The prevalence of Birt-Hogg-Dubé syndrome in spontaneous pneumothorax patients.

Published: 13-10-2011
Last updated: 30-11-2024

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

Ethical review	Approved WMO
Status	Will not start
Health condition type	Chromosomal abnormalities, gene alterations and gene variants
Study type	Observational invasive

Summary

ID

NL-OMON36416

Source
ToetsingOnline

Brief title
BHD in pneumothorax patients.

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Renal disorders (excl nephropathies)
- Respiratory disorders NEC

Synonym
Pneumothorax

Research involving
Human

Sponsors and support

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

Intervention

Keyword: Birt-Hogg-Dubé, Folliculin, Renal cancer, Spontaneous Pneumothorax

Outcome measures

Primary outcome

- Yield of FLCN mutation analysis (in DNA)

-Prevalence of fibrofolliculomas

- Prevalence of lung cysts (on the CT-Thorax)

- Prevalence of renal tumoutrs (on ultrasound & MRI)

Prevalence of familial cases

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

In 2009 a retrospective study was performed wherein 523 patients were included who, in the period 1990-2008 were treated for spontaneous pneumothorax at our hospital. For the present study we selected patients aged 18 years or older. Patients who died were excluded from evaluation. After exclusion the remaining 380 patients received a letter with questions about their medical history, pneumothorax, smoking behaviour, medication, and familial incidence of pneumothorax and other diseases.
Information was received from in total 94 patients. These patients consented to participate in a follow-up study.

The patients who gave permission for the follow up study will receive:
- An Invitation letter for the follow up study
- An Information brochure with information on Birt-Hogg-Dubé syndrome
- A consent form for examination at the outpatient Clinical Genetics
The investigations will consist of the following components:
- personal and family history
- expert dermatological examination which may include a skin biopsy
- The subgroup patients with familial pneumothorax will undergo CT-thorax (5mSv) and ultrasound & MRI of the kidneys.
- Two blood tubes will be taken for FLCN mutation analysis

Study burden and risks

None

Contacts

Public
Vrije Universiteit Medisch Centrum

De Boelelaan 1117
1007 MB Amsterdam
NL
Scientific
Vrije Universiteit Medisch Centrum

De Boelelaan 1117
1007 MB Amsterdam
NL

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:	Will not start
Start date (anticipated):	01-04-2014
Enrollment:	93
Type:	Anticipated

Ethics review

Approved WMO

Date:	13-10-2011
Application type:	First submission
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	NL31417.029.11