In search of thyroid hormone receptor alpha-1 mutations in adolescents with delayed puberty

Published: 30-11-2009
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To identify patients with TRalpha-1 mutations among adolescents with delayed puberty.

Ethical review	Approved WMO
Status	Pending
Health condition type	Thyroid gland disorders
Study type	Observational invasive

Summary

ID

NL-OMON33241

Source
ToetsingOnline

Brief title
TR alpha-1 mutations in adolescents with delayed puberty

Condition

- Thyroid gland disorders

Synonym
constitutional delay of puberty, delayed puberty

Research involving
Human

Sponsors and support

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

Intervention

Keyword: delayed puberty, thyroid hormone receptor alpha-1

Outcome measures

Primary outcome

TRalpha-1 mutations

Secondary outcome

not applicable

Study description

Background summary

Thyroid hormone receptor (TR) alpha and beta are nuclear receptors that mediate the transcriptional effects of triiodothyronine (T3). Many different mutations in TR beta are known to cause the syndrome of *resistance to thyroid hormone* (RTH). Surprisingly, no patients with germline mutations in TR alpha-1 have been identified. Recently the phenotype of mice with a TR alpha-1 mutation has been described. These mice are biochemically euthyroid but show severely delayed development and seem to catch up during adult life.
Our research aims to identify TR alpha-1 mutations in biochemically euthyroid patients who have a phenotype compatible with that of the TR alpha-1 mutant mice. We will identify candidate patients with delayed puberty, for which no other underlying cause has been found. Since these patients show a delayed puberty of unknown cause, but catch up during adult life they are good candidates for harbouring a mutation in the TR alpha-1 gene.

Study objective

To identify patients with TRalpha-1 mutations among adolescents with delayed puberty.

Study design

Patients with a previous diagnosis of delayed puberty will be asked to visit the outpatient clinic of the department of Pediatric Endocrinology at Emma Children*s Hospital AMC. The visit will consist of a short (structured) interview and a venous blood collection.
The blood will be analyzed for thyroid function determinants and possible

TRalpha-1 mutations.

Study burden and risks

The study could provide knowledge into a cause of delayed puberty. The burden to the subjects is minimal and consists of a one-time visit to the hospital, answering a questionnaire and undergoing one venous punction.

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

Diagnosis of delayed puberty of unknow origin, or defined as constitutional delay of puberty

Exclusion criteria

Permanent forms of hypogonadotropic hypogonadism
Hypergonadotropic hypogonadism
Age < 18 years

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-10-2009
Enrollment:	40
Type:	Anticipated

Ethics review

Approved WMO

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	NL29106.018.09