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## TREAT-NMD Care and Trial Sites Registry Information Chart

Below you find a list of the information that is asked in the CTSR of all registered neuromuscular disease sites. For more information please visit [www.treat-nmd.eu/ctsr](http://www.treat-nmd.eu/ctsr) or contact us by e-mail ([ctsr@uniklinik-freiburg.de](mailto:ctsr@uniklinik-freiburg.de)).

### Information collected by the CTSR:

#### General Information

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Contact details from all the registered sites are collected

- Site name
- Title
- First name
- Last name
- Address
- Zip-Code / City
- Country
- E-Mail
- Phone
- Fax
- Homepage

#### Patient Population

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Patient Population (number of patients) has been separated into the following age groups

- Infants and Toddlers (0-2)
- Children (3-11)
- Adolescents (12-17)
- Adults (18-45)
- Senior Adults (45+)

The information is collected for the following diseases

- Duchenne Muscular Dystrophy (DMD)
- Becker Muscular Dystrophy (BMD)

- Limb Girdle Muscular Dystrophies (LGMD)
- Congenital Muscular Dystrophies (CMD)
- Congenital Myopathies (CM)
- Facioscapulohumeral Muscular Dystrophy (FSHD)
- Myotonic Dystrophy 1 (DM1)
- Myotonic Dystrophy 2 (DM2)
- Other genetic skeletal muscle diseases
- Acquired skeletal muscle diseases
- Spinal Muscular Atrophy I (SMA I)
- Spinal Muscular Atrophy II (SMA II)
- Spinal Muscular Atrophy III/IV (SMA III/IV)
- Other genetic motor neuron diseases
- Amyotrophic lateral sclerosis (ALS)
- Acquired motor neuron diseases
- Congenital Myasthenic Syndrome (CMS)
- Muscular Channelopathies (periodic paralysis, non-dystrophic myotonias, episodic weakness with ataxia and/or migraine, other muscular channelopathies)
- Genetic neuromuscular junction diseases
- Acquired neuromuscular junction diseases
- Genetic motor neuropathies
- Acquired peripheral neuropathies
- Mitochondrial disorders

## Diagnostic Tools

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### Diagnosis of DMD

Availability of the following methods to establish the diagnosis “DMD” (categorized into *available and funded / available but not funded / not available*)

- MLPA analysis
- Limited deletion/duplication analysis
- Point mutation detection
- Dystrophin analysis on muscle biopsy

### Diagnosis of SMA

Availability of the following methods to establish the diagnosis “SMA” (categorized into *available and funded / available but not funded / not available*)

- SMN 1 deletion test
- SMN 1 point mutation test
- SMN 2 copy-number

### Diagnosis of UCMD dystrophy by

Availability of the following methods to establish the diagnosis “UCMD” (categorized into *available and funded / available but not funded / not available*)

- muscle biopsy
- COL6A1/2/3 sequencing

- fibroblast culture

### **Diagnosis of MDC1A by**

Availability of the following methods to establish the diagnosis “MDC1A” (categorized into *available and funded / available but not funded / not available*)

- muscle biopsy
- LAMA2 sequencing
- fibroblast culture

### **Diagnosis of CMS by**

Availability of the following methods to establish the diagnosis “CMS” (categorized into *available and funded / available but not funded / not available*)

- motor nerve conduction
- repetitive nerve stimulation
- single fibre EMG
- EMG
- CHRNE genetic testing
- COLQ genetic testing
- DOK7 genetic testing
- RAPSN genetic testing
- Expanded panel (the above plus MUSK, AGRN, ALG2, ALG14, DPAGT1, GFPT1, CHRNA, CHRNB, CHRND, CHAT)

### **Diagnosis of HMN by**

Availability of the following methods to establish the diagnosis “MDC1A” (categorized into *available and funded / available but not funded / not available*)

- motor nerve conduction
- repetitive nerve stimulation
- PMP22 deletion test
- EMG

### **Diagnosis of muscular channelopathy by**

Availability of the following methods to establish the diagnosis “MDC1A” (categorized into *available and funded / available but not funded / not available*)

- EMG
- CACNA1S genetic testing
- SCN4A genetic testing
- CLCN1 genetic testing
- DM1 genetic testing
- DM2 genetic testing
- CACNA1A genetic testing
- KCN1A genetic testing

## **General Care Settings**

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**Availability of specialists or services internal/as team members/joint clinics or as external referral.**

- Paediatric Neurologist
- Neurologist
- Pulmonologist
- Cardiologist
- Orthopedic Surgeon
- Genetic Counseling
- Social Worker
- Orthotist
- Psychologist
- Physiotherapist
- Occupational Therapist
- Speech/Language Therapist
- Care Coordinator
- Endocrinologist

**Arrangement for transition from paediatric to adult care**

- Joint clinic
- Regular personal contact between paediatric and adult neurologist
- No transitional arrangement

**Availability of pulmonary function tests**

- Forced Vital Capacity (FVC)
- Peak cough flow
- Blood gas analysis (CO<sub>2</sub>, O<sub>2</sub>)
- Transcutaneous measurement of pCO<sub>2</sub>
- Transcutaneous measurement of spO<sub>2</sub>
- Polysomnography

**Available tests for heart function**

- 2-D Trans-thoracic echo-cardiogram
- Tissue Doppler imaging evaluations of the left ventricle and/ or wall motions
- 12-lead ECG with rhythm strip
- 24h ECG
- Cardio-MRI

**Availability of tests for muscle and bone health**

- Muscle MRI
- Dual energy X-ray absorptiometry (DXA)
- Peripheral quantitative computed tomography (pQCT)
- Others (freetext)

**Available facilities and equipment for physical therapy**

- Standardised four-stairs climbing test
- 10m straight stretch of floor for timed walk test
- 30m straight hallway for the Six-Minutes Walk Test

- 15cm high box step
- Hand-held Myometry
- Other devices for QMT (Quantitative Muscle Testing)

### **Muscle biopsies**

- The sites are asked if they are performing skeletal muscle biopsies and if yes how many in average annually:
  - open biopsies under general anaesthesia
  - open biopsies under local anaesthesia with/without sedation
  - needle biopsies under general anaesthesia
  - needle biopsies under local anaesthesia with/without sedation

### **Duchenne Care**

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**Rough percentage about how many of all DMD patients treated at the centre come at least once a year.**

**Reasons why DMD patients come less than once a year** (very important / somewhat important / insignificant / don't know):

- Not recommended to attend the neuromuscular clinic at least once a year
- Difficulty to provide an appointment / long waiting list
- Economic difficulties / travel is too expensive
- Distance: clinic too far away / journey is too long
- Patients prefer their general practitioner for regular assessment
- Patients don't want to come so often
- Others (freetext)

**Typical length of time a patient with DMD spends with each of the following health care providers (in minutes):**

- Neuromuscular doctor
- Physiotherapist
- Nurse

**Type and frequency of assessments an ambulatory child with DMD usually gets** (2x per year / 1x year / < 1x per year / not regularly – with symptoms / not performed):

- Functional abilities (e.g. time testing)
- Muscle force (myometry or manual muscle testing)
- Range of motion of joints
- Echocardiography
- Measurement of Forced Vital Capacity (FVC)
- DEXA or CT Scan to measure bone density
- No paediatric patients seen in the clinic
- Comments (freetext)

**Type and frequency of assessments a non-ambulatory boy without respiratory complications or cardiomyopathy usually gets** (2x per year / 1x year / < 1x per year / not regularly – with symptoms / not performed) :

- Functional abilities (e.g. time testing)
- Muscle force (myometry or manual muscle testing)
- Range of motion of joints
- Echocardiography
- Measurement of Forced Vital Capacity (FVC)
- DEXA or CT Scan to measure bone density
- No paediatric patients seen in the clinic
- Comments (freetext)

**Type and frequency of assessments an adult with DMD usually gets** (2x per year / 1x year / < 1x per year / not regularly – with symptoms / not performed):

- Functional abilities (e.g. time testing)
- Muscle force (myometry or manual muscle testing)
- Range of motion of joints
- Echocardiography
- Measurement of Forced Vital Capacity (FVC)
- DEXA or CT Scan to measure bone density
- No adult patients seen in the clinic
- Comments (freetext)

**Approaches to the treatment of ambulant DMD patients with corticosteroids. Closest approach:**

- Corticosteroids are offered to all patients
- Corticosteroids are only prescribed upon request
- Corticosteroids use not offered
- Others (freetext)

**Most important reason to start a treatment with corticosteroids:**

- Patient's age (with age specified)
- Stage of disease (motor progress / plateau phase / motor decline)
- Other reasons (freetext)
- Not applicable

**Approaches concerning the ending of the treatment with corticosteroids (most appropriate answer):**

- Usually stop when the patient has lost ambulation
- Usually continue after loss of ambulation but generally stop if side effects don't allow continuation of treatment
- Not applicable because steroids are not used routinely
- Others (freetext)

**Rough estimate of the percentage of DMD patients receiving corticosteroids:**

- ambulant patients
- non-ambulant patients

**Recommended measures on a regular basis to recognize or prevent side effects of corticosteroid treatment:**

- Completion of vaccination before start of steroids
- Regular measurements of weight and height
- Contact with a dietician
- Ophthalmologic control for cataracts
- Control of blood glucose level or glucosuria
- Blood pressure measurement
- Control of vitamin D levels
- Control of bone mineral density
- Comments (freetext)

**Availability of specific equipment or treatment for all patients, when it is indicated:**

- Manual wheelchair
- Electric wheelchair
- Regular physiotherapy
- Assisted ventilation (non-invasive via mask or invasive via tracheostomy)
- Scoliosis surgery
- Comments (freetext)

**Most important reason for not receiving the equipment / treatment as mentioned above. The reason should be specified according to the following possibilities:**

- not applicable / available for all patients
- economic reasons
- not available in our region
- patients refuse
- other reasons (freetext)

**Interest in attending specific training courses about the treatment of DMD:**

- Yes
- No
- Don't know

**The subjective relevance of the following topics should be rated (yes, very important / yes, less important / no, not necessary):**

- Psychosocial care
- Cardiac management
- Pulmonary management
- Neuromuscular management
- Corticosteroid therapy
- Orthopaedic management
- Gastrointestinal management
- Bone health
- Palliative care
- Others (freetext)

**Awareness of the family guide “The diagnosis and management of Duchenne Muscular Dystrophy” created by TREAT-NMD and the patient organizations PPMD, UPPMD and the MDA.**

- Yes
- No
- Don't know



## Clinical Research

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**Use of clinical data from neuromuscular patients for clinical research within the last 2 years (e.g. participation in clinical trials, natural history, imaging studies)**

- Yes
- No
- Don't know

**(Co)authorship of the contact person and/or neuromuscular team members on clinical research in peer reviewed journals within the last two years**

- More than 5 times
- 1-5 times
- None
- Don't know

**External funding for clinical research in neuromuscular disorders during the last two years**

- More than 1 academic position per year (or equivalent)
- Less than 1 academic position per year (or equivalent)
- None
- Don't know

## Education and Networking

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**Lectures or trainings on care for neuromuscular disorders given by the contact person and/or neuromuscular team members within the last two years**

- More than 5 times
- 1-5 times
- No
- Don't know

**Active participation in networks for neuromuscular diseases (Active means participation in meetings, sharing data, receive funding etc)**

- None
- National
- International

**Name(s) of the network(s) for neuromuscular diseases in which the site is participating (free text)**

## Personnel and experience

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### Potential Principal Investigator

Contact details of the potential principal investigator are gathered:

- Name
- Job title
- Telephone
- E-mail
- Fax
- Comment

### Staff that is available for clinical trials

- Information about type and number of personnel per function as well as the experience in clinical trials is collected :
  - Subinvestigator
  - Study nurses / Study coordinator
  - Lung specialist
  - Cardiologist
  - Pharmacist
  - Physiotherapist
  - IT-specialist

### Possibility to recruit additional patients for clinical trials through cooperation with other medical centres for research

- Yes
- No
- Don't know

### Clinical Trials Experience

- Familiarity with GCP (Good Clinical Practice)
- The sites are asked if they have already participated in a clinical trial or observational study and in which trial phase:
  - Phase I
  - Phase II
  - Phase III
  - Phase IV
- Is a site currently conducting a clinical trial? (title of the trial(s))
- Does the study team have experience with IVRS?
- Does the study team have experience with eCRF?
- Experience of study team with IATA packaging instructions
- Does the study team have been audited for a clinical trial, and if yes with which result

- What kind of audit was it? Sponsor audit or regulatory authority audit

## Equipment

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### Availability of the following facilities

- Certified Laboratory
- Centrifuge
- Ice
- Dry ice
- Liquid nitrogen
- Refrigerator lockable/not lockable with the following temperatures:
  - $-80^{\circ}\text{C}$  ( $-112^{\circ}\text{F}$ )
  - $-20^{\circ}\text{C}$  ( $-4^{\circ}\text{F}$ )
  - $2-8^{\circ}\text{C}$  ( $36^{\circ}\text{F}-46,4^{\circ}\text{F}$ )
- Lockable rooms or filling cabinets
- Lockable safes to keep study drugs
- Temperature recorder for the fridges and safes
- Possibility for inpatient care
- Dedicated clinical trials unit for children and/or adults
- Emergency care unit, if yes for whom:
  - Neonates and infants
  - Children / adolescents
  - Adults

### Data collection

- Internet access
- Possibility for international phone and fax calls