

# Molecular genetics of familial heart disease

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<b>Ethical review</b>	Approved WMO
<b>Status</b>	Recruitment stopped
<b>Health condition type</b>	Congenital cardiac disorders
<b>Study type</b>	Observational invasive

## Summary

### ID

NL-OMON37798

### Source

ToetsingOnline

### Brief title

Genetics of familial heart disease

### Condition

- Congenital cardiac disorders
- Cardiac and vascular disorders congenital

### Synonym

congenital heart defect, Congenital heart disease

### Research involving

Human

### Sponsors and support

**Primary sponsor:** Academisch Medisch Centrum

**Source(s) of monetary or material Support:** ICIN;Nederlandse Hartstichting;EU

## Intervention

**Keyword:** Aortic aneurysm, Congenital heart disease, Genetics

## Outcome measures

### Primary outcome

Genetic defects underlying CHD and aortic aneurysms.

### Secondary outcome

Not applicable

## Study description

### Background summary

It is becoming increasingly clear that genetic defects are involved in a significant number of cardiac disorders, such as rhythm disturbances, cardiomyopathies and a subset of congenital heart diseases (CHD). Through identification of genes that are involved in these disorders, insight in the pathophysiology of the disorders and the functions of the mutated genes was gained, and the effects of early treatment on the prognosis of the disorder could be evaluated. However, in the majority of families with multiple affected individuals with CHD or aortic aneurysm no mutation is identified in any of the genes that are currently known to be involved. This implies genetic heterogeneity of these disorders.

In this study we aim to identify genetic causes for several familial cardiac disorders, i.e. (different types of) CHD and aortic aneurysms. This will provide insight in the aetiology and heredity of these diseases. This knowledge can contribute to improved medical care for patients and their relatives, in terms of counselling, screening and treatment.

### Study objective

The aim of this study is to identify genetic defects underlying CHD and aortic aneurysms. This will provide insight in the aetiology and heredity of these diseases.

### Study design

Families with two or more affected patients with CHD and/or an aortic aneurysm will be approached for participation. Participants will undergo the following

investigations: medical history taking, ECG, echocardiogram and cardiac MRI (when indicated). Furthermore, blood will be drawn (20 ml), from which DNA will be isolated (in very young children a mouth-swab or saliva kit can be used instead). In each individual family it will be determined which DNA analysis technique is feasible for identification of involved genes. Techniques that may be used are linkage analysis, Sanger-sequencing, exome sequencing, whole genome sequencing, array CGH and RNA-sequencing.

### **Study burden and risks**

No significant risks are associated with participation in this study. However, there is a possibility that results will be generated that may be of clinical relevance to the participant or his/her relatives. This may involve genetic results related to cardiac disease, as well as coincidental findings not related to the cardiac disorder though clinically relevant. In case of such finding, we will inform the participant in general terms about the finding and its clinical relevance. He or she can decide if he wants to be informed more extensively and to seek treatment. If necessary, participants will be counseled about this finding at the clinical genetics outpatient clinic. If a participant does not want to be informed in general terms about such findings, he or she cannot participate in the study.

## **Contacts**

### **Public**

Academisch Medisch Centrum

Meibergdreef 9  
1105 AZ Amsterdam  
NL

### **Scientific**

Academisch Medisch Centrum

Meibergdreef 9  
1105 AZ Amsterdam  
NL

## **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

Individuals from families with two or more affected persons with congenital heart disease and/or (thoracic) aortic aneurysm are included. Affected family members as well as unaffected family members are included.

### Exclusion criteria

None

## Study design

### Design

**Study type:** Observational invasive

Masking: Open (masking not used)

Control: Uncontrolled

Primary purpose: Basic science

### Recruitment

NL

Recruitment status: Recruitment stopped

Start date (anticipated): 12-06-2012

Enrollment: 300

Type: Actual

## Ethics review

Approved WMO

Date: 12-06-2012

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	NL38157.018.11