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 or contact us by e-mail (ctsr@uniklinik-freiburg.de).

Information collected by the CTSR:

General Information

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

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)

- Spinal Muscular Atrophy I (SMA I)
- Spinal Muscular Atrophy II (SMA II)
- Spinal Muscular Atrophy III (SMA III)
- Limb Girdle Muscular Dystrophies (LGMD)
- Congenital Muscular Dystrophies (CMD)
- Congenital Myopathies (CM)
- Facioscapulohumeral Muscular Dystrophy (FSHD)
- Myotonic Dystrophy 1 (MD1)
- Myotonic Dystrophy 2(MD2)
- Hereditary Motor Neuropathies (HMN)
- Congenital Myasthenic Syndrome (CMS)
- Muscular Channelopathies

Diagnostic Tools

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

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

- (Paediatric) Neurologist
- Pulmonologist
- Cardiologist
- Orthopedic Surgeon
- Genetic Counseling
- Social Worker
- Orthotist

- Psychologist
- Physiotherapist
- Occupational Therapist
- Speech/Language Therapist
- Care Coordinator

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_2, O_2)
- Transcutaneous measurement of pCO_2
- Transcutaneous measurement of spO2
- 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

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 <u>ambulatory child</u> 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 <u>a non-ambulatory</u> 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 <u>adult</u> 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 if Duchenne Muscular Dystrophy" created by TREAT-NMD and the patient organizations PPMD, UPPMD and the MDA.

- Yes
- No
- Don't know

Clinical Research

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

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

Personnel and experience

Potential Principal Investigator

Contact details of the potential principal investigator are gathered:

- Name
- Telephone
- E-mail
- Fax
- Qualifications

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
    - IT-specialist
    - Lung specialist
    - Cardiologist
    - Pharmacist
    - Physiotherapist

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)
- Availability of a dedicated clinical trials unit for children and / or adults
- The sites are asked if they have already participated in a clinical trial and in which trial phase:
    - Phase I
    - Phase II
    - Phase III
    - Phase IV
- Is a site currently conducting a clinical trial? (short description / title of the trial(s))
- Does the study team know how adverse events and reactions are defined?
- 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

Equipment

Availability of the following facilities

- Certfied Laboratory
- Centrifuge
- Ice
- Dry ice
- Liquid nitrogen
- Refrigerator lockable/not lockable with the following temperatures:
    - -80°C(-112°F)
    - -20°C (-4°F)
    - 2-8°C(36°F-46,4°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