Gene mutations and -variations in congenital heart defects

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Identification of genes that are involved in congenital heart defects.

Ethical review	Approved WMO
Status	Pending
Health condition type	Congenital cardiac disorders
Study type	Observational invasive

Summary

ID

NL-OMON31069

Source
ToetsingOnline

Brief title
Gene mutations and -variations in congenital heart defects

Condition

- Congenital cardiac disorders
- Cardiac and vascular disorders congenital

Synonym
congenital cardiovascular malformation, congenital heart defect

Research involving
Human

Sponsors and support

Primary sponsor: Academisch Medisch Centrum
Source(s) of monetary or material Support: Europese Unie

Intervention

Keyword: congenital heart defects, genes, mutations, variations

Outcome measures

Primary outcome

A genetic variation in relatives, with or without congenital heart defects, of patients with a congenital heart defect and a proven genetic variation.

Occurrence of (mild) congenital heart defects in relatives, with or without the genetic variation found in de index patient, of patients with a congenital heart defect and a proven genetic variation.

Secondary outcome

Not applicable

Study description

Background summary

Congenital heart defects are frequent, affecting approximately 1% of newborn babies (8 per 1,000). Little is known about the genes that are involved in isolated congenital heart defects. Research on genetic causes of congenital heart defects provides insight into the developmental processes underlying these heart defects. In the future, this may lead to a personalized therapeutic strategy and adequate genetic counseling for patients and their relatives. It could also identify interacting environmental elements that might be prevented.

Study objective

Identification of genes that are involved in congenital heart defects.

Study design

Non-randomized monocenter observational cross-sectional study.
Patients registered in the CONCOR DNA-bank with a specific congenital heart defect will be approached by their cardiologist at first, to receive permission for contacting them again for participation in this study, if a variation has been found in their DNA .
Patients with a variation who declared to agree with being approached, will be

contacted by mail and asked to participate in this particular study. If participating, DNA-analysis will be repeated for confirmation of the genetic variation. History and family history will be taken and a physical examination will be performed.
Patients* first degree relatives are contacted as soon as the researcher receives a list of relatives willing to participate from the index patient. Relatives are asked about medical history and family history and undergo a physical examination, as well as a cardiologic exam, consisting of ECG and echocardiography. If relevant, previous medical records will be reviewed. Blood will be taken for DNA-analysis.
If the mutation found in the index patient is familial, and/or if the family history is positive for congenital heart defects, other relatives will also be asked to participate in the study.
Data-analysis: DNA-analysis takes place in the laboratories of the departments of Anatomy/Embryology and Clinical Genetics in the AMC. The molecular biologist/geneticist will be blinded for patient data. ECG and echocardiography are performed by a cardiologist with special expertise in congenital heart defects, who will be blinded for the results of the DNA-analysis.
Data will be presented as a description of genetic segregation analysis.

Study burden and risks

Patients will have a history, family history and physical examination taken. Blood will be taken for confirmation of the mutation that was previously found. Relatives will have a history, family history and physical examination taken and they will undergo echocardiography and ECG. As well, blood will be taken for DNA analysis.
To our opinion, these investigations hold no risks for the participants.

Contacts

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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

1) Patients with a congenital heart defect, with a variant in the DNA.
2) First degree relatives of patients with a congenital heart defect with a variant in the DNA.

Exclusion criteria

1) Patients with a congenital heart defect, without a variant in the DNA.

Study design

Design

Study type: Observational invasive

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

Recruitment

NL

Recruitment status:	Pending

Start date (anticipated):	01-08-2007
Enrollment:	160
Type:	Anticipated

Ethics review

Approved WMO

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	NL18161.018.07