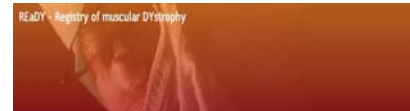


Czech National Registry of Myotonic Disorders

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The patient registries belong to the core activities which can help us in planning of the effective health care, assessing standards of diagnosis and care, and answer the questions concerning on epidemiologic data. Besides of the local hospital-based databases and registries we can find four national registries of hereditary neuromuscular disorders associated under unique name: ReaDy (registry of muscular dystrophy). Four registries are currently running: Duchenne/Becker muscular dystrophy, spinal muscular atrophy, myotonic disorders, and facioscapulohumeral muscular dystrophy. Each registry is independent and has its own curator. The registries are under the supervision of Czech neuromuscular society.



Technological remarks

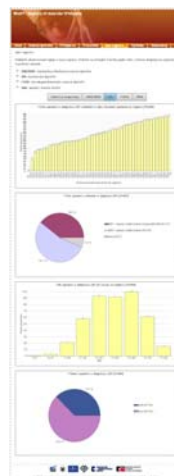
The technological aspect of the project, the data collection, storage and backup and their analysis are provided by the Institute of Biostatistics and Analyses, Masaryk University, Brno, CR. On-line data collection is based on a TRIALDB system developed on Yale University, Connecticut, USA, which is widely used for this purpose. So it is not necessary to install any additional computer software. The database can be accessed only by authorized persons using their login and password. For each patient is generated a unique ID; all data transfer is encrypted and the system is designed to prevent their unauthorized use during data transfer. Laws and regulations in CR require having an informed consent from all patients whose data are used in the registry. All claims for personal data protection were met. Data are stored on the central server on Masaryk University in Brno in Oracle 9i database.

Other (user-friendly) functions

- Database users are allowed to print the submitted patient forms
- Selected data are on-line exported to the web pages of the ReaDy (ready.registry.cz)
- Each patients can see her/his own data after secure individual authentication
- The battery of questionnaires are available, partially for self-evaluation of patients
- Documents repository for patients and physicians
- Helpdesk

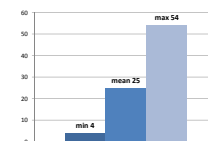


Průběh onemocnění	Průběh onemocnění	Průběh onemocnění	Průběh onemocnění	Průběh onemocnění	Průběh onemocnění	Průběh onemocnění	Průběh onemocnění	Průběh onemocnění	Průběh onemocnění
1998-2000	2000-2002	2002-2004	2004-2006	2006-2008	2008-2010	2010-2012	2012-2014	2014-2016	2016-2018

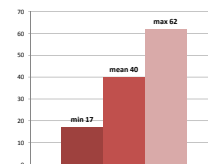


DM1 vs. DM2

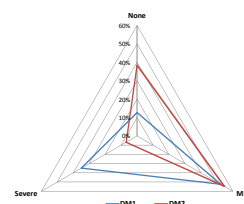
DM1 age of onset (y)



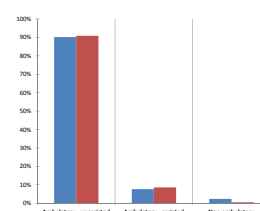
DM2 age of onset (y)



Myotonia (DM1, DM2)



Walking ability



Up to January 2015 422 patients from 8 centres has been included. The majority (84%) of all records are from centres in Prague and Brno.

The average annual recruitment during total 3.5 years period are 121 patients, but the last year the acquisition rate decreased to 69 patients.

The mean follow-up time in the registry is 16 months.

The majority consists from patients with DM2 (n=207, 49.1%) and DM1 including congenital form (n=157, 37.2%).

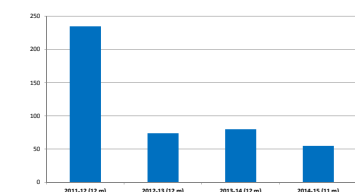
Non dystrophic myotonias (chloride and sodium channelopathies) are represented with 26 persons (6.1%). The rest are asymptomatic mutation carriers and files with poor defined or missing data (32 items).

Among patients with DM (1 and 2) there are 219 females (63%) and 129 males (37%). Mean age in the time of the registry entering is 45 years, approximately 10 years after disease manifestation which was in patient with DM1 25 (10- 54) years and in persons with DM2 40 (17-62) years.

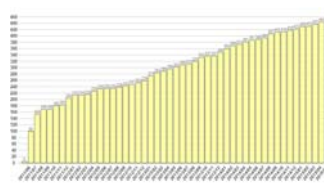
Nearly all patients with DM1 and DM2 are ambulatory (assisted or unassisted). Only 4 patients are wheelchair bound.

(Some pictures are based on last data update May 30, 2015)

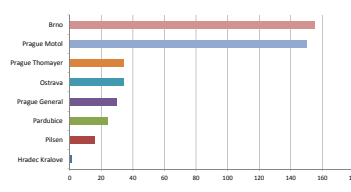
Annual recruitment rate



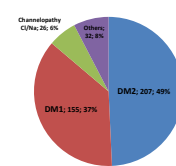
Recruitment rate since launching



Contributions of the neuromuscular centres



Diagnostic groups



Acknowledgement

The registry is the common work of all Czech neuromuscular centres, molecular biology laboratories (Centre of Molecular Biology and Gene Therapy, University Hospital Brno and Institute of Biology and Medical Genetics Prague-Motol), and Institute of Biostatistics and Analyses (Masaryk University Brno).

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