

## **157TH ENMC INTERNATIONAL WORKSHOP ON “PATIENT REGISTRIES FOR RARE, INHERITED MUSCULAR DISORDERS”**

**25-27 January 2008, Naarden, the Netherlands**

At the end of January, the European Neuromuscular Centre (ENMC), in collaboration with TREAT-NMD partners, hosted a group of experts in Naarden for a discussion on patient registries for rare, inherited muscular disorders. Patients' registries represent crucial start-ups for new therapeutic strategies for neuromuscular diseases, as well as to answer research questions such as the prevalence of neuromuscular disorders in Europe and support other activities such as assessing standards of care. A major goal of the integrating activity within TREAT-NMD is the introduction of European patient databases for DMD, SMA and other muscular dystrophies, and harmonized TREAT-NMD global registries for DMD and SMA have been already set up, together with a Charter for TREAT-NMD patient database/registries. A “toolkit” applicable to registry development in other diseases including a discussion on the selection of mandatory items, ethical and confidentiality issues, as well as the Charter for data sharing are available on the TREAT-NMD website.

The ENMC workshop aimed to create a collaborative action towards the set up, development and harmonization of patient registries for additional inherited neuromuscular disorders, both in Europe and world-wide. This workshop provided an excellent development of the work of TREAT-NMD and ENMC into rare muscle diseases towards clinical trial readiness, complementing the ongoing TREAT-NMD activities for DMD and SMA registries. The workshop was attended by 20 participants from 7 European countries (Finland, France, Germany, The Netherlands, Spain, Switzerland, United Kingdom) and from the U.S.A. Participants included bioinformatics experts, geneticists, neurologists, paediatricians, representatives of patient advocacy organizations, as well as members of the TREAT-NMD and ENMC. The discussion was focused on existing databases and registries, ranging from laboratory and clinical based databases to patient led initiatives, relating to several types of congenital muscular dystrophy and myopathy, limb girdle muscular dystrophies, oculopharyngeal muscular dystrophy, myofibrillar myopathies, Pompe Disease, facioscapulohumeral muscular dystrophy and myotonic dystrophy. Technical and legal issues, objectives and requirements for each registry, development and agreement on mandatory items across genes and diseases, agreement on best practices and further plans for collaboration were discussed as well. Agreement was reached on the importance of European and global harmonized and coordinated patient registries for accelerating progress, in particular in translational research. This will be facilitated by action taken by individual, disease-specific ENMC workshops and consortia, but also through joint action across diseases. This includes a web-based inventory of patient registries to be published, harmonization of registration practices and registry content, increased collaboration with patient organizations and empowerment of patients, and sharing of resources and dissemination through ENMC and TREAT-NMD.

The participants of the workshop wish to invite our partners and additional representatives for rare, inherited muscle disorders to get involved and form a consortium for further work in this area. Additional inputs are welcome! A full report will be soon published in Neuromuscular Disorders and will be complemented by an inventory of the registries available. Further details will be released on the TREAT-NMD website.