

Brain Imaging and Cognition in Adults living with Duchenne Muscular Dystrophy

Published: 21-10-2021

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Objective: we aim to achieve a better understanding of the effect of age on NPE and MRI changes in the adult DMD patients and hypothesize that the cerebral growth curves have an altered trajectory.

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

Summary

ID

NL-OMON52255

Source

ToetsingOnline

Brief title

Adult DMD Brain research

Condition

- Muscle disorders

Synonym

Duchenne Muscular Dystrophy / Duchenne

Research involving

Human

Sponsors and support

Primary sponsor: Leids Universitair Medisch Centrum

Source(s) of monetary or material Support: Grant entitled 'Brain Imaging and Cognition in adults living with Duchenne muscular dystrophy' from the Duchenne Parent Project The Netherlands

Intervention

Keyword: Brain, Cognition, Duchenne, Neuromuscular

Outcome measures

Primary outcome

Main study parameters/endpoints: Presence or absence or significant differences in brain volumes structural connectivity, cerebral perfusion and functional connectivity between patients and healthy controls with respect to age, after applying the appropriate statistics per measure. Neurocognitive parameters and behavioural functioning will be assessed to describe the cohorts.

Secondary outcome

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Study description

Background summary

Rationale: Duchenne muscular dystrophy (DMD) is a neuromuscular disorder caused by mutations in the dystrophin gene. Dystrophin is not only expressed in muscle, but also in the brain. About 40% of DMD patients show learning disabilities, especially problems with automatization, working memory and reading. We have previously shown altered total brain and grey matter volume, white matter microstructure and cerebral perfusion in boys with DMD compared to age-matched healthy controls using quantitative magnetic resonance imaging (MRI). We also found lower scores on neuropsychological examination (NPE). Preliminary data show that brain volume decline with age seems to be faster in DMD than in controls suggesting a neurodegenerative process. The cognitive impairment in DMD is thought to be non-progressive; however, the relationship between age, intellectual functioning and MRI-visible changes is still a matter of debate.

Study objective

Objective: we aim to achieve a better understanding of the effect of age on NPE and MRI changes in the adult DMD patients and hypothesize that the cerebral

growth curves have an altered trajectory.

Study design

Study design: an observational case control study in which we will apply the same MRI and NPE investigations used in our previous study in young DMD patients (mean age 13.4 years) (P09.121) to an older group of DMD patients (>18 years).

Study burden and risks

Nature and extent of the burden and risks associated with participation, benefit and group relatedness: The participants for both MRI and NPE are required to come to the LUMC once for half a day (approximately 4 hours). They will undergo an MRI of the brain and neuropsychological testing. These adult patients with DMD are most likely non-ambulant and will require a hoist to be transferred onto the MRI bed. They may also have contractures that prohibit them from stretching out their legs on the MRI bed. We have extensive experience and MRI accessories (pillows, soft bands etc.) to optimize their comfort and safety. The participants with MRI contraindications who only take part in the NPE can do so from home via either a live home visit from the researcher or online consultation which will take approximately 2 hours. The participants have no personal benefits to taking part, but the data will aid our understanding of the brain involvement in DMD and help improve clinical care.

Contacts

Public

Leids Universitair Medisch Centrum

Albinusdreef 2
Leiden 2333ZA
NL

Scientific

Leids Universitair Medisch Centrum

Albinusdreef 2
Leiden 2333ZA
NL

Trial sites

Listed location countries

Netherlands

Eligibility criteria

Age

Adults (18-64 years)

Inclusion criteria

Group A: DMD patients, MRI and NPE

Group B: controls, MRI and NPE

Group C: DMD patients, NPE

For group A/C:

Able to provide written informed consent

Age 18 years or older

Genetically confirmed diagnosis for DMD

Male

For group B:

Able to provide written informed consent

Age: maximally one year apart from a DMD participant (expected to be between 18-30 years old)

Male

Exclusion criteria

A potential subject who meets any of the following criteria will be excluded from participation in this study in group A:

- MRI contraindications
- Inability to complete the NPE

A potential subject who meets any of the following criteria will be excluded from participation in this study in group B:

- A diagnosis for any neurological or neuropsychological disorder
- MRI contraindications
- Inability to complete the NPE

A potential subject who meets any of the following criteria will be excluded from participation in this study in group C:

- Inability to complete the NPE

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:	Other

Recruitment

NL	
Recruitment status:	Completed
Start date (anticipated):	27-06-2022
Enrollment:	40
Type:	Actual

Ethics review

Approved WMO	
Date:	21-10-2021
Application type:	First submission
Review commission:	METC Leiden-Den Haag-Delft (Leiden)
	metc-ldd@lumc.nl

Approved WMO	
Date:	25-03-2022
Application type:	Amendment
Review commission:	METC Leiden-Den Haag-Delft (Leiden)

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	NL75238.058.21