# Generation of induced pluripotent stem cells from patients with genetic neurodevelopmental disorders to study disease mechanisms

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Here we will generate iPS cells from patients with genetic neurodevelopmental disorders and (related) healthy control individuals, that can be used to generate in vitro disease models to further study the mechanisms behind these disorders.

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| **Ethical review** | Approved WMO |
| **Status** | Recruiting |
| **Health condition type** | Neurological disorders congenital |
| **Study type** | Observational invasive |

## Summary

### ID

NL-OMON44331

**Source**
ToetsingOnline

**Brief title**
iPSCs for understanding neurodevelopment

### Condition

- Neurological disorders congenital

**Synonym**
genetic disorders of brain development

**Research involving**
Human

### Sponsors and support

**Primary sponsor:** Erasmus MC, afdeling Klinische Genetica

**Source(s) of monetary or material Support:** Ministerie van OC&W,NWO (ZonMW Veni aan dr. Barakat)

## Intervention

**Keyword:** -clinical genetics, -disease modelling, -Induced pluripotent stem cells, -neurodevelopment

## Outcome measures

### Primary outcome

the generation of iPS cells from patient material (blood or skin cells), that

have either already been sampled during the diagnostic work-up of the patients,

or will be collected when other procedures are scheduled.

### Secondary outcome

the generation of disease models from the generated iPS cells which will allow

further functional investigations into the disease mechanisms.

# Study description

### Background summary

Neurodevelopmental disorders are frequently occurring, and incompletely understood. Patients are frequently referred to Clinical geneticists for diagnostics and counseling. Recent advantages in DNA diagnostics has discovered many novel disease associated genes and mutations, but it is still unclear how they are mechanistically implicated in these disorders. Additional research is needed to obtain novel insights in these processes. Recently, induced pluripotent stem (iPS) cells have been described, that can be generated from blood or skin cells from patients by reprogramming. These cells, and made from patients and proper controls, can be differentiated in vitro towards neuronal cell lineages, and can be used as disease models to study genetic disorders. By using these models, novel insights can be obtained in the disease pathogenesis contributing to these disorders.

### Study objective

Here we will generate iPS cells from patients with genetic neurodevelopmental

disorders and (related) healthy control individuals, that can be used to generate in vitro disease models to further study the mechanisms behind these disorders.

**Study design**

iPS cells will be generated from blood or skin cells of patients, by iPS reprogramming. Patients routinely visit our clinic for DNA diagnostics and counseling. Cells from patients will be collected in combination with routine clinical diagnostic procedures, or when other procedures are planned. In addition, already sampled cell lines from patients can be used. Therefore, the burden for the individuals participating in this research will be reduced to a minimum.

**Study burden and risks**

Blood samples or skin biopsies will be collected from patients and control individuals. Cells that have already been collected during routine clinical diagnostic work-up can be used, or cells will be collected when additional planned procedures happen, so that additional burden for the patients will be reduced to a minimum

# Contacts

**Public**
Selecteer

Wytemaweg 80
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**Scientific**
Selecteer

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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

patients with genetic causes of neurodevelopmental disorders. In principle patients of all ages are eligible, as some forms of neurodevelopmental disorders present already at young age. Healthy individuals either related or unrelated to the patient.

## Exclusion criteria

-Patient tested as HIV or hepatitis positive.
-Healthy control individuals: Partners of patients and employees of Erasmus MC department of Clinical Genetics are excluded.

# Study design

## Design

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|---|---|
| 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: | Recruiting |
| Start date (anticipated): | 29-10-2017 |

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|---|---|
| Enrollment: | 40 |
| Type: | Actual |

## Ethics review

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| Approved WMO Date: | 01-09-2017 |
| Application type: | First submission |
| Review commission: | METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam) |

## 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 | NL62031.078.17 |