Pituitary hormone secretion in patients with IGSF1 mutation

Published: 21-12-2012
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The objective of the current proposal is to further explore the pathophysiological changes that occur in patient with an IGSF1 mutation, using an established method for assessing hormone secretory characteristics.

Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Hypothalamus and pituitary gland disorders
Study type	Observational invasive

Summary

ID

NL-OMON37000

Source
ToetsingOnline

Brief title
Hormone secretion in IGSF1 mutation

Condition

- Hypothalamus and pituitary gland disorders

Synonym
Congenital hypothyroidism, decreased function of the thyroid gland present at birth

Research involving
Human

Sponsors and support

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

Intervention

Keyword: Circadian rhythm, Congenital central hypothyroidism, IGSF1, Pituitary hormone secretion

Outcome measures

Primary outcome

24-hour hormone profiles of:

- Growth hormone

- Follicle stimulating hormone

- Luteinizing hormone

- Thyroid stimulating hormone

Potentially in a later analysis:

- ACTH

- Cortisol

- Prolactin

Secondary outcome

Crosscorrelations between hormonal axes

Study description

Background summary

A defect in the IGSF1 gene causes a heterogenous phenotype of TSH deficiency, prolacin deficiency, growth hormone deficiency, macroorchidism and delayed testosteron rise during puberty. This syndrome has only recently been discovered, and so far 43 patients are known to have this gene defect worldwide (of which 30 live in the Netherlands). Little is known about the pathophysiology of this syndrome, and further studies are warrented to

elucidate the role of IGSF1 in the regulation of pituitary hormones.

Study objective

The objective of the current proposal is to further explore the pathophysiological changes that occur in patient with an IGSF1 mutation, using an established method for assessing hormone secretory characteristics.

Study design

We propose to perform 24-hr rhythm profiles (drawn blood every 10 minutes for 24 hr) to quantitate hormone secretion and to study correlations between several pituitary hormones. The 24-hr profiles will be analyzed with established techniques (deconvolution analysis, Approximate Entropy, and Cosinor analysis).

Study burden and risks

Burden:
Patients are only allowed to leave their hospital beds for toilet visits, which is a considerable restriction in their freedom of movement. Also, an intravenous canule will stay in situ for 24 hours, further limiting their freedom of movement. Finally, having an investigator draw blood every 10 minutes during the night might interfere with a subject's sleep comfort. The investigators consider this burden as not to be neglected, but within reasonable and acceptable boundries.

Risk:
Patient are at risk of undergoing more than one attempts to (re)place the intravenous canule.

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

1. Male patients who are known to have with a pathogenic IGSF1 mutation
2. Female carriers who are known to have with a pathogenic IGSF1 mutation
All eligible patients were previously tested for an IGSF1 mutation based on specific symptoms (familial central hypothyroidism) or based on susceptibility from pedigree analyses. The patients are familiar with their gene defect and have already received information from their physician about the possible implications of having this defect.
Patients will be informed about the nature of this study by their treating physician. If they are interested in participating, the primary investigators will contact them by phone and send written information about the study details.

Exclusion criteria

None.

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):	18-02-2013
Enrollment:	32
Type:	Actual

Ethics review

Approved WMO

Date:	21-12-2012
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	NL42392.058.12