40
Type:	Anticipated

Ethics review

Approved WMO

Date:	03-12-2015
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	NL55315.018.15