Optimal care for Duchenne and Becker muscular dystrophy: Learning, behavior and careful care

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1. Inventory of the current care of boys/men with Duchenne Muscular Dystrophy in the Netherlands: To establish the extent of implementation of the international guidelines and its updates in the Netherlands and to explore the relationship between...

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

Summary

ID

NL-OMON46818

Source
ToetsingOnline

Brief title
Optimal care for Dystrophinopathies

Condition

- Muscle disorders
- Cognitive and attention disorders and disturbances

Synonym
DMD&BMD, Duchenne and Becker muscular dystrophy, Dystrophinopathy

Research involving
Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum
Source(s) of monetary or material Support: Spieren voor Spieren

Intervention

Keyword: Becker, Care, Duchenne, Questionnaires

Outcome measures

Primary outcome

- Percentage in which the international guideline of 2010 is followed; the diagnosis, hereditary advice, the frequency of medical examinations and rehabilitation treatments

- Inventory of the suggestions made by patients themselves, their parents and/or their caregivers to improve care

- After implementation of the updated guideline: The percentages in which this guideline is followed with regard to, among other things, the diagnosis, hereditary advice, the frequency of performing medical examinations and rehabilitation treatments

- The prevalence (%) of learning problems (automating reading and arithmetic, working memory and executive thinking functions) in a prospective longitudinal design

- The prevalence (%) of behavioral problems (ADHD - OCD - ASS) in a prospective longitudinal design

- The prevalence (%) of the degree of dysfunctional psychosocial adaptation in a prospective longitudinal design

- Relationship between learning problems, longitudinally assessed (automating reading and arithmetic, working memory and executive thinking functions), the genotype (Dp140 expression) and type of muscular dystrophy (Duchenne or Becker). Covariation will be checked as well: personality (PLOT questionnaire).

- Relationship between longitudinally measured behavioral problems (ADHD, OCD, ASD), degree of psychosocial adjustment, the genotype (Dp140 expression) and type of muscular dystrophy (Duchenne or Becker). Covariation will be checked as well: personality (PLOT outcome).

Secondary outcome

not applicable

Study description

Background summary

Duchenne muscular dystrophy (DMD) is an X-linked hereditary progressive muscle disorder. In DMD, the dystrophin protein is absent, leading to contraction-induced muscle damage. On average, non-treated DMD patients become wheelchair-bound at the age of 10 and die at their twenties due to respiratory failure or cardiomyopathy. In Becker muscular dystrophy (BMD) the dystrophin mutation leads to a shorten, partially functional dystrophin protein. The clinical presentation of BMD is therefore milder than DMD, ranging from patients becoming wheelchair around the age of 16 and die at late-middle age to patients who remain ambulant having normal life-span. Unfortunately, at date no therapy for DMD and BMD is present.
The last decades, several developments of care consideration for DMD patients have been taken place. In addition to general improvements, like vaccination, antibiotics, the use of corticosteroids and the start of home ventilation have been important developments that have contributed to a longer mobility and survival for boys with Duchenne. Since 2010, international multidisciplinary guidelines have been made available (and an update was published in 2018), which describes the minimum care for boys with DMD. However, a recent study shows that the healthcare in England, Germany, Italy and the USA partly does not comply with these international guideline. The extent of compliance to the care guidelines in the Netherlands and the health status of boys and men with DMD is unknown. Potentially, lower health status influences the daily functioning. Therefore, it is important to optimize care and health.
In addition to the physical problems, boys/men with DMD and BMD also experience learning and behavioral disorders, such as dyslexia, autism spectrum disorders (ASD), obsessive compulsive disorder (OCD) and attention deficit disorder (ADHD). Recent studies have shown that these disorders may be caused by no and/or reduced presence of dystrophin in the brain, such as the Dp140 dystrophin isoform. Behavior and learning disorders can have important

consequences for the everyday functioning of these boys and men. With improved care, longer survival rates and increasing future perspectives, it is important that behavioral and learning disorders are systematically diagnosed and treated in these boys/men with DMD and BMD. Because longitudinal research in this patient population is lacking, the course of these learning and behavioral disorders over time is (also) not yet clear.

Study objective

1. Inventory of the current care of boys/men with Duchenne Muscular Dystrophy in the Netherlands: To establish the extent of implementation of the international guidelines and its updates in the Netherlands and to explore the relationship between given care, health status, social participation and quality of life.

2. Prevalence of learning and behavioral disorders from a longitudinal perspective in relation to type of muscular dystrophy (Duchenne or Becker) and in relation to the genotype (presence of Dp140 expression, and possible other mutations).

Study design

A descriptive longitudinal study in patients with Duchenne or Becker muscular dystrophy.
Digital questionnaires will be used to make an inventory of the extent of implementation of the international guideline and its updates in the Netherlands and the prevalence of specific behavioral and learning problems, the degree of psychosocial adjustment, personality aspects and correlation with the genetic defect.

Study burden and risks

If the patient and/or the parent (s)/ carer giver (s) (when boys <18 years old) provide consent, they will receive both in year one and year three digital questionnaires. Completing the questionnaires will take a time investment of approximately 40-60 minutes for both the parent as well as fior the patient. Based on the results of the questionnaire, telephone contact can be made to discuss any questions/uncertainties.

A direct therapeutic benefit is not to be expected.
No invasive procedures are performed during the study.

This research will provide insight
1. The extent of compliance to the care guidelines in the Netherlands
2. The prevelance of bahavior and learning problems in boys/men with Duchenne and Becker muscular dystrophy.

This study will provide insight/ an overview of the current care of patients with Duchenne and Becker muscle dystrophy. With these results care can be improved in the Netherlands

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
- Confirmed diagnosis of dystrophinopathy: defined mutation in the dystrophin gene and / or either reduced or lack of dystrophin levels in a muscle biopsy
- Age > 4 years

- Dutch as mother tongue

Exclusion criteria

- No informed consent

Study design

Design

Study type: Observational non invasive

Masking:	Open (masking not used)
Control:	Uncontrolled
Primary purpose:	Health services research

Recruitment

NL

Recruitment status:	Pending
Start date (anticipated):	01-10-2018
Enrollment:	131
Type:	Anticipated

Ethics review

Approved WMO

Date:	11-10-2018
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	NL65159.058.18