Sex chromosome abnormality as co-finding when performing routine prenatal diagnostic procedures: diagnostic gain or damage? Study on the views of parents and professionals.

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1. To evaluate the consequences of the diagnosis of a sexchromosomal abnormality during routine prenatal diagnostic testing, in the course of which the experiences of the parents and professionals involved will be noted.2.To determine if it would be...

Ethical review	Approved WMO
Status	Pending
Health condition type	Other condition
Study type	Observational non invasive

Summary

ID

NL-OMON31988

Source
ToetsingOnline

Brief title
Routine fetal gender determination in prenatal testing: gain or damage?

Condition

- Other condition
- Chromosomal abnormalities, gene alterations and gene variants

Synonym
gender determination

Health condition

foetale geslachtsbepaling

Research involving
Human

Sponsors and support

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

Intervention

Keyword: gender determination, prenatal diagnosis, sex chromosomal abnormality, targeted testing

Outcome measures

Primary outcome

1.Did these parents desire to know the fetal sex after amniocenteses in week 15

of the pregnancy?

2.Were these parents willing to wait 4 weeks to know the fetal sex when sex

determination would be performed by ultrasound examination in week 20 of the

pregnancy?

3.How have they experienced the consequences of the diagnosis of a

sexchromosomal abnormality during routine prenatal diagnostic testing?

4.How do they look back on the events and how is their general quality of life

at this moment?

5. What is the opinion about this specific problem of the professionals

involved?

Secondary outcome

Factors associated with the parents* desire to know the sex of their baby.

(ref Shipp, 2004)

Study description

Background summary

At this moment, routine prenatal invasive tests in pregnancies at risk for Down syndrome automatically result in fetal sex determination. Pregnant women undergoing amniocenteses have the option to know the sex of their unborn baby after invasive testing even though there is no medical reason for one.
Detection of sex chromosomes with karyotyping or molecular testing (MLPA) often leads to unexpected findings. The phenotype of most sex chromosome aneuploidy cases is usually mildly affected; the risk for sexual development problems is increased, but most individuals fall in the normal range of development, and marked abnormality is not usually seen. Unexpected findings of the sex chromosomes or sex chromosomal mosaicisms present the prospective parents with a very difficult personal decision whether to continu or terminate the pregnancy.
With molecular testing, like the recently diagnostic implemented MLPA test, the possibilty arises to exclude the sex chromosomal targets in the test. Exclusion of the sex chromosomes in prenatal molecular testing results in a decrease of unexpected findings concerning the sex chromosomes.
For prospective parents fetal sex determination before birth is still possible with a prenatal ultrasound examination. In the Dutch prenatal screening programme, the fetal anomaly scan is routinely performed in week 20 of the pregnancy. Compared with fetal sex determination after amniocenteses, parents will have to wait 4 weeks to know the sex of their baby. Fetal sex detemination after prenatal invasive testing can potentially be replaced by the fetal anomaly scan.
The consequences of the diagnosis of a sexchromosomal abnormality as a co-finding during routine prenatal diagnostic testing will be evaluated.

Study objective

1. To evaluate the consequences of the diagnosis of a sexchromosomal abnormality during routine prenatal diagnostic testing, in the course of which the experiences of the parents and professionals involved will be noted.

2.To determine if it would be preferable to provide fetal gender determination by a non-invasive procedure (ultrasound) as opposed to gender determination by

invasive prenatal testing.

Study design

Retrospective qualitative study by semi-structured interviews and the administration of a validated Quality of Life Scale to parents who have been confrontated with a sexchromosomal abnormailty as a co-finding in a prenatal diagnostic procedure.

Professionals, who are in any way involved in these invasive diagnostic procedures, will be asked their opinion about above mentioned problem of a sexchromosomal abnormality as a co-finding and the specific problems this brings along for the parents involved.

Study burden and risks

An interview during 60-90 minutes, with the completion of a standardised questionnaire.
In the unexpected case, that talking about these events may bring up grieve, we will provide adequate psychological support.

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

Parents that have undergone an invasive prenatal diagnostic test in the last 15 years in the Nijmegen region because of maternal age or to exclude other serious chromosomal anomalies, who were faced with a fetal sex chromosomal abnormality as a co-finding in the karyotyping procedure.

Exclusion criteria

. Patients with a clear risk at a sex chromosomal abnormality, because of an earlier child with this same problem.
. Patients with insufficient fluency in Dutch.

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-05-2008
Enrollment:	30
Type:	Anticipated

Ethics review

Approved WMO
Application type: First submission

Review commission: CMO regio Arnhem-Nijmegen (Nijmegen)

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	NL21572.091.08