Effect of treatment with levothyroxine in a patient with thyroid hormone receptor *1 deficiency

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Objective: The primary objective of the study is to determine the effect of standard treatment with levothyroxine in TR*1 deficiency on:- Neutrophil and macrophage function- Erythropoiesis- Thyroid hormone target gene expression- Plasma metabolome

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Endocrine disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON42720

Source
ToetsingOnline

Brief title
Effect of treatment with levothyroxine in TR*1 deficiency

Condition

- Endocrine disorders congenital
- Thyroid gland disorders

Synonym
Thyroid hormone receptor alpha 1 deficiency

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Erythropoiesis, Innate immunity, Metabolomics, Thyroid hormone receptor 1 deficiency

Outcome measures

Primary outcome

Main study parameters/endpoints:

* Neutrophil and macrophage function prior to and following levothyroxine

treatment

* Erythropoiesis function

* Thyroid hormone target gene expression levels prior to and following

levothyroxine treatment

* Plasma metabolome prior to and following levothyroxine treatment

Secondary outcome

n.a.

Study description

Background summary

Thyroid hormone receptor *1 (TR*1) deficiency is a recently identified syndrome in which patients present with resistance to thyroid hormone at the tissue level characterised by growth retardation, cognitive defects and delayed bone development. The standard form of treatment for this disorder is levothyroxine. The effects of TR*1 deficiency and levothyroxine treatment on innate immune function, erythropoiesis, metabolomics and thyroid hormone target gene expression have not yet been studied. We have recently identified a new patient with TR*1 deficiency who has not yet started treatment. This patient presents a unique opportunity to study the role of TR* and the effects of treatment with levothyroxine in these physiological processes.

Study objective

Objective: The primary objective of the study is to determine the effect of standard treatment with levothyroxine in TR*1 deficiency on:
- Neutrophil and macrophage function
- Erythropoiesis
- Thyroid hormone target gene expression
- Plasma metabolome

Study design

Study design: Observational laboratory study

Study burden and risks

Nature and extent of the burden and risks associated with participation, benefit and group relatedness: The TR*1 deficient patient will undergo venipuncture 4 times in total (max. 73,5 ml). Once prior to the start of treatment and at 3, 6 and 12 months following the start of treatment. Venipunctures after the start of treatment will be scheduled to coincide with diagnostic venipunctures to avoid additional hospital visits. Healthy controls will undergo venipuncture only once (18 ml). Venous blood collection may result in minor discomfort and carries a small risk of bruising and bleeding.

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

For patient:
- Known carrier of inactivating thyroid hormone receptor alpha1 mutation
- Aged *18
- Willing and able to provide signed informed consent prior to study-related procedures.;For healthy controls:
- Aged *18
- Willing and able to provide signed informed consent prior to study-related procedures.

Exclusion criteria

Healthy controls:
- Fever or other symptoms of infection during the past 7 days.
- Use of medication that could affect haematological parameters or leukocyte function as specified by the Farmacotherapeutisch Kompas (http://www.fk.cvz.nl)

Study design

Design

Study type:	Observational invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

Recruitment

NL
Recruitment status: Recruitment stopped

Start date (anticipated):	27-05-2015
Enrollment:	12
Type:	Actual

Ethics review

Approved WMO Date:	20-05-2015
Application type:	First submission
Review commission:	METC Amsterdam UMC
Approved WMO Date:	23-07-2015
Application type:	Amendment
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	NL53342.018.15