Genetic control of the number of lumbar vertebrae in humans

Published: 14-12-2015
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Primary Objective: to investigate whether the variation of the number of lumbar vertebrae in humans is coded by the same genes which are responsible for this as well as in other mammals.

Ethical review	Approved WMO
Status	Pending
Health condition type	Musculoskeletal and connective tissue disorders congenital
Study type	Observational invasive

Summary

ID

NL-OMON41911

Source
ToetsingOnline

Brief title
Genetic control human spine

Condition

- Musculoskeletal and connective tissue disorders congenital

Synonym
back pain, development

Research involving
Human

Sponsors and support

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

Intervention

Keyword: genetics, spine

Outcome measures

Primary outcome

The endpoint of the study is the identification of the genes responsible for

the number of lumbar vertebrae in the human spinal column.

Secondary outcome

not applicable

Study description

Background summary

The number of vertebrae in mammals is roughly constant within single species. The number of cervical vertebrae is 7 in all mammals, while the number of thoracic and lumbar vertebrae varies from 17 to 24. For instance humans usually have 17 thoracic and lumbar vertebrae while pigs usually have 19. Approximately 10000 years ago man switched from a hunter-gatherer society to a farming society. Animals were domesticated and the genes of these domesticated species were modified by selective breeding. This had an effect on the number of lumbar vertebrae of pigs. In the domesticated European pig 6 lumbar vertebrae are found, while only 5 are found in Asian pigs and in European wild pigs. Similar variations in number of vertebrae occur naturally in many species. By selecting animals with a large number of vertebrae, European farmers changed the population into a uniform group with 6 lumbar vertebrae. In pigs, the variation in number of lumbar vertebrae is controlled by a specific group of genes.
In humans the same natural variation in the number of vertebrae occurs. A majority of 88% of the population has 5 vertebrae, while 4% of the population has 6 vertebrae and 8% of the population has 4. Furthermore, apes (chimpanzee, gorilla, orang-utan and gibbon) usually have only 4 lumbar vertebrae. This suggests that there is a genetic advantage for humans to have 5 lumbar vertebrae. Since humans are the only one to have bipedal locomotion this could be linked to motion. More specifically, the higher number of vertebrae might allow more rotation between pelvis and thorax which facilitates bipedal gait.
Like all mammals, pigs and humans share a common history. Approximately 85 million years ago, both groups separated into different lineages. The traces of their common past can still be found in the genes of both species. Large part

of the DNA of both species is the same, especially the genes that belong to the basic mammalian heritage. Since the variation in number of vertebrae in pigs is controlled by a specific group of genes, it can be hypothesized that the number of lumbar vertebrae in humans is controlled by these same group of genes.

Study objective

Primary Objective: to investigate whether the variation of the number of lumbar vertebrae in humans is coded by the same genes which are responsible for this as well as in other mammals.

Study design

observational study. The patient is selected by the physician on the basis of the number of vertebrae. Twenty five unrelated consecutive patients with 4 and 25 unrelated consecutive patients with 6 lumbar vertebrae will be recruited. In addition, 25 unrelated patients with 5 vertebrae will be recruited to obtain three groups of 25 subjects. From each of the 75 participants a 5 ml blood sample will be taken for the present study. Each patient will be assigned a unique number, which will allow the researchers to combine CT-images with blood samples. The tube, containing the blood will be labeled with M for male, F for female, a number describing the number of vertebrae and the assigned subject number. Blood samples will be taken according to the standard protocol at the *VU-medisch centrum prikpoli*.

Study burden and risks

burden and risks are low.

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

people who belong to the group of people with 4,5 or 6 lumbar vertebrae.
Subjects are recruited among patients of the department of orthopaedic surgery at the VU University Medical Center.
Only mentally competent patients, aged 18 years or older, are included in the study.

Exclusion criteria

congenital deformations and/or blood relatives to a person who is already included in the study.
Minors and/or incompetent patients are excluded from the study.
Patients who do not want to be informed of chance findings may not participate in this study.

Study design

Design

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

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-01-2016
Enrollment:	75
Type:	Anticipated

Ethics review

Approved WMO

Date:	14-12-2015
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	NL49786.029.14