Abnormal number of fetal ribs or vertebrae and associated congenital abnormalities and adverse fetal outcome; The need for prenatal detection of rib and vertebral abnormalities by 2D&3D-ultrasound.

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Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Congenital and hereditary disorders NEC
Study type	Observational non invasive

Summary

ID

NL-OMON40300

Source
ToetsingOnline

Brief title
ASAP (Abnormal Spine And Prognosis)

Condition

- Congenital and hereditary disorders NEC
- Foetal complications

Synonym
rib and vertebral anomalies, vertebral column abnormalities

Research involving
Fetus in utero

Sponsors and support

Primary sponsor: Erasmus MC, Universitair Medisch Centrum Rotterdam
Source(s) of monetary or material Support: Ministerie van OC&W

Intervention

Keyword: Anomaly, Fetus, Rib, Vertebra

Outcome measures

Primary outcome

The number of ribs and/or vertebrae in fetuses with and without congenital malformations, aneuploidy and adverse fetal outcome.

Secondary outcome

Secondary objectives are to determine:

i) the most suitable and earliest gestational age for assessing number of vertebrae and ribs and shape by ultrasound

ii) whether it is feasible, when vertebral or rib anomalies are seen by ultrasound, to identify the level of anomalies in the vertebral column, i.e. cervical, thoracic or lumbar

iii) whether there is an association between an abnormal number of ribs or vertebrae and abnormalities of specific organ systems

iv) whether the severity of the malformations is correlated to the severity of the anomaly of the vertebral column

v) whether anomalies in vertebral or rib number are associated with specific genetic anomalies

vi) whether the malformations can be classified into recognizable phenotypic

patterns which may be caused by or associated with a single genetic anomaly

vii) whether genetic anomalies can be classified according to the vertebral

level where the disruption occurs

Study description

Background summary

Variations in number of vertebrae and ribs are more common in stillbirths and fetuses with congenital abnormalities and/or aneuploidy. This is probably caused by strong interactions in early organogenesis between axial patterning, mediated by Hox genes and other, simultaneously occurring morphogenetic processes.
Assessment of the number of vertebrae and ribs and their shape during prenatal sonographic examinations might be useful in determining prognosis of fetuses with a congenital abnormality.
Hypothetically, assessment of the fetal vertebral column by ultrasound can be reliably done from a gestational age of 13 weeks. The incidence of variations in number of ribs and vertebrae, especially in the cervical region, of fetuses without congenital anomalies is expected to be lower than in fetuses with congenital malformations. Furthermore, the severity of disturbance of the fetal vertebral column will probably be associated with the severity of congenital malformations and adverse fetal outcome. More severe disturbances will be located more cervical or involve more than one vertebral boundary.

Study objective

The primary objective is to determine if there is an association between the presence of vertebral and/or rib anomalies and congenital malformations, aneuploidy or adverse fetal outcome, in order to assess whether detailed evaluation of the vertebral column and ribs is therefore indicated.

Study design

1. Retrospective analysis of fetal volumes from 12 weeks of gestation in 4D view and/or the I-space (A four-walled CAVE Automatic Virtual Environment. A hologram is created by the V-Scope volume rendering application and this hologram can be manipulated by means of a virtual pointer that is controlled by a wireless joystick) to determine which gestational age provides the most suitable images for assessment of number of vertebrae and ribs and their shape.
2. Retrospective cohort study; assessment of number of vertebrae and ribs in fetuses with aneuploidy from whom a prenatal 3-dimensional ultrasound volume is

available.
3. Retrospective study of fetuses with congenital anomalies or aneuploidy of whom a prenatal ultrasound, autopsy, X-ray and/or genetic array are available, from 2009 * 2013. Analysis of possible correlation between an abnormal umber of ribs and/or vertebrae and structural anomalies and the presence of genetic anomalies seen on genomic SNP array.
4. Prospective study of fetuses without structural malformations; determination of number of vertebrae and ribs with 2D&3D-ultrasound in fetuses without congenital anomalies in a high-risk population at a gestational age around 20 weeks.
5. Prospective study of fetuses with malformations; determination of number of vertebrae and ribs with 2D&3D-ultrasound, and if available, correlation with X-ray from postnatal period or autopsy.
6. Analysis of genetic material of the fetuses and their parents in order to determine if specific genetic abnormalities are associated with an abnormal number of ribs and/or vertebrae and possible associated anomalies.

Study burden and risks

During the ultrasound examination, a 3-dimensional volume of the fetal vertebral column will be made and saved. No extra examinations will be performed. There are no indications for harmful effects of ultrasound in human subjects. The risk of participation in this study is therefore negligible.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Inclusion criteria

Pregnant women
- with a gestational age of at least 12 weeks
- of at least 18 years old
- who are mentally competent
- who are willing to sign the informed consent form

Exclusion criteria

Language barrier
No consensus on interpretation of ultrasound or Xrays between different observers
Insufficient ossification (taking visibility of ossification centres of the phalanges as a reference)

Study design

Design

Study type:	Observational non invasive
Intervention model:	Other
Allocation:	Non-randomized controlled trial
Masking:	Open (masking not used)
Control:	Active
Primary purpose:	Basic science

Recruitment

NL
Recruitment status: Recruitment stopped

Start date (anticipated):	19-05-2014
Enrollment:	260
Type:	Actual

Ethics review

Approved WMO Date:	24-04-2014
Application type:	First submission
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)
Approved WMO Date:	09-09-2015
Application type:	Amendment
Review commission:	METC Erasmus MC, Universitair Medisch Centrum Rotterdam (Rotterdam)

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	NL45536.078.14