THE IMMUNE SYSTEM IN FOXP1 SYNDROME

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Primary Objective: We aim to provide a clear phenotype of the immune system in patients with FOXP1 syndrome. Secondary Objectives:We aim to correlate the immunophenotype of patients with FOXP1 syndrome with frequency and type of infections reported...

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

Summary

ID

NL-OMON52094

Source
ToetsingOnline

Brief title
FOXP1 immune system

Condition

- Chromosomal abnormalities, gene alterations and gene variants

Synonym
FOXP1 syndrome; FOXP1 mutation

Research involving
Human

Sponsors and support

Primary sponsor: Klinische genetica
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: FOXP1 syndrome, immune system, Regulatory T cells

Outcome measures

Primary outcome

Immune system phenotype

o Differential blood count

o Immunoglobins (IgA, IgG including IgG subclasses, IgM)

o Lymphocyte subset analysis

* CD19/CD20 B cell counts, including B cell subset analysis* CD3+ T cell

counts, including (naïve and memory) CD4+, CD8+ T cell counts

Second tier

o FOXP3+ regulatory T cell counts

o Functional analysis

Secondary outcome

Answers to questionnaires F1 and F2.

Demographics of the patient (age, gender)

Genetic mutation in FOXP1.

Study description

Background summary

Many persons with FOXP1 syndrome have frequent infecties.
Based on in vitro studies there seems to be an important role for FOXP1 in the immune system.

For a more detailed background, we refer to the study protocol.

Study objective

Primary Objective:
We aim to provide a clear phenotype of the immune system in patients with FOXP1 syndrome.

Secondary Objectives:
We aim to correlate the immunophenotype of patients with FOXP1 syndrome with frequency and type of infections reported by patients and/or their parents.
We aim to correlate the immunophenotype of patients with FOXP1 syndrome with frequency and type of auto-immune problems reported by patients and/or their parents.

Study design

This is a cross-sectional study without an intervention.

Study burden and risks

This study can only be performed in subjects having FOXP1 syndrome. There are no additional risks(since it is added to a planned clinical venapuncture).
Since many individuals with FOXP1 syndrome have frequent infections, the analysis of the immune system might lead to an explanation of the phenotype and possible preventive and/or therapeutic measures. The results will therefore be interpreted by an experience (paediatric) immunologist
The possible benefits for the group are that preventive and/or therapeutic measures may become available, and that the collection of these data may indicate whether analysis of the immune system should be performed in all individuals with FOXP1 syndrome.

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)

Inclusion criteria

- Pathogenic mutation in the FOXP1 gene
- Phenotype in accordance with genotype (i.e. neurodevelopmental problems, congenital abnormalities and/or dysmorphic features)
- Since age-appropriate references will be used for immune cell counts, there are no age limitations

Exclusion criteria

Allogenic stem cell transplantation (not a regular treatment for FOXP1 syndrome)

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled

Primary purpose:	Other

Recruitment

NL

Recruitment status:	Completed
Start date (anticipated):	05-10-2021
Enrollment:	30
Type:	Actual

Ethics review

Approved WMO

Date:	13-05-2022
Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden) metc-ldd@lumc.nl

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	NL78568.058.21