Prevalance of Birt-Hogg-Dubé syndrome among patients with (hereditary) spontaneous pneumothorax

Published: 18-06-2015
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To assess the prevalence of BHD among patients with spontaneous sporadic and familial pneumothorax.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON44161

Source
ToetsingOnline

Brief title
Prevalence of BHD in spontaneous pneumothorax

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Renal disorders (excl nephropathies)
- Lower respiratory tract disorders (excl obstruction and infection)

Synonym
Pneumothorax

Research involving
Human

Sponsors and support

Primary sponsor: Vrije Universiteit Medisch Centrum
Source(s) of monetary or material Support: Stichtingen zoals Swieringa Stichting en

Bakhuis Roozeboom stichting

Intervention

Keyword: Birt-Hogg Dubé syndrome, Folliculin, renal cancer, spontaneous pneumothorax

Outcome measures

Primary outcome

FLCN mutation analysis (on DNA)

Lungcysts (CT)

Secondary outcome

None

Study description

Background summary

Birt-Hogg-Dubé (BHD) syndrome is a rare autosomal dominant disease, characterized by skin fibrofolliculomas, renal cancer, multiple lung cysts and pneumothorax.
The BHD-Syndrome is caused by a mutation in the FLCN-gene, which is localised on chromosome 17, and encodes for the protein folliculin.
In over 80 percent of the patients with clinical BHD syndrome, a pathogenic FLCN mutation can be demonstrated. The clinical expression is variable: patients may present with skin lesions, renal cancer or pneumothorax.
The skin signs consist of are 1-5 mm multiple white or skin-coloured papules, in particular on the face, neck and torso. These fibrofolliculomas are benign lesions originating from hair follicles. Treatment is cosmetic with laser techniques .
The observed prevalence of renal cancer in BHD-patients has varied between 6 and 35%. Renal cancer in BHD is diagnosed at a relative young age and is often multifocal and/ or bilateral. For early detection and treatment FLCN mutation carriers are advised to undergo yearly renal ultrasound.
Lung cysts have been found in 80-90% of patients with BHD syndrome, more often localised basally than in the apical regions. These lung cysts may cause pneumothorax. In about 25% of FLCN mutation carriers pneumothorax before the age of 50 has been observed. Recurrent pneumothorax is common.
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Familial occurrence of pneumothorax is found in about 10% the patients with

spontaneous pneumothorax.
As stated above, the clinical expression of BHD is variable. BHD families with *pneumothorax-only*have been observed without skin or renal lesions.
Based on recent studies, 15-25% of the familial pneumothorax is due to an underlying FLCN mutation.

Study objective

To assess the prevalence of BHD among patients with spontaneous sporadic and familial pneumothorax.

Study design

Based on the results of our pilotstudy in VUmc, in which 3 out of 40 tested patients had a pathological FLCN mutation, we decided to extend the study to a second center; Rijnstate ziekenhuis Arnhem.
In this hospital was performed retrospective research in february 2014. We searched in 750 patientfiles. Totally, we included 450 PSP patients who were admitted in the pulmonary ward between 2003 and 2013. In the dossiers we searched for medical history, pneumothorax side and recurrence, diagnostic imaging, treatment, co-morbidity, complications of treatment, skinabnormalities, kidneydisease, smoking behaviour, medication, and familial incidence of pneumothorax and other diseases.
This group of patients will recieve a lettre with explanation of the research and a questionary in which we ask them their medical status, co-morbidty, pneumothorax (number and side), smkoing behaviour, use of drugs, familial incidence of pneumothorax and other diseases.

Study burden and risks

Patients who have given consent to be included in further research, will be invited to come to our outpatient clinic for one visit. As above discribed, it concernes a onetime non-invasive examination, the patient will sign a consent form. The participation will include a low dose CT scan (2mSv) and the 2 venous blood samples (2x8ml).

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

18 years and older, spontaneous pneumothorax in medical history

Exclusion criteria

younger than 18 years old, traumatic / iatrogenic pneumothorax

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	21-04-2016
Enrollment:	200
Type:	Actual

Ethics review

Approved WMO Date:	18-06-2015
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	15-03-2016
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

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	NL50605.091.14