Semen analysis in males with IGSF1 deficiency syndrome

Published: 08-04-2015
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Primary objective: 1) To examine the effect of IGSF1 deficiency syndrome on semen quality when compared to WHO reference ranges;2) The comparison of semen quality of these patients to that of healthy controls.To investigate the association between...

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

Summary

ID

NL-OMON44850

Source
ToetsingOnline

Brief title
Semen analysis in IDS

Condition

- Endocrine disorders congenital
- Sexual function and fertility disorders

Synonym
Macroorchidism; enlarged testes

Research involving
Human

Sponsors and support

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

Intervention

Keyword: IGSF1 deficiency syndrome, Macroorchidism, Semen analysis

Outcome measures

Primary outcome

1) Conventional semen analysis parameters, including: volume, concentration, sperm count, motility, vitality, pH, leucocytes, morphology, MAR-test and immunobead test compared to WHO reference ranges.

2) Serum concentrations of testosterone (and all metabolites), SHBG, FSH, LH and inhibin B, and their association with semen analysis parameters.

Secondary outcome

na

Study description

Background summary

In 2012 it was discovered that mutations in the X-bound IGSF1 gene leads to an illness characterized by central hypothyroidism, a delayed puberty and macroorchidism. Due to the recent discovery of this syndrome, there is still a lot unknown about the disease. For instance, it is unsure what causes the marked macroorchidism, and what it's effects are on the spermatogenesis.
In Fragile X-syndrome, also an X-bound disease with a similar growth pattern and macroorchidism, men were found to have a decreased semen quality. While these patients have a diminished fertility, they, like patients with IGSF1 mutations, have the ability to reproduce. There is nothing known about the fertility of men with IGSF1 mutations.
In the current study, the semen quality of men with IGSF1 mutations is evaluated.This study does not only provide more information about the fertility of these men, but may also offer more insight in the pathofysiology behind the macroorchidism.

Study objective

Primary objective:

1) To examine the effect of IGSF1 deficiency syndrome on semen quality when compared to WHO reference ranges;
2) The comparison of semen quality of these patients to that of healthy controls.To investigate the association between semen quality and endocrine parameters.

Study design

A cross-sectional study.

Study burden and risks

Venous blood sampling carries a small risk of bleeding and bruising.

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

- IGSF1 deficiency syndrome
- Aged *18 years
- Able to masturbate

Exclusion criteria

- Use of any medication known to affect sperm quality

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	05-04-2016
Enrollment:	20
Type:	Actual

Ethics review

Approved WMO

Date:	08-04-2015
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	NL48270.018.14