

Heart muscle changes in facioscapulohumeral muscular dystrophy type 1 applying cardiac magnetic resonance

Submission date 13/06/2017	Recruitment status No longer recruiting	☐ Prospectively registered
		☐ Protocol
Registration date 25/07/2017	Overall study status Completed	☐ Statistical analysis plan
		☒ Results
Last Edited 08/04/2020	Condition category Nervous System Diseases	☐ Individual participant data

Plain English summary of protocol

Background and study aims

Facioscapulohumeral muscular dystrophy (FSHD) is a genetic disease that causes weakness in the muscles of the limbs, shoulders and face. Patients may suffer from shortness of breath, dizziness and loss of consciousness. In the researchers' patient group heart problems seem to be less common, but problems including sudden death have been reported. It may be that an unknown injury of the heart leads to death or dangerous heart rhythm disturbances. Over the last couple of years cardiac (heart) MRI has become the gold standard method for looking at scars and other changes in the heart muscle (fat, inflammation). The aim of this study is to identify early heart muscle changes in patients with FSHD and their relationship with heart rhythm disturbances.

Who can participate?

Patients age 18 or over with FSHD, and healthy volunteers

What does the study involve?

All participants undergo a full examination by cardiologists including a cardiac MRI scan with both standard and new techniques to look for heart tissue injuries. Heart rhythm disturbances are assessed using an electrocardiogram (ECG).

What are possible benefits or risks of participating?

Participants receive a written report for their records including the basic assessment. The detection of early heart changes with MRI may help to improve treatment in the future. The MRI scan is prolonged by only about ten minutes. From a participant's perspective no other burden is created by this study.

Where is the study run from?

Charité University Medicine Berlin (Germany)

When is the study starting and how long is it expected to run for?

September 2015 to May 2017

Who is funding the study?
Bayer Health Medical Care

Who is the main contact?
Prof. Jeanette Schulz-Menger

Contact information

Type(s)
Scientific

Contact name
Prof Jeanette Schulz-Menger

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Additional identifiers

Protocol serial number
FSHD-CMR (internal study code)

Study information

Scientific Title
Cardiac involvement in facioscapulohumeral muscular dystrophy type 1 patients with preserved ejection fraction – assessment by cardiovascular magnetic resonance

Study objectives
A prospective diagnostic trial to evaluate the efficacy of gadobutrol-enhanced cardiovascular magnetic resonance (CMR) at identifying myocardial tissue injury in facioscapulohumeral muscular dystrophy type 1 patients with preserved left ventricular function. This proof of concept trial is intended to extend the indications for CMR.

Ethics approval required
Old ethics approval format

Ethics approval(s)
Ethics board of the Charité University Medicine Berlin Campus Mitte, 03/09/2015, ref: EA1/169/15

Primary study design

Observational

Study design

It is a prospective diagnostic trial to evaluate the efficacy of gadobutrol-enhanced CMR to identify myocardial tissue injury in Facioscapulohumeral Muscular Dystrophy Type 1. This proof of concept trial is intended to extend the indications for CMR.

Study type(s)

Diagnostic

Health condition(s) or problem(s) studied

Facioscapulohumeral muscular dystrophy type 1

Interventions

After inclusion, the study participants undergo a CMR. Furthermore, an age- and gender-matched healthy control group is identified. They receive the same CMR protocol.

CMR is applied at a 1.5 T Scanner (MAGNETOM AvantoFit®, Siemens Healthcare, Erlangen, Germany) using a 32 channel surface coil. Cine imaging is performed applying state of the art steady state precession sequences to determine the global cardiac performance. For myocardial tissue differentiation, parametric T1 and T2 mapping, fat/water separated imaging and focal fibrosis imaging (Late Gadolinium Enhancement, LGE) are performed.

Intervention Type

Other

Primary outcome(s)

Myocardial tissue injuries, detected using magnetic resonance imaging visually (qualitative) and quantitative including the presence and extent of lesions like fat and scar, measured at a single timepoint

Key secondary outcome(s)

Heart rhythm disturbances, assessed using ECG and ECG monitoring at a single timepoint

Completion date

15/12/2018

Eligibility

Key inclusion criteria

1. Genetically confirmed diagnosis of FSHD1
2. Age ≥ 18 years (no upper limit)
3. Age- and gender-matched healthy control group

Participant type(s)

Mixed

Healthy volunteers allowed

No

Age group

Adult

Lower age limit

18 Years

Sex

All

Total final enrolment

52

Key exclusion criteria

Known vascular, cardiac diseases (e.g. coronary artery disease, significant valvular disease, myocarditis), malign diseases or known contraindications for CMR or Gadolinium- based contrast-media

Date of first enrolment

03/09/2015

Date of final enrolment

31/12/2016

Locations**Countries of recruitment**

Germany

Study participating centre**Working Group on Cardiovascular Magnetic Resonance**

Experimental and Clinical Research Center, a joint cooperation between the Charité University Medicine Berlin and the Max-Delbrueck Center for Molecular Medicine, and HELIOS Klinikum Berlin Buch, Department of Cardiology and Nephrology

Lindenberger Weg 80

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13125

Sponsor information**Organisation**

Charité University Medicine Berlin

ROR

<https://ror.org/001w7jn25>

Funder(s)

Funder type

Industry

Funder Name

Bayer Health Medical Care

Results and Publications

Individual participant data (IPD) sharing plan

The datasets generated during and/or analysed during the current study are not expected to be made available due to data protection laws in Germany. However, upon request methodology and dataset structure can be shared.

IPD sharing plan summary

Not expected to be made available

Study outputs

Output type	Details	Date created	Date added	Peer reviewed?	Patient-facing?
Results article	results	29/04/2019		Yes	No