FSHD-FOCUS 2: Five years follow up in FSHD

Published: 04-04-2019
Last updated: 12-04-2024

Obtain longterm follow-up data in a large and clinically and genetically well-characterized cohort and thereby contributing to knowledge about the disease course and clinical trial preparedness for FSHD research.

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

Summary

ID

NL-OMON48308

Source
ToetsingOnline

Brief title
FSHD-FOCUS 2

Condition

- Musculoskeletal and connective tissue disorders congenital

Synonym
FSHD (facioscapulohumeral muscular dystrophy), Landouzy Dejerine disease

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum
Source(s) of monetary or material Support: Prinses Beatrix Fonds

Intervention

Keyword: (epi)genetics, follow up, FSHD (facioscapulohumeral muscular dystrophy), imaging techniques, phenotype

Outcome measures

Primary outcome

Primary outcomes will be a description of the natural history of FSHD, using the patient reported (questionnaires), clinical (muscle strength, functional assessment, clinical severity scores) and radiological outcome measures (MRI and ultrasound).

Secondary outcome

Secondary outcomes are the sensitivity to change of commonly used clinical outcome measures, as were used in the baseline FSHD-study and the validation of recently developed outcome measures. Furthermore we assess the evolvement of muscle MRI abnormalities in terms of fatty infiltration and TIRM positivity and the evolvement of muscle ultrasound abnormalities. We will also add bloodsamples to look for epigenetic factors of influence for FSHD and to our Radboudumc Biobank for future research.

Study description

Background summary

Facioscapulohumeral muscular dystrophy (FSHD) is an autosomal dominant inherited progressive muscular dystrophy, characterized by asymmetrical weakness and wasting of facial, shoulder girdle and upper arm muscles, followed by weakness of the muscles of the trunk and the lower extremities. Over the last few years, our knowledge on the pathogenic mechanism in FSHD has expanded. However, we have not yet explained the variability in onset, disease course and penetrance, nor the asymmetric nature of the disease. Our hypothesis is, that

other (epi)genetic and environmental or lifestyle factors must be involved in this disease. It is important to identify these factors, as some of them may be considered as *natural moderators* of the disease and may contribute to development of new treatment strategies. If such a treatment becomes available, well controlled clinical trials will be warranted. Conditions for clinical trial readiness should therefore be met, such as validating measures of activity impairment and patient reported outcome measures, identification of serum biomarkers and longitudinal muscle MRI data with a correlation of imaging to muscle function. This study will provide all this for a cohort of 200 FSHD patients with a follow-up of five years.

Study objective

Obtain longterm follow-up data in a large and clinically and genetically well-characterized cohort and thereby contributing to knowledge about the disease course and clinical trial preparedness for FSHD research.

Study design

Cross-sectional, observational study.

Study burden and risks

Participants will visit the outpatient clinic at the department of neurology. Their medical history will be taken, they will undergo a clinical examination and they will fill out questionnaires online (at home). Blood samples will be collected for storage of blood in a biobank for future research and to check for more epigenetic factors of interest. During the same visit a magnetic resonance imaging (MRI) of muscles of both legs and possibly leg muscle ultrasound will be performed. Complications of venapunctions are very uncommon and include hematoma, which will resolve itself.

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

All 203 genetically confirmed FSHD patients that participated in the FSHD-FOCUS study (CMO 2014-121) and were (at the time of the study or before) informed about the genetic confirmation of FSHD.

Exclusion criteria

Incapacitated persons will not be included in this study. FSHD patients that participated in the FSHD-FOCUS study but were not informed about the genetic testing results for FSHD will not be included in this study.
Persons with contra-indications for MRI-scan are excluded for that one procedure, but can still be included in the study. Contra-indications for MRI-scan include metallic implants (vascular clips, foreign bodies like metallic splinters in the eye, coronary and peripheral artery stents, prosthetic heart valves, pacemakers and ICD*s, cochlear implants, breast tissue expanders and some other electronic implants or devices), renal insufficiency, previous allergic reaction to contrast fluids and known claustrophobia.

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):	09-05-2019
Enrollment:	200
Type:	Actual

Ethics review

Approved WMO Date:	04-04-2019
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	11-06-2019
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	NL68245.091.18