

# Brain study



## Brain imaging and Cognition in patients with DMD

### Hub Summary

This 2-year natural history study is designed to study the progression of pathological changes in the brain associated with the absence of dystrophin. The study will focus on cognitive impairment as well as looking at the relationship between the outcome measures and behavioural functioning.

**Study Number: NOT ON clinicaltrials.gov**

**Description by Newcastle-upon-Tyne Hospitals NHS trust**

### Primary Outcome Measures

Quantitative assessment of progressiveness of pathological changes in brain associated with dystrophin absence and cognitive impairment by:

Comparison upon follow-up of

- cerebral (sub-)volumes
- localization of differences in cerebral volumes
- cortical thickness
- cerebral perfusion
- anatomical connectivity
- functional connectivity
- metabolism as assessed by diffusion weighted spectroscopy (DWS)

### Secondary Outcome Measures

To study the relationship between the above mentioned MR parameters and:

- the results from neuropsychological testing and behavioral functioning
- the *DMD* gene mutation

### Can I take part?

#### Inclusion Criteria

- ✓ DMD patients of eight years and older with a known genetic mutation in the dystrophin gene.
- ✓ Male disease controls of similar age to the DMD boys with mutations affecting the dystrophin glycoprotein complex, but intact dystrophin.
- ✓ Healthy boys of eight years or older form the control group.

#### Exclusion Criteria

General exclusion criteria are:

- ✗ Claustrophobia
- ✗ Pacemakers and defibrillators
- ✗ Nerve stimulators
- ✗ Intracranial clips
- ✗ Intraorbital or intraocular metallic fragments
- ✗ Cochlear implants
- ✗ Ferromagnetic implants (e.g. thoracic implant for scoliosis)
- ✗ Inability to lie supine during less than 45 minutes
- ✗ not having a general practitioner

Exclusion criteria for healthy controls

- ✗ any muscle disease
- ✗ a brain disorder (such as severe brain concussion in past history, congenital brain anomalies, epilepsy)

## Trial Status

### Trial complete

**UK Locations**  
Newcastle, Trial complete /terminated

**Trial Sponsor**  
Newcastle-upon-Tyne Hospitals NHS trust

**Phase**  
Observational

**Length Of Participation**  
2 years

**Recruitment Target**  
32

**Ambulatory**  
Ambulant and non-ambulant, Not specified

**Therapeutic Category**  
Observational study

**Age**  
8 and over

**Mutation Specific**  
All treatment types

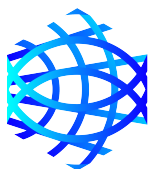
**Muscle Biopsy**  
No Muscle Biopsy Required

**MRI**  
Yes

[dmdhub.org](https://dmdhub.org)



For contact details and to find out more, please refer to [dmdhub.org](https://dmdhub.org).



Duchenne  
UK

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