Development of non-invasive diagnostic test based on fetal DNA isolated from maternal blood (NATERA-study)

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Our primary aim is to develop en perfection the diagnostic accuracy of the non-invasive tests for chromosomal aberrations in maternal blood.

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

Summary

ID

NL-OMON37477

Source
ToetsingOnline

Brief title
NATERA-studie

Condition

- Chromosomal abnormalities, gene alterations and gene variants

Synonym
chromosomal aberriations, genetic defect

Research involving
Human

Sponsors and support

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

Intervention

Keyword: chromosomal, fetal DNA, noninvasive

Outcome measures

Primary outcome

The development and perfection of a noninvasive test in maternal blood

detecting chromosomal aberrations.

Secondary outcome

not applicable

Study description

Background summary

Since the 1970s, pregnant women are offered prenatal screening or prenatal diagnostic testing for trisomies. The main aim of diagnostic testing is the detection of most common chromosomal aberrations.

The invasive tests (amniocentesis and chorion villus sampling) acquire fetal cells for further chromosomal research. De accuracy is highly reliable with a sensitivity and specificity of practically 100%. The main disadvantage however is the invasive nature of the diagnostic tests. Both tests are associated with a procedure-related fetal loss rate of 0.5-1%.

Recently, new approaches are introduced to analyze free fetal DNA in maternal blood in a non-invasive way. Published studies show a sensitivity and specificity of nearly 100%

For the further development of this test blood samples are needed of women with fetus with a known chromosomal aberration.

Study objective

Our primary aim is to develop en perfection the diagnostic accuracy of the non-invasive tests for chromosomal aberrations in maternal blood.

Study design

Pregnant women with a fetus with a proven chromosomal aberrations are asked to donate 2 blood tubes. The biological father is asked to donate either blood or a buccal or salivary samples.

In total internationally the following samples need to be collected:
* Up to 2,000 maternal blood samples along with their corresponding paternal blood, buccal or saliva samples (1,000 required for final analysis).
* Up to 200 maternal blood samples from carrying a fetus with a confirmed chromosomal abnormality or genetic disorder, along with their corresponding paternal blood, buccal or saliva samples (50 required for final analysis). For women who opted for termination, a genetic sample of the fetus may also be collected.
* Up to 1,000 buccal or saliva samples from paternal grandfathers and/or the biological father*s brothers.
* Up to 1,000 cord, buccal or saliva samples from the born children.
* Up to 40 blood samples (20 non-pregnant females and 20 males) from healthy volunteers (20 required for final analysis)
* Up to 400 blood samples from women undergoing D&C procedure following a miscarriage along with corresponding paternal blood (or buccal or saliva samples) (200 required for final analysis).

In the Netherlands the following samples will be collected:
* Up to 100 maternal blood samples from carrying a fetus with a confirmed chromosomal abnormality or genetic disorder, along with their corresponding 100 paternal blood, buccal or saliva samples. For women who opted for termination, a genetic sample of the fetus may also be collected.

Study burden and risks

Venous puncture: the amount of blood required for the study will not harm the participant or the fetus. The buccal or salivary swab will not harm the participant either.

Contacts

Public
Leids Universitair Medisch Centrum

Albinusdreef 2
2333 ZA Leiden
NL
Scientific
Leids Universitair Medisch Centrum

Albinusdreef 2
2333 ZA Leiden
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

Maternal blood samples:
> 18 years of age, informed consent given
diagnosis of abnormal or genetic disorder diagnosed by either amniocentesis or chorionic villus sampling
before (spontanious) abortion;Blood, buccal or saliva collection biological father:
>18 years, informed consent

Exclusion criteria

Maternal blood samples:
After spontanious abortion or dilation and curettage

Study design

Design

Study type: Observational invasive

Masking:	Open (masking not used)
Control:	Uncontrolled

Primary purpose:	Diagnostic

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	21-10-2013
Enrollment:	300
Type:	Actual

Ethics review

Approved WMO

Date:	01-02-2013
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	NL39980.058.12