A natural history study on infantile facioscapulohumeral muscular dystrophy

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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-OMON42518

Source
ToetsingOnline

Brief title
iFSHD

Condition

- Musculoskeletal and connective tissue disorders congenital
- Neuromuscular disorders

Synonym
FSHD, Landouzy-Dejerine

Research involving
Human

Sponsors and support

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

Intervention

Keyword: childhood onset, facioscapulohumeral dystrophy, natural history study, neuromuscular disease

Outcome measures

Primary outcome

The main study parameter will be the description of the phenotype of childhood-onset FSHD. Therefore we propose extensive qualitative and quantitative measurements specified by age and use a modified format of the International Classification of Functioning, Disability and Health criteria (ICF-Y). ICF is the WHO framework for measuring health and disability at both individual and population levels. The domaines are body structure, body function, activities and participation and environmental factors.

Secondary outcome

Secondary outcome measures are:

- Prevalance and incidence estimations of infantile FSHD

- (epi)genetical changes

- Disease modifying factors

- extensive quantitative and qualitative genotype and fenotype characterisation.

Study description

Background summary

Facioscapulohumeral muscular dystrophy (FSHD) is an autosomal dominant inherited muscular dystrophy with a characteristic distribution of weakness. In >95% of the patients, FSHD is caused by a mutation on chromosome 4q35.

Although the genetic basis is very homogenous, there is remarkable inter- and intrafamilial variability ranging from wheelchair bound individuals to asymptomatic carriers. The most severely affected subgroup has a childhood onset (first symptom before the age of 5) and extramuscular symptoms such as epilepsy, hearing loss and mental retardation occur. There is very limited knowledge on this severely affected subgroup, accounting for 2-15% of the total patient population. Natural history, disease and longitudinal data on infantile FSHD are essential for the design of best standard of care, therapeutic trials and in assessment of genetic heterogeneity.

Study objective

The primary objective is to describe the clinical characteristics of infantile FSHD.
The secondary objectives are:
- To determine the incidence and prevalence of infantile FSHD
- To describe the genetic characteristics of infantile FSHD.
- To identify (epi)genetic and environmental disease modifying factors that contribute to the variable clinical phenotype of FSHD.
- To obtain a well-documented cohort of children with FSHD-patients to be recruited for future clinical trials.

Study design

Explorative, cross-sectional, observational study.

Study burden and risks

Participants will be asked for a visit to the outpatient clinic at the department of pediatric neurology. Their medical history will be taken, they will undergo a clinical examination and their parents will be asked to fill out questionnaires. Medical charts will be reviewed on FSHD history. Blood samples will be collected for DNA- and RNA-analysis and gene expression profiling. Also, participants will undergo a hearing test, muscle ultrasonogaphy, fundoscopy and ECG-recording. We classify the risk of this study as negligible. The investment for patients and their family is time.

Contacts

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Trial sites

Listed location countries

Netherlands

Eligibility criteria

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

Inclusion criteria

- Patients aged 0-17 with genetically proven FSHD1 (mutation on chromosome 4q35, leading to a reduced number of less than 10 D4Z4 subunits) or FSHD2 (SMCHD1 gene mutation on chromosome 18)
- Patients aged 0-17 with a clinical suspicion of FSHD. The clinical suspicion is defined as: based on the opinion of the treating medical specialist (paediatricians or neurologists) or children with delayed motor development with clinical weakness of the facial or upper-arm muscle.

Exclusion criteria

- Patients not able to visit the outpatient clinic at the Radboudumc
- If not genetically confirmed: clinical suspicion not confirmed by a specialized neuromuscular child neurologist.

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):	11-11-2015
Enrollment:	40
Type:	Actual

Ethics review

Approved WMO Date:	03-11-2015
Application type:	First submission
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	02-06-2016
Application type:	Amendment
Review commission:	CMO regio Arnhem-Nijmegen (Nijmegen)
Approved WMO Date:	23-07-2018
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	NL53213.091.15