# Which role do genetic polymorphisms play in relation to respiratory tract infections in children with Down syndrome ?

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To investigate the relation between a known genetic variation in a set of selected genes involved in the recognition of microorganisms and inflammation and the frequency of respiratory tract infections in a group of children with DS.

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|---|---|
| **Ethical review** | Approved WMO |
| **Status** | Recruiting |
| **Health condition type** | Chromosomal abnormalities, gene alterations and gene variants |
| **Study type** | Observational non invasive |

## Summary

### ID

NL-OMON33938

**Source**
ToetsingOnline

**Brief title**
TLR polymorphisms in Down syndrome

### Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Immune disorders NEC
- Hepatobiliary neoplasms malignant and unspecified

**Synonym**
Down Syndrome, innate immunity

**Research involving**
Human

## Sponsors and support

**Primary sponsor:** Vrije Universiteit Medisch Centrum
**Source(s) of monetary or material Support:** Janssen-Cilag

## Intervention

**Keyword:** children, Down syndrome, respiratory tract infection, TLR ( Toll-like receptor)

## Outcome measures

### Primary outcome

To investigate the genetic variation in TLRs and related genes in children with

DS compared to a cohort of healthy controls.

### Secondary outcome

To investigate if a relationship exists between certain genetic polymorphisms

in TLR genes and an increased susceptibility to respiratory tract infections

within the group children with DS.

# Study description

### Background summary

Children with Down syndrome ( DS) are more prone to respiratory tract infections than healthy children: both the severeness and frequency of respiratory tract infections are increased in children with DS.
This results in an increased frequency of hospitalizations. One of the causes of this increased frequency of respiratory tract infections in DS is an altered immune response : former investigations revealed a diminished cellular and humoral immune response. "Toll-like" receptors play a prominent role in initiating the cellular immune response.
Our hypothesis is that polymorphisms in the TLR genes in children with DS contribute to the higher frequency of respiratory tract infections in this group.

### Study objective

To investigate the relation between a known genetic variation in a set of

selected genes involved in the recognition of microorganisms and inflammation and the frequency of respiratory tract infections in a group of children with DS.

**Study design**

Study type: observational.

Duration of the study: one year

Location: Outpatient clinic of pediatric VU University Medical Center and Laboratory of Immunogenetics VU University Medical Center

A. Patient-related part: questionnaire for the parents of participating children with DS, concerning the frequency of infections in their children. Obtaining DNA of the child with DS by means of a buccal swab, which can be performed by the parents at home, by means of a small brush.

B.Laboratory-related part: analysis of Single Nucleotide Polymorphisms (SNPs) in TLR- and NOD genes by TaqMan analyses, PCR based RFLPs and VNTR analyses.

**Study burden and risks**

The burden and risks for the patient are minimal: the parents can obtain a buccal swab from their child ,at home, which is a very simple and painless procedure. The parents are asked to fill in a questionnaire concerning the frequency of infections their child has had. Participants will not have a direct benefit from this study. We consider however that the knowledge that we can obtain with this study can be helpful to develop future treatments for this group of patients.

# Contacts

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# Trial sites

## Listed location countries

Netherlands

# Eligibility criteria

**Age**
Adolescents (12-15 years)
Adolescents (16-17 years)
Children (2-11 years)

## Inclusion criteria

1.written informed consent from both parents or legal guardian(s)
2. age 0 until 18 years
3. sex: male and female
4. Down syndrome: trisomy 21

## Exclusion criteria

1. not meeting inclusion criteria
2. Down syndrome: translocation or mozaicism

# Study design

## Design

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|---|---|
| Study type: | Observational non invasive |
| Intervention model: | Other |
| Allocation: | Non-randomized controlled trial |
| Masking: | Open (masking not used) |
| Control: | Active |

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| Primary purpose: | Basic science |

### Recruitment

NL

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|---|---|
| Recruitment status: | Recruiting |
| Start date (anticipated): | 14-09-2009 |
| Enrollment: | 600 |
| Type: | Actual |

## Ethics review

Approved WMO

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|---|---|
| Date: | 09-03-2009 |
| 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 | NL24208.029.08 |