# Nasal Potential Difference in suspected CF patients with 5T polymorphism * Comparison between patients from Israel and Netherlands

Published: 04-09-2013
Last updated: 26-04-2024

Clarify the effect of 5T polymorphism with or without another CF causing mutation on the other allele, on the CFTR function.

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| **Ethical review** | Not approved |
| **Status** | Will not start |
| **Health condition type** | Congenital reproductive tract and breast disorders |
| **Study type** | Observational invasive |

## Summary

### ID

NL-OMON39838

**Source**
ToetsingOnline

**Brief title**
NPD in 5T patients

### Condition

- Congenital reproductive tract and breast disorders
- Respiratory disorders NEC

**Synonym**
Cystic Fibrosis, Mucoviscidosis

**Research involving**
Human

### Sponsors and support

**Primary sponsor:** Universitair Medisch Centrum Utrecht

**Source(s) of monetary or material Support:** Ministerie van OC&W

## Intervention

**Keyword:** 5T polymorphism, CF, CFTR function, NPD

## Outcome measures

### Primary outcome

Potential differences in NPD measurement.

### Secondary outcome

None.

# Study description

### Background summary

For different CFTR mutations is is known that they can ause CF. However for some mutations in the CFTR gene it is not certain if they cause CF or in combination with which mutations they might. 5T polymorphisme is one of those mutations. Persons who are homozygous for this mutation, or persons who have this mutation on one allele and have no CF causing mutation on the other, have an increased chance of CBAVD (congenital bilateral absence of vas deferens). Hyterozygocy for 5T polymophism combined with a known CF-causing mutation is associated with CF with atypical presentation. In Ashkenazi Jews this mutation is present in 18% of CF patients with atypical presentation, in not-Ashkenazi Jews this is 10%. In CBAVD in Askenazi Jews, 32% of cases is due to 5T polymorphism and in non-Askenazim this is 36%. This mutation is also present in other ethnic groups. Is is not know to what extent this mutation in itself or in combination with another mutation leads to a reduction in CFTR function, and to what extent this leads to health complaints. Through this study we want to clarify this. Possibly CFTR function is dependent on the mutation that is present on the other CFTR allele. Those mutations probably differ between the Dutch and Israeli population, which is why we want to compare Israeli patients (data already available) with Dutch patients.

### Study objective

Clarify the effect of 5T polymorphism with or without another CF causing mutation on the other allele, on the CFTR function.

### Study design

An NPD test is performed in about 30 persons with a 5T polymophism. The average values of the test are calculated for the group of 5T homozygotes and for de group of persons with one 5T in combination with another CF causing mutation. These values are compared with the referece values for NPD. Through an ANOVA test the significance of the difference between the outcomes of the groups is assessed.
Additionally the 95% CI is calculated per group and overlap in the values is evaluated.
The same is done or de patients in Israel, and the Dutch patient groups will be compared to de Israeli patient groups.

### Study burden and risks

The burden and risk of perticipation are minimal.

The patient needs to sit still throughout the measurement in a neutral position.
The salt solutions and subcutaneous needle used, are safe. Some iiritation or bleeding of the nasal mucosa may occur.

## 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

presence of 5T polymorphism on one or both alleles

### Exclusion criteria

chronic irritation of nasal epithelium

## Study design

### Design

**Study type:** Observational invasive

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

### Recruitment

NL

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|---|---|
| Recruitment status: | Will not start |
| Enrollment: | 30 |
| Type: | Anticipated |

## Ethics review

Not approved

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|---|---|
| Date: | 04-09-2013 |
| Application type: | First submission |
| Review commission: | METC Universitair Medisch Centrum Utrecht (Utrecht) |

# 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 | NL42164.041.12 |