Pedigree research in families with Common Variable Immunodeficiency Disease.

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Using pedigree research in two CVID-families to find clues for genetic testing.

Ethical review	Approved WMO
Status	Pending
Health condition type	Immune system disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON30598

Source
ToetsingOnline

Brief title
CVID families

Condition

- Immune system disorders congenital
- Immunodeficiency syndromes

Synonym
common variable immunodeficiency disease, late onset hypogammaglobulinemia

Research involving
Human

Sponsors and support

Primary sponsor: Jeroen Bosch Ziekenhuis
Source(s) of monetary or material Support: interne financiering uit JBZ research fonds

Intervention

Keyword: pedigree, variable

Outcome measures

Primary outcome

1. With the help of department of clinical genetics: to make a pedigree of the

two families with CVID to see if they are related to each other. Are there

enough clues for genetic testing?

2. To determine immunologic parameters in CVID-patients and family members with

recurrent infections or other symptoms of CVID.

Secondary outcome

1. If there are enough clues for a genetic background of CVID in a family:

genetic testing of known CVID-genes.

2. If a genetic mutation is found: is there a correlation between immunologic

parameters and the found mutation?

Study description

Background summary

Early identification of children with a primary immunodeficiency (PID) in the large pool of children presenting with recurrent infections is not an easy task, but important for their prognosis. A delay in diagnosis is associated with considerable morbidity and increased mortality. The most common PID is Common Variable Immunodeficiency (CVID). Once a CVID has been established on clinical grounds, genetic identification of the defect is possible. A detailed family history can help to unravel the mode of inheritance in a family, and enables the attending physician to select the appropriate family members for

genetic testing.

Study objective

Using pedigree research in two CVID-families to find clues for genetic testing.

Study design

Observational study.

Study burden and risks

Per visit per person: 30 minutes. One to two blood samples are taken. Risk: hematoma associated with punction.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age

Adolescents (12-15 years)

Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)
Elderly (65 years and older)

Inclusion criteria

*patients with CVID treated at the department of pediatrics of the Jeroen Bosch Hospital.
*genetic interesting relatives of the CVID patients, based on the family history and pedigree.
*relatives with suspicion of immunodeficiency based on clinical records.

Exclusion criteria

Illness or recent infection (<2 weeks prior to inclusion).

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-10-2006
Enrollment:	20
Type:	Anticipated

Ethics review

Approved WMO

Application type:	First submission
Review commission:	METC Brabant (Tilburg)

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	NL15096.028.06