Gene expression during ciliagenesis: Towards a novel candidate gene list for Primary Ciliary Dyskinesia.

No registrations found.

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

Summary

ID

NL-OMON27807

Source
Nationaal Trial Register

Health condition

Primary Ciliary Dyskinesia

Sponsors and support

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

Intervention

Outcome measures

Primary outcome

Gene expression levels and pattern.

Secondary outcome

Cell culture success rate with brush biopsies.

Study description

Background summary

Primary Ciliary Dyskinesia (PCD) is a hereditary disorder occurring in 1:15.000-30.000 live births, with increased frequency in genetically isolated populations. PCD is characterised by dyskinesia of epithelial cilia causing chronic respiratory disease. Diagnosing patients can be challenging, as there is no gold standard test. Due to the many unknown disease causing gene defects, genetic testing is still not possible in most cases. To this date, our understanding of the molecular composition of cilia is far from complete. Investigating which genes are important in cilia genesis will contribute to a more complete candidate gene list and development of a diagnostic test for Primary Ciliary Dyskinesia.

Objective of the study:

We aim to assess gene expression patterns in cilia-producing cells from healthy controls to develop a candidate gene list for PCD.

Study design:

This observational study will be conducted at the VU University Medical Center. Nasal curette and brush biopsies will be performed on healthy volunteers to obtain respiratory epithelial cells. Cells will be cultured and RNA will be isolated at three different time points during cilia genesis. Subsequently, Affymetrix gene expression arrays will be performed on cDNA and analyzed by cluster analysis.

Study population:

Six healthy volunteers (employees from the Dept. of Pulmonary Medicine/Pediatrics/Clinical Genetics) will be recruited to participate in this study.

Primary study parameters/outcome of the study:

Gene expression levels and pattern.

Secundary study parameters/outcome of the study:

Cell culture success rate with brush biopsies.

Study objective

We aim to assess gene expression patterns in cilia-producing cells from healthy controls to develop a candidate gene list for PCD. We hypothesise that:

1. Known PCD genes show similar gene expression patterns during ciliagenesis;

2. Novel candidate genes can be identified by clustering genes with similar expression patterns.

Study design

RNA will be isolated at three different time points during ciliagenesis (before, during and after the growth of cilia).

Intervention

Nasal curette and brush biopsies will be performed on healthy volunteers to obtain respiratory epithelial cells. Cells will be cultured and RNA will be isolated at three different time points during cilia genesis. Subsequently, Affymetrix gene expression arrays will be performed on cDNA and analyzed by cluster analysis.

Contacts

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Eligibility criteria

Inclusion criteria

≥ 18 years of age.

Exclusion criteria

1. Any signs of upper or lower airway infection;

2. Coagulation disorders (or any symptoms of easy bruising, heavy bleedings);

3. Use of anti-coagulants.

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):	01-05-2013
Enrollment:	6
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	NL3717
NTR-old	NTR3880
Other	CMO VUmc : Pro 12/99
ISRCTN	ISRCTN wordt niet meer aangevraagd.

Study results

Summary results
N/A