Inflammation in facioscapulohumeral muscular dystrophy

Published: 01-08-2016
Last updated: 16-04-2024

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Ethical review	Approved WMO
Status	Recruitment stopped
Health condition type	Muscle disorders
Study type	Observational non invasive

Summary

ID

NL-OMON43090

Source
ToetsingOnline

Brief title
Inflammation in FSHD

Condition

- Muscle disorders

Synonym
muscular dystrophy

Research involving
Human

Sponsors and support

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

Intervention

Keyword: Cytokines, DUX4, FSHD, Inflammation

Outcome measures

Primary outcome

We expect to better understand if and which role inflammation plays in the disease pathogenesis by evaluating the following outcomes: percentage and features of PBMC, cytokine profiling and autoantibodies inquiry both in FSHD1 patients and in healthy controls. Moreover, disease severity of FSHD patients will be assessed using a clinical severity score (Ricci-score).

Secondary outcome

not applicable

Study description

Background summary

Facioscapulohumeral dystrophy (FSHD) is one of the most prevalent inherited myopathies and, although the role of inflammation in its pathogenesis is strongly suggested, evidence in literature remains equivocal. Recent studies describe muscle inflammatory infiltrates mainly composed by CD8+ T cells in muscles showing hyperintensity features on T2-weighted short tau inversion recovery magnetic resonance imaging (T2-STIR-MRI) sequences. A further understanding of the inflammatory mechanism involved is crucial, especially as it may open up new possibilities for pharmaceutical interventions, anti-inflammatory drugs being available.

Study objective

We aim at investigating the role played by the innate and adaptive immune system and orienting a proper treatment. We will evaluate the percentage and features of circulating activated immune cells and cytokine production. As a second step, we will investigate the reactivity of FSHD patient sera with human skeletal muscle protein extracts to detect autoantibodies.

Study design

We will perform a case-control study to evaluate peripheral blood mononuclear cells (PBMC), cytokine profiling, autoantibodies finding.

Study burden and risks

A total of 10 subjects will be included and invited for one visit at the outpatient clinic of the department of neurology at the Radboudumc. During the visit, medical and family history will be collected and a clinical examination will be performed. A total volume of approximately 50 mL will be taken via venipuncture from each participant: 4 blood samples of 10mL EDTA and 1 sample of 10 mL serum. The blood sampling collection has numerous advantages: affordable, rapid, easy to obtain, availability of several body sites. Furthermore, there are minimal associated risks: bleeding, a slight risk of infection, fainting or feeling light-headed. These risks can be controlled and avoided by antiseptic measures and expert personnel. This study has a considerable relevance, as inflammation is a potential key to new treatment strategies. Therefore, participation of patients and healthy volunteers is crucial to gain insight into this debilitating disease.

Contacts

Public
Radboud Universitair Medisch Centrum

Reinier Postlaan 4
Nijmegen 6525 GC
NL
Scientific
Radboud Universitair Medisch Centrum

Reinier Postlaan 4
Nijmegen 6525 GC
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adults (18-64 years)
Elderly (65 years and older)

Inclusion criteria

-18-65 year old
-Genetically confirmed FSHD1

Exclusion criteria

-Age <18 or > 65 years old
-Diabetes mellitus
-Chronic obstructive pulmonary disease
-Current malignancy
-Previous treatment with chemotherapy and/or radiation therapy
-Use of corticosteroids during more than two weeks in the past 5 years
-Use of statins in the past year

Study design

Design

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

Recruitment

NL
Recruitment status:	Recruitment stopped
Start date (anticipated):	20-07-2017

Enrollment:	10
Type:	Actual

Ethics review

Approved WMO Date:	01-08-2016
Application type:	First submission
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	NL57308.091.16