One year follow-up in FSHD: biomarkers and outcome measures on the road to therapy

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

Summary

ID

NL-OMON49566

Source
ToetsingOnline

Brief title
FSHD-FOCUS 2.1

Condition

- Musculoskeletal and connective tissue disorders congenital

Synonym
FSHD (facioscapulohumeral muscular dystrophy), Landouzy

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum
Source(s) of monetary or material Support: FSHD Stichting;Prinses Beatrix fonds

Intervention

Keyword: Follow-up, FSHD (facioscapulohumeral dystrophy), Muscle Ultrasound, Phenotype

Outcome measures

Primary outcome

Muscle ultrasound on facial, trunk and extremity muscles will be repeated to assess structural muscle changes over one year. A subgroup of 30 patients will undergo muscle ultrasound every three months over the span of one year to assess the evolvement of ultrasound abnormalities. Raw muscle echo intensities will be converted to z-scores*that is, the number of standard deviations from the mean score for gender, age, and weight using previously established reference values.

Secondary outcome

Changes in functional status will be assessed by the Motor Function Measure, 6-minute walk test, clinical severity scores in combination with facial weakness and muscle strength (MRC-scale), and the newly developed anti-gravity tests. The responsiveness of existing and recently developed patient-reported outcome measures (SIP68, Rasch-built FSHD-RODS, FSHD-specific questionnaire on facial weakness) will be evaluated. Also in 50 participants a muscle MRI will be performed, to assess the fatfraction in the muscles of both legs. Furthermore, blood samples will be taken immunological studies and for storage in the Radboudumc biobank for future research.

Study description

Background summary

Facioscapulohumeral muscular dystrophy (FSHD) is one of the most common adult muscular dystrophies with a prevalence of 12/100.000. It is characterized by progressive muscle weakness and wasting of muscle of the face, extremities and trunk, which leads to loss of function and major disability. In 2014 we characterized a large cohort of over 200 FSHD patients both clinically and genetically in great detail in the FSHD-FOCUS study. A five year follow-up study is currently ongoing that will provide extensive data on the natural history of the disease through various clinical outcome measures and muscle imaging biomarkers. However, five year follow-up is too long to assess the responsiveness of these outcomes over a reasonable timeframe for a clinical trial. Additionally, after the baseline measurements of this cohort in 2014 new highly promising patient-reported and functional outcome measures and imaging biomarkers have been developed on which data baseline data is collected in the current follow-up study.

Study objective

The goal of this study is to perform an additional follow-up visit on the FSHD-FOCUS cohort one year after the five year follow-up visit to assess the responsiveness over one and a half year, a reasonable timeframe for a clinical trial, of highly promising clinical outcome measures identified in the FSHD-FOCUS study and muscle ultrasound as an imaging biomarker.

Study design

Longitudinal, observational study

Study burden and risks

Participants will be asked for a single or multiple visit(s) to the outpatient clinic at the department of neurology. Their medical history will be taken and they will undergo a clinical examination. Several questionnaires can be completed at home through on online system (Castor). A muscle ultrasound and pssibly a muscle MRI will be performed and blood samples will be collected. We classify the risk of this study as negligible.

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

100 genetically confirmed FSHD patients that participated in the FSHD-FOCUS 2 study (CMO 2014-121) who underwent muscle ultrasound as part of the study.

Exclusion criteria

1. Incapacitated persons will not be included in this study.
2. Patients who are unable to visit the Radboudumc.

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):	12-10-2020
Enrollment:	100
Type:	Actual

Ethics review

Approved WMO Date:	05-08-2020
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	15-09-2020
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	17-02-2021
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)

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	NL73343.091.20