Genetic analysis of short stature

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a. analysis of the usefulness of new molecular biological techniques in the diagnostic process of short stature, especially the detection of small chromosomal anomalies.b. identification of genetic factors causing short stature or influencing the...

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

Summary

ID

NL-OMON30788

Source
ToetsingOnline

Brief title
Genetic analysis of short stature

Condition

- Chromosomal abnormalities, gene alterations and gene variants
- Endocrine and glandular disorders NEC
- Musculoskeletal and connective tissue disorders congenital

Synonym
dwarfism, shortness

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Gestreefd wordt naar externe subsidiëring;maar dit is nog niet gerealiseerd.

Intervention

Keyword: Genetics, Growth, Short stature

Outcome measures

Primary outcome

a. diagnostic possibilities in patients with short stature

b. genetic factors which are involved in short stature with or without

disproportion

c. the potential value of proteomics techniques in the diagnostic process of

short stature.

Secondary outcome

Not applicable.

Study description

Background summary

Short stature is a common diagnostic problem in pediatric and clinical genetic practice. With the routine diagnostic methods the cause of short stature remains unknown in a high percentage of patients, despite extensive diagnostic analyses. This study focuses on the improvement of diagnostic techniques, in order to come to a diagnosis more often.
In a series of patients with short stature, proportionate as well as disproportionate, blood will be taken. This material will be used for several molecular analyses, also known as system biology. We expect this approach to provide a better understanding of the causes of growth disorders leading to short stature with or without disproportion.

Study objective

a. analysis of the usefulness of new molecular biological techniques in the diagnostic process of short stature, especially the detection of small chromosomal anomalies.
b. identification of genetic factors causing short stature or influencing the severity of it.

c. analysis of protein profiles in plasma with proteomics techniques.

Study design

a. a human material bank (DNA) of persons with short stature will be constructed for research of causes of short stature. To facilitate the search for responsible genes DNA of both parents will also be stored.
b. the material will be used to validate new molecular biological techniques as a diagnostic tool in patients with short stature.
c. after the genetic confirmation of a diagnosis, associations with a certain protein profile in this genetic disorder will be investigated.

Study burden and risks

From every person blood will be taken and height measurements performed once. When DNA is available in the Laboratory of Diagnostic Genome Analysis (LDGA) in Leiden, or in another diagnostic DNA laboratory, blood sampling in this person will be abated. Blood will be taken from the parents for isolation of DNA.
These actions will hardly be hazardous.

Contacts

Public
Academisch Medisch Centrum

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Scientific
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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)
Elderly (65 years and older)

Inclusion criteria

Patients with short stature and their parents

Exclusion criteria

No permission;
biological parents not available;
medical information of the patient not available;
known diagnosis, confirmed by molecular, cytogenetic or cytological analysis

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruitment stopped
Start date (anticipated):	29-06-2009
Enrollment:	150
Type:	Actual

Ethics review

Approved WMO Date:	04-02-2009
Application type:	First submission
Review commission:	METC Leids Universitair Medisch Centrum (Leiden)

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	NL16112.058.07