Genetic characteristics of Primary Ciliary Dyskinesia.

No registrations found.

Ethical review	Not applicable
Status	Pending
Health condition type	-
Study type	Observational non invasive

Summary

ID

NL-OMON24319

Source
NTR

Health condition

Primary Ciliary Dyskinesia

Sponsors and support

Primary sponsor: VU University Medical Center
Source(s) of monetary or material Support: VU University Medical Center
Fonds NutsOhra

Intervention

Outcome measures

Primary outcome

1. Accuracy of MPS in detecting PCD mutation (validation);

2. Possible pathogenic mutations causing PCD.

Secondary outcome

N/A

Study description

Background summary

Primary Ciliary Dyskinesia (PCD) is an autosomal recessive hereditary disorder that causes dysfunction of cilia. Patients suffer from frequent respiratory infections and often develop bronchiectasis. Diagnosing PCD is difficult as a single gold standard is lacking. The diagnosis is usually based on a combination of clinical symptoms, abnormal movement of cilia on microscopic evaluation of respiratory epithelial biopsies and epithelial cell cultures, and/or identification of an ultra structural defect in the cilia by electron microscopy. Genetic testing is time consuming and very costly as there are many large genes involved. However, recent developments enable rapid DNA sequencing of many fragments in parallel.

We aim to validate Massive Parallel Sequencing (MPS) in Primary Ciliary Dyskinesia and identify novel disease causing mutations in our Dutch PCD population.

We aim to include all children and adults with PCD visiting the VU University Medical Center, Amsterdam.

Study objective

We aim to validate Massive Parallel Sequencing in Primary Ciliary Dyskinesia (PCD) and identify novel disease causing mutations in the Dutch PCD population.

Study design

N/A

Intervention

N/A

Contacts

Public
Postbus 7057
T. Paff
Department of pulmonology
VU University Medical Center, Amsterdam

Room PK 4X 023
Amsterdam 1007 MB
The Netherlands
+31 (0)20 4445491
Scientific
Postbus 7057
T. Paff
Department of pulmonology
VU University Medical Center, Amsterdam
Room PK 4X 023
Amsterdam 1007 MB
The Netherlands
+31 (0)20 4445491

Eligibility criteria

Inclusion criteria

Primary Ciliary Dyskinesia.

Exclusion criteria

Other recessive hereditary disorders.

Study design

Design

Study type:	Observational non invasive
Intervention model:	Parallel
Allocation:	Non controlled trial
Masking:	Open (masking not used)
Control:	N/A , unknown

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-01-2012
Enrollment:	83
Type:	Anticipated

Ethics review

Not applicable
Application type: Not applicable

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
NTR-new	NL2990
NTR-old	NTR3138
Other	CWO VUmc : pro 11/68
ISRCTN	ISRCTN wordt niet meer aangevraagd.

Study results

Summary results
N/A