Unique epigenetic, transcriptional, metabolomic and proteomic characterization of the heart in cardiomyopathy patients

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Using epigenetic, transcriptional, metabolomic and proteomic approaches in cardiac and skeletal muscle tissue, we want to reveal distinct biological pathways affected in ACM, DCM and PLN r14del cardiomyopathy.

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Cardiac disorders, signs and symptoms NEC
Study type	Observational invasive

Summary

ID

NL-OMON49085

Source
ToetsingOnline

Brief title
DECIPHER-PLN part 2

Condition

- Cardiac disorders, signs and symptoms NEC
- Cardiac and vascular disorders congenital

Synonym
cardiomyopathy, Heart disease

Research involving
Human

Sponsors and support

Primary sponsor: Universitair Medisch Centrum Groningen
Source(s) of monetary or material Support: Astra Zeneca,Bedrijf

Intervention

Keyword: Cardiology, Heart disease

Outcome measures

Primary outcome

Define the biological pathways affected in several types of cardiomyopathies

using a **multi**-omics approach. We will furthermore define how cardiac PLN

expression correlates to skeletal muscle PLN expression to see whether PLN

expression in skeletal muscle can be used as a proxy for PLN expression in the

heart.

Secondary outcome

N/A

Study description

Background summary

The (epi)-genetic background is known to be an important risk factor for the development of arrhythmogenic (ACM) and dilated cardiomyopathy (DCM). Phospholamban (PLN) r14del cardiomyopathy is a heritable disease that bears characteristics of both ACM and DCM and is the most frequently identified genetic mutation in cardiomyopathy patients in the Netherlands.

Study objective

Using epigenetic, transcriptional, metabolomic and proteomic approaches in cardiac and skeletal muscle tissue, we want to reveal distinct biological pathways affected in ACM, DCM and PLN r14del cardiomyopathy.

Study design

Non-therapeutic study, exploring ACM, DCM and PLN r14del cardiomyopathy patients.

Study burden and risks

Cardiac transplantation and LVAD implantation are major procedures, however the collection of a cardiac tissue during this procedure will not add any additional burden or risks to the patient, nor will it extend the procedure. The burden or risks associated to the skeletal muscle biopsy collection are minimal. Participants can experience some pressure or mild cramping, but can resume their daily activities right away and participate in physical activity after 48 hours. Risks include bruising, infection (<1%) and muscle or nerve damage (<1%). This study will not provide a direct benefit to the patients, but will prove beneficial to the research community with potential new insights in ACM, DCM and PLN r14del cardiomyopathy pathogenesis. This study can ultimately contribute to the development of treatment and disease prevention strategies for these patients.

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

• A minimum age of 18.
• Of adequate communication.
• Informed consent is obtained.

Exclusion criteria

• Known allergy for local anaesthetics.

Study design

Design

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	26-11-2020
Enrollment:	40
Type:	Actual

Ethics review

Approved WMO Date:	06-10-2020
Application type:	First submission
Review commission:	METC Universitair Medisch Centrum Groningen (Groningen)

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	NL73976.042.20