Epidemiology, natural course and registration of dystrophinopathies in the Netherlands

Published: 25-04-2008
Last updated: 07-05-2024

1. Description of the epidemology of dystrophinopathies in the Netherlands2. Description of the natural course of dystrophinopathies in the Netherlands and the influence of medical development on it.3. The initiation of a database for patients...

Ethical review	Approved WMO
Status	Recruiting
Health condition type	Neuromuscular disorders
Study type	Observational non invasive

Summary

ID

NL-OMON32292

Source
ToetsingOnline

Brief title
Dystrophinopathies in the Netherlands

Condition

- Neuromuscular disorders

Synonym
Duchenne/Becker, muscular dystrophy

Research involving
Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum
Source(s) of monetary or material Support: TREAT-NMD (met subsidie van Europese Unie)

Intervention

Keyword: Becker muscular dystrophy, database, Duchenne muscular dystrophy, Epidemiology

Outcome measures

Primary outcome

- registration in national and international database

- year of birth

- age when able to walk independantly for the first time

- age at diagnosis Duchenne/Becker

- mutation in dystrophin gene

- level of education

- age at becoming wheelchair dependant

- scoliosis and possible surgery

- cardiomyopathy

- age at which time home ventilation was initiated, if applicable

- age at death

- use of corticosteroids

Secondary outcome

none

Study description

Background summary

Duchenne Muscular Dystrophy is an X-chromosomal inherited condition, caused by a mutation in the dystrophin gene. This protein has an important function in stabilisation of the muscle membrane. In Duchenne patients this protein is

absent, leading to contraction-induced muscle damage. Clinically this results in progressive muscle weakness, leading, without treatment, to a wheelchair dependancy at approximately the age of ten and death due to respiratory failure or a cardiomyopathy around the age of twenty.
With Becker Muscular Dystrophy there is also a mutation in the dystrophin gene. However, in this condition there is dystrophin production, nut the dystrophin is shorter than normal and only partially functional. As a result, the clinical picture is milder than with Duchenne, ranging from wheelchair dependancy around 16 years of age to patients with a normal life expectancy.
The preceiding decades there have been several developments in the care for Duchenne patients. Besides general improvements (vaccination, antibiotics) the usage of corticosteroids and the start of home ventilation for Duchenne patients are important developments that contribute to a longer mobility and a better survival respectively.
Currently, there are eleven clinical phase I/II trials being planned, started or recently ended for eleven medicines. Of these, the exon-skipping technology is one of the most promising. Many of the possible therapies are mutation specific, which makes it important to have an overview of which patient has which mutation and is therefor eligable for a specific therapy or can participate in a specific trial.

Study objective

1. Description of the epidemology of dystrophinopathies in the Netherlands
2. Description of the natural course of dystrophinopathies in the Netherlands and the influence of medical development on it.
3. The initiation of a database for patients willing to participate in future research and/or trials.

Study design

Retrospective, observational cohortstudy. Inclusion of patients occurs through the clinical-genetics database, internet, treating physicians, patient organisations and the home ventilation centers. The expected duration of this research project is 2 years.
After receiving the informed consent form, patients will receive a questionaire and are asked to give permission to enquire about their medical records from treating physicians. Following the results from the questionaire and the medical record there will be a telephonic contact to discuss any remaining questions/unclarities. The data will be stored in a national database. Besides this, patients are asked to give permission to anonymously store their data in the international TREAT-NMD database. By registration in these databases a patient can be contacted in future to participate in possible (therapeutic) research.

Study burden and risks

none

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)
Elderly (65 years and older)

Inclusion criteria

Progressive muscle weakness (from childhood)
Confirmation of dystrophinopathy diagnosis by a mutation in the dystrophin gene or the absence of dystrophin in a muscular biopt

Exclusion criteria

no informed consent

Study design

Design

Study type: Observational non invasive

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

Recruitment

NL

Recruitment status:	Recruiting
Start date (anticipated):	01-06-2008
Enrollment:	800
Type:	Actual

Ethics review

Approved WMO

Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden) metc-ldd@lumc.nl

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	NL21411.058.08