Creation of a disease model system to investigate the mechanisms causing intellectual disability.

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1) to gain insight in the pathophysiological mechanisms leading to intellectual disability and
2) to create a model system which allows screening of therpeutic interventions for efficacy.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Mental impairment disorders
Study type	Observational non invasive

Summary

ID

NL-OMON38756

Source
ToetsingOnline

Brief title
Disease model system for intellectual disability

Condition

- Mental impairment disorders

Synonym
developmental delay, intellectual disability

Research involving
Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum
Source(s) of monetary or material Support: Gisela Thier fellowship LUMC

Intervention

Keyword: genetics, intellectual disability, neuronal development

Outcome measures

Primary outcome

Insight in the pathophysiological mechanisms leading to intellectual

disability.

Secondary outcome

NA

Study description

Background summary

Although each individual genetic cause of intellectual disability is rare, altogether about 1% of the population has some degree of intellectual disability. Although the genetic cause is known in a subgroup of these patients, there is usually very little knowledge on the pathophysiological mechanisms that lead to intellectual disability. An important problem in this research is the nervous tissue is not accessible. THis problem can now be overcome using induced pluripotent stem cells.

Study objective

1) to gain insight in the pathophysiological mechanisms leading to intellectual disability and 2) to create a model system which allows screening of therpeutic interventions for efficacy.

Study design

Comparing patient cell lines with control cell lines.

Study burden and risks

A single skin biopsy. Typically patients report this to be slightly more uncomfortable than a blood draw. The risks are 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

Intellectual disability
Either known molecular or clinical diagnosis

Exclusion criteria

Geen

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	30-04-2013
Enrollment:	50
Type:	Actual

Ethics review

Approved WMO

Date:	06-03-2013
Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden) metc-ldd@lumc.nl

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	NL43069.058.12