
TREAT-NMD Care and Trial Sites Registry Information Chart

The following information is collected from all registered sites using a web-based questionnaire.

Each site has a user account and is regularly asked to log in and to update their data set. The current database contains information from more than 200 sites (June 2011) with a worldwide coverage and can be searched for specific selection criteria.

Further disease groups or other questions can be added to the questionnaire if needed. For more information please contact ctcc-info@uniklinik-freiburg.de.

General Information

Contact details about all the registered sites are collected

- Site name
- Title
- Firstname
- Lastname
- Address
- Zip-Code / City
- Country
- E-Mail
- Phone
- Fax

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 (MD1)

Diagnosis of DMD/SMA

The following types of DMD and SMA diagnosis are collected and categorised into *available* and *funded* / *available but not funded* / *not available*

- Limited deletion/duplication analysis
- MLPA analysis
- Point mutation detection
- Dystrophin analysis on muscle biopsy
- SMN 1 deletion test
- SMN 1 point mutation test
- SMN 2 copy-number

Diagnosis of UCMD/MDC1A by

The following UCMD/MDC1A diagnosis are collected and categorised into *available* and *funded* / *available but not funded* / *not available*

- clinical presentation
- muscle biopsy
- molecular testing
- fibroblast culture

Care Settings

The availability of specialists or services are collected and it is asked if they are available internally (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 pediatric 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 (CO₂, O₂)
- Transcutaneous measurement of pCO₂
- Transcutaneous measurement of saO₂?
- Polysomnography

Available tests for heart function

- Facility to perform 2-D Trans-thoracic echo-cardiogram?
- Tissue Doppler imaging evaluations of the left ventricle and/ or wall motions
- 12-lead ECG with rhythm strip?
- 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-Minute Walk Test
- 15cm high box step
- Hand-held Myometry
- Other devices for QMT (Quantitative Muscle Testing)

Availability of emergency care facilities

- Does a centre have an emergency care unit? If yes for whom:
 - Neonates and infants
 - Children / adolescents
 - Adults

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

Research Activities

Have clinical data from your neuromuscular patients been used for clinical research within the last 2 years? (e.g. participation in clinical trials, natural history, imaging studies)

- Yes
- No
- Don't know

Have you and/or your neuromuscular team members been (co)author of articles on clinical research in peer reviewed journals within the last two years?

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

Have you/your team members given training/lectures on care for neuromuscular disorders within the last two years?

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

Are you actively participating in networks for neuromuscular diseases? (Active means participation in meetings, sharing data, receive funding etc).

- None
- National
- International

Have you received 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

Clinical Trial Infrastructure

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

- Personnel information is gathered and the number of personnel per function is collected and if they are experienced in clinical trials:
 - 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

- Yes
- No
- Don't know

Clinical Trials Experience

- Familiarity with GCP (Good Clinical Practice)
- Availability of a dedicated clinical trials unit
- 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 teams know about 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

Other equipment

Availability of the following facilities is recorded:

- Centrifuge for blood sample
- Possibility for inpatient care
- Refrigerator lockable/not lockable with the following temperatures:
 - -80°C (-112°F)
 - -20°C (-4°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
- Certified laboratory

- Lab provision of normal ranges

Data collection

- Internet access
- IBM compatible computer
- Version of MS Windows
- Possibility for international phone and fax calls