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

Gepubliceerd: 04-03-2013 Laatst bijgewerkt: 18-08-2022

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...

Ethische beoordeling	Niet van toepassing
Status	Werving nog niet gestart
Type aandoening	-
Onderzoekstype	Observationeel onderzoek, zonder invasieve metingen

Samenvatting

ID

NL-OMON27807

Bron
Nationaal Trial Register

Aandoening

Primary Ciliary Dyskinesia

Ondersteuning

Primaire sponsor: VU University Medical Center, Amsterdam
Overige ondersteuning: Sponsor and Fonds NutsOhra

Onderzoeksproduct en/of interventie

Uitkomstmaten

Primaire uitkomstmaten

Gene expression levels and pattern.

Toelichting onderzoek

Achtergrond van het onderzoek

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.

Doel van het onderzoek

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.

Onderzoeksopzet

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

Onderzoeksproduct en/of interventie

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.

Contactpersonen

Publiek

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

Wetenschappelijk

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

Deelname eisen

Belangrijkste voorwaarden om deel te mogen nemen (Inclusiecriteria)

≥ 18 years of age.

Belangrijkste redenen om niet deel te kunnen nemen (Exclusiecriteria)

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.

Onderzoeksopzet

Opzet

Type:	Observationeel onderzoek, zonder invasieve metingen
Onderzoeksmodel:	Parallel
Toewijzing:	N.v.t. / één studie arm
Blindering:	Open / niet geblindeerd
Controle:	N.v.t. / onbekend

Deelname

Nederland
Status: Werving nog niet gestart

(Verwachte) startdatum:	01-05-2013
Aantal proefpersonen:	6
Type:	Verwachte startdatum

Ethische beoordeling

Niet van toepassing

Soort:	Niet van toepassing

Registraties

Opgevolgd door onderstaande (mogelijk meer actuele) registratie

Geen registraties gevonden.

Andere (mogelijk minder actuele) registraties in dit register

Geen registraties gevonden.

In overige registers

Register	ID
NTR-new	NL3717
NTR-old	NTR3880
Ander register	CMO VUmc : Pro 12/99
ISRCTN	ISRCTN wordt niet meer aangevraagd.

Resultaten

Samenvatting resultaten
N/A