Exploring the presence and characteristics of physical fatigability in children and adolescents with facioscapulohumeral dystrophy (FSHD)

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The primary objective is to determine if physical fatigability is present in infantile and young adult patients with FSHD.The secondary objectives are to determine if there is an association between fatigability ando Disease severityo Muscle...

Ethical review	Approved WMO
Status	Completed
Health condition type	Musculoskeletal and connective tissue disorders congenital
Study type	Interventional

Summary

ID

NL-OMON56024

Source
ToetsingOnline

Brief title
iFocus fatigability

Condition

- Musculoskeletal and connective tissue disorders congenital
- Musculoskeletal and connective tissue deformities (incl intervertebral disc disorders)
- Neuromuscular disorders

Synonym
FSHD, Landouzy-Dejerine

Research involving
Human

Sponsors and support

Primary sponsor: Radboud Universitair Medisch Centrum
Source(s) of monetary or material Support: Spieren voor spieren;FSHD stichting.

Intervention

Keyword: Childhood onset, Facioscapulohumeral dystrophy, Fatigability, Neuromuscular disease

Outcome measures

Primary outcome

The main study parameter will be whether fatigability is present in children en young adults with FSHD. Therefore, we propose to use the endurance shuttle box and block test (ESBBT) and the endurance shuttle walk test (ESWT).

In short, we will instruct subjects to repeatedly move 10 blocks over a partition (ESBBT) or walk 10 meters (ESWT) at 75% of their previously determined, individualized maximum speed. The test will be ended when the subject is not able to keep up the pre-set pace during two consecutive shuttles or when the maximum duration of 20 minutes is reached (test completion). Further, we will describe these two tests in more detail.

For each performed test we will document two outcomes:

- Drop out (yes/no): the inability to endure the maximum duration of 20 minutes
- Time to limitation (s)

Increased fatigability is defined as the presence of drop out on the ESBBT or ESWT.

The time to limitation is used as a measure of severity of fatigability

Secondary outcome

Secondary study parameters are:

- Disease severity (muscle strength, FSHD score, Ricci score, Motor function measure)

- Muscle structure (muscle ultrasonography)

- Quality of life (questionnaires)

- Pain (questionnaire)

- Fatigue (questionnaire, observed signs of fatigue)

Other study parameters are:

- Genotype characteristics, if present (D4Z4 repeat length)

- Demographic and clinical characteristics (age, sex, length and weight)

- Identification of environmental disease modifying factors (physical activity, co-morbidity and medication use)

Study description

Background summary

Facioscapulohumeral dystrophy (FSHD) is one of the most common hereditary muscular dystrophies in the Netherlands.
The age of symptom onset is mostly in the second or third decade, but in approximately 20% of all the patients symptoms start in childhood.
This subgroup is historically associated with a faster disease progression, severe weakness and more systemic complications (epilepsia, hearing problems, mental retardation and cardial arrhytmias).

Previously, we studies since 2016 the natural history of FSHD in childhood.
This showed that 70% of children suffers from fatigue. This high prevalence is

a reason for concern, especially when we take into account the correlation between age and fatigue severity, with increasing age leading to a further deterioration of quality of life. In this way, fatigue could have a substantial effect on participation in school and sports and on their professional development. Knowlede of fatigue and its causes is essential in order to improve the management of childhood FSHD.

In children with spinal muscular atrophy (SMA) recent research showed that fatigability (the inability to continue exercise at the same intensity wit a resultant deterioration in performance) was present in 54-73% of patients. This was measured by some recently developed and validated shuttle test.
In our earlier observations, the functional exercise capacity of children with FSHD measured by the 6 minute walk test was diminished, and the average gait velocity of the last minute decreased compared to the first minute. An important limitation of this test is that the result is affected by the effort of the child. However, in clinical practice we found that children often complain about fatigue in the ocntext of long walks or during a day to a theme park.

By combining this, we suspect that muscle fatigability might be involved in childhood FSHD.

Study objective

The primary objective is to determine if physical fatigability is present in infantile and young adult patients with FSHD.

The secondary objectives are to determine if there is an association between fatigability and
o Disease severity
o Muscle strength
o Motor function
o Perceived fatigue
o Perceived pain
o Quality of life

Study design

This study is a cross-sectional, observational study.

Intervention

No intervention

Study burden and risks

This study is an observational study on a childhood disease and therefore must include minors as subjects. The burden associated with participation is minimized and the risk of participation is negligible. There are no invasive procedures, the used test are not painful. If we observe any inconveniences during participation, we will stop the test. .
Participants visit the hospital for half a day. We d

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age
Adolescents (12-15 years)
Adolescents (16-17 years)
Adults (18-64 years)
Children (2-11 years)

Inclusion criteria

- patients with genetically proven FSHD
- Disease onset in childhood (<18 years)

-Current age 8-25 years
- The ability to follow test instructions

Exclusion criteria

- Loss of ambulation (the inability to perform the endurance shuttle walk test)
- A history of myasthenia gravis or another neuromuscular disorder known to cause fatigability or affect the neuromuscular junction function
- The use of drugs that change the neuromuscular transmission

Study design

Design

Study type: Interventional

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Basic science

Recruitment

NL

Recruitment status:	Completed
Start date (anticipated):	27-03-2024
Enrollment:	20
Type:	Actual

Ethics review

Approved WMO Date:	07-12-2023
Application type:	First submission
Review commission:	CCMO: Centrale Commissie Mensgebonden Onderzoek (Den Haag)

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	NL80728.000.23