PATIENT REGISTRY OF DMD/BMD IN THE CZECH REPUBLIC

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Duchenne muscular dystrophy

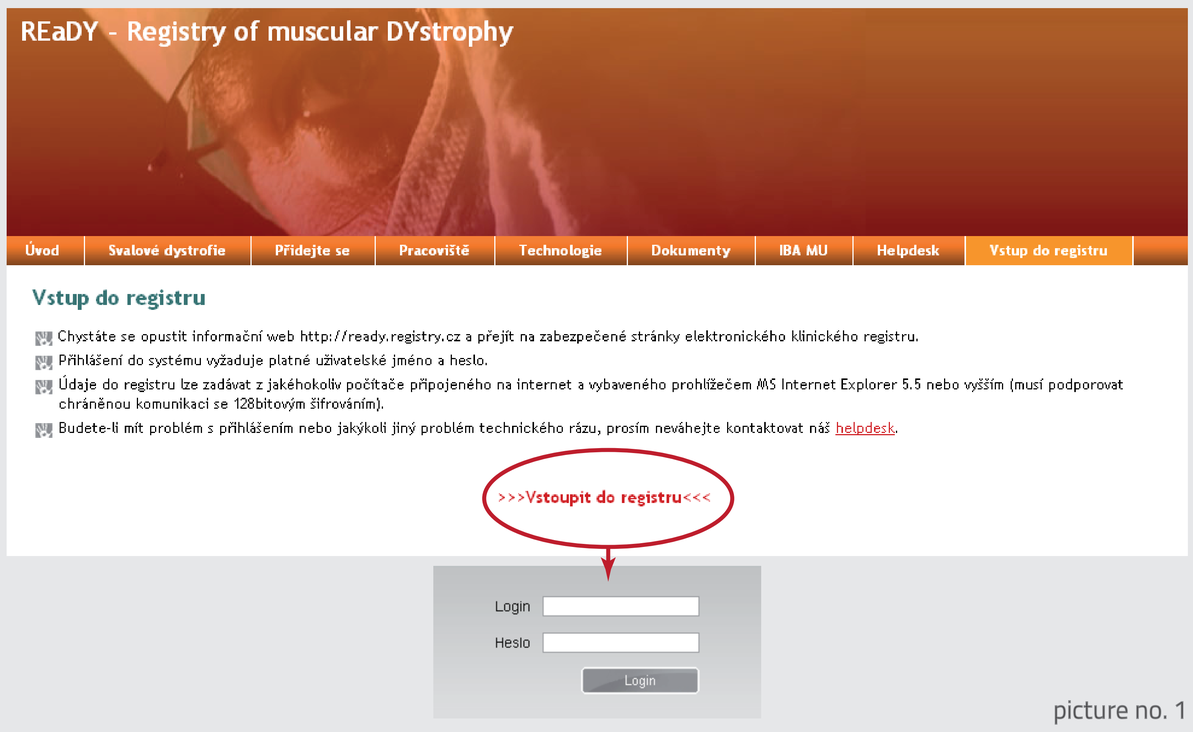
Duchenne muscular dystrophy (DMD) as well as other neuromuscular diseases belongs to orphan diseases. It means that its prevalence is less than 1 case in 2,000 inhabitants.
DMD is the most common of muscular dystrophies, affecting approximately 1 in every 3,500 newborn boys. It is caused by a fault in a gene called the dystrophin or DMD gene. The fault in this gene stops the body from making a protein called dystrophin which is important in muscle fibres and its absence results in muscle weakness.
The dystrophin gene is located on the X chromosome and that's why DMD affects only boys. DMD can run in a family even if only one person in the biological family has it. This is because of the ways in which genetic diseases are inherited. A mother who is a carrier has a 50 percent chance of having a son with DMD.
DMD is a progressive disease, affected boys usually walk later than other boys their own age, have troubles with jumping, running etc. One of the typical signs of DMD is Gower's sign (or maneuver). At the age of ten (or a little bit later) boys lose their ability to walk. Over time they develop breathing difficulties and they need permanent ventilatory support. They usually die very young, between 20-30-years-old .
There is still no cure for DMD, but there is active research ongoing and new types of treatment are being tested in clinical trials.
Manipulation of pre-mRNA splicing by antisense oligonucleotides (AOs) offers considerable potential for a number of genetic disorders.
As these diseases are rare, one of the most important parts of care for these patients are patient registries which collect information about patients affected by this disease. They represent an essential tool for researchers and health care providers as they contain genetic and clinical data and patient contact information.

REaDY patient registry (REgistry of muscular DYstrophy)

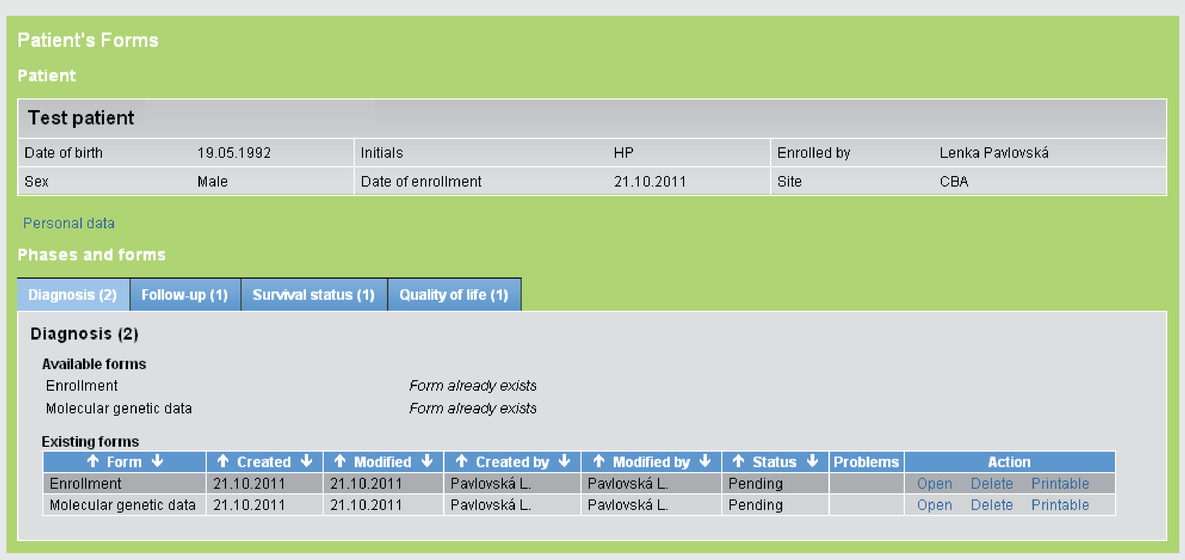
REaDY was developed in cooperation with IBA MU (Institute of Biostatistics and Analyses, Masaryk University), FN Brno (The University Hospital Brno) and a non-profit organisation PARENT PROJECT.
The project was established in May 2011 in continuation of an international project DMD/BMD established in 2004.
REaDY is a multicenter non-interventional study with the main aim to monitor Czech and Slovak patients diagnosed with orphan diseases – Duchenne muscular dystrophy, Becker muscular dystrophy, spinal muscular atrophy myotonic dystrophy etc.
Its data structure has been created on the basis of documents from TREAT-NMD, a network for the neuromuscular field. The registry is collecting data from 12 centres in the Czech Republic and Slovakia and is available only in English.
REaDY registry currently provides a clinical picture of 150 DMD patients, 39 BMD patients, 3 suspect carriers, 45 verified carriers. Patients enrolled in the registry also participate actively in the data collection by means of filling out Quality of life form-available only in Czech language. Six forms have been filled out since May 2011 already.

Trial Registry Platform

Access to the trial registry is allowed only to authorized users - Data Managers. This rule applies to everyone - to patients and to medical doctors as well - see picture no. 1 Entry to registry.



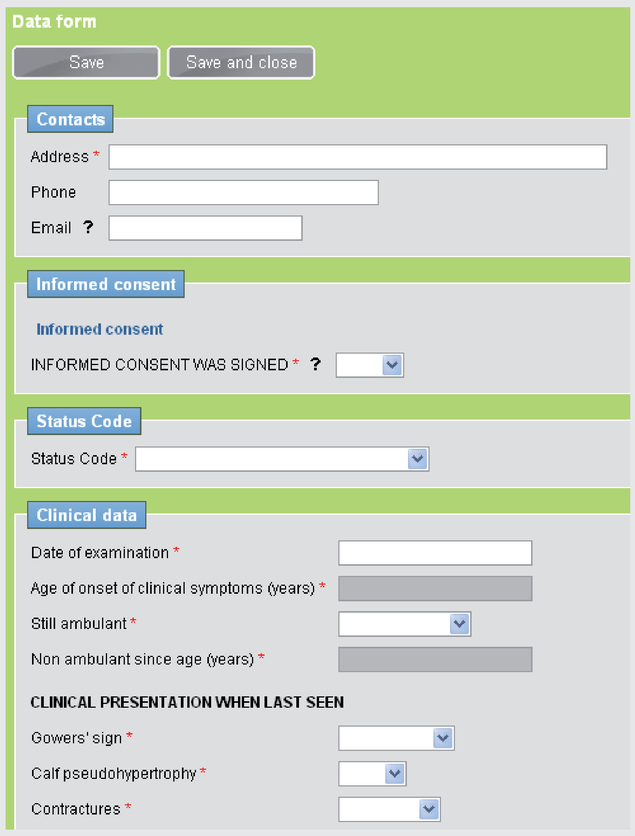
picture no. 1



picture no. 2

Case report form is divided into 4 phases - Diagnosis, Follow-up, Survival Status, Quality of life (see picture no. 2). All of them are available to doctors. Patients are allowed to fill in the phase Quality of life.
Each phase includes at least one form.
Diagnosis - Enrollment (picture no. 3) and Molecular genetic data - contain basic information about dystrophies such as Clinical data, Biochemistry, Genetic etc.
Follow-up - Follow-up examination - this form is filled in once a year.
Survival status - this form describes current information about patient's condition.
Quality of life - the form evaluates quality of patient's life from his point of view
Data from the REaDY registry enable to enroll our patients quickly into international clinical studies and help medical doctors in the development and treatment of these hereditary diseases.

More information can be found on the web site **www.ready.registry.cz**



picture no. 3

PARENT PROJECT svalová dystrofie
Výzkum Léčba Naděje

PARENT PROJECT is a non-profit organization that is run by parents of children diagnosed with Duchenne (DMD) and Becker (BMD) Muscular Dystrophy. It was founded in 1994 in the USA and it has been registred in the Czech Republic since 2001. It is participating with FN Brno and IBA in this registry.
It helps keep affected families in touch and helps patients find appropriate clinical trials.