# Genetic sequencing study in Primary Ciliary Dyskinesia.

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Objectives: The aim of this study is to:I Identify pathogenic mutations in novel genes, causing PCDII Develop a diagnostic PCD test, based on MPSIII Validate MPS technique for PCD.

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| **Ethical review** | Approved WMO |
| **Status** | Recruitment stopped |
| **Health condition type** | Respiratory disorders congenital |
| **Study type** | Observational invasive |

## Summary

### ID

NL-OMON37743

**Source**
ToetsingOnline

**Brief title**
Genetic sequencing study in Primairy Ciliary Dyskinesia.

### Condition

- Respiratory disorders congenital
- Respiratory tract infections

**Synonym**
immotile cilia syndrome, Primary Cliliary Dyskinesia

**Research involving**
Human

### Sponsors and support

**Primary sponsor:** Vrije Universiteit Medisch Centrum
**Source(s) of monetary or material Support:** Ministerie van OC&W,Fonds NutsOhra

## Intervention

**Keyword:** DNA, PCD, Sequence analysis

## Outcome measures

### Primary outcome

Main study endpoints:

I Pathogenic mutations found in novel genes: gene, type, number

II Pathogenic mutations found in selected known genes: gene, type, number.

III Diagnostic test, based on a set of PCD genes

IV Accuracy of the MPS technique after validation. Sensitivity and specificity

for identifying

known mutations.

### Secondary outcome

None

# Study description

### Background summary

Rationale:
Early diagnosis of Primary Ciliary Dyskinesia (PCD) is important for the preservation of lung function and quality of life. The diagnosis of PCD is difficult, as a single gold standard is lacking. At present there is no genetic test available for PCD due to the many genes that are involved, of which 60-70% are still unknown. Recent advances in genetic sequencing technologies enable DNA sequencing for many patients and hundreds of genes simultaneously. Massive Parallel Sequencing (MPS) is a fast and reliable technique to find mutations in DNA. This offers the possibility to develop a diagnostic test for PCD and find mutations in novel genes associated with the disease.

### Study objective

Objectives:

The aim of this study is to:
I Identify pathogenic mutations in novel genes, causing PCD
II Develop a diagnostic PCD test, based on MPS
III Validate MPS technique for PCD.

### Study design

This is an observational study, performed by the department of clinical genetics in collaboration with the department of pediatric pulmonology and pulmonary diseases of the VU University Medical Center, Amsterdam, the Netherlands. This study is intended to run from February 2012 till February 2015.

### Study burden and risks

Children are important to include in this study because it is likely that every family has its own unique mutation(s). To design a proper diagnostic test for PCD, as many as possible associated mutations should be included. Due to the recessive heredity of the disease we would not be able to find most mutations that cause PCD in the Netherlands if only adult patients would be included in this study. Participation of patients requires a DNA sample which will be obtained from saliva. By using this non-invasive procedure to obtain DNA we ensure that there are no risks and the burden for children and adults is minimized.

## 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

Primary Ciliary Dyskinesia

### Exclusion criteria

Other recessive hereditary diseases, known Volendam PCD mutation

## Study design

### Design

**Study type:** Observational invasive

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

### Recruitment

NL

| | |
|---|---|
| Recruitment status: | Recruitment stopped |
| Start date (anticipated): | 15-01-2013 |
| Enrollment: | 83 |
| Type: | Actual |

# Ethics review

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|---|---|
| Approved WMO Date: | 05-04-2012 |
| Application type: | First submission |
| Review commission: | METC Amsterdam UMC |
| Approved WMO Date: | 12-12-2012 |
| Application type: | Amendment |
| 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 | NL38804.029.12 |