a 88.6% of the maximum score. Especially activities above shoulder level are problematic. Patterns of limitations in arm function are further analyzed as are correlations with disease course, age, and gender. The project McArm is a national Dutch ‘Piekien in de Delta’ project. It is partially funded by: Agentschap.nl, an agency of the Dutch Ministry of Economic Affairs, Agriculture and Innovation, the Province of Noord Brabant and the Province of Limburg.

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S.P.15
Utility of subjective pain scales for assessments of standing with knee–ankle–foot-orthoses in Duchenne muscular dystrophy
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Standing with knee–ankle–foot orthoses (KAFOs) in rehabilitation for Duchenne muscular dystrophy (DMD) helps prevent lower limb contracture and progression of scoliosis. As joint contracture progresses, compression from orthoses can cause pain, making it difficult to continue standing. The purpose of this study was to evaluate the utility of subjective pain scales in assessments of continued standing with KAFOs. Subjects were 69 boys with DMD. The following were assessed 30 minutes after standing with KAFOs: visual analogue scale (VAS), Wong-Baker face scale (WBFS), and Lorish–Maisiak face scale (LMFS). Joint contracture was evaluated during hip extension, knee extension, and ankle dorsiflexion by measuring range of motion (ROM) with a goniometer. Spearman’s correlation coefficients were used to assess the relationship between pain scales and ROM. Receiver operating characteristic (ROC) curve analysis was used to calculate the sensitivity and specificity of each pain scale during continued standing with KAFOs. Factors associated with continued standing with KAFOs were determined using the cut-off values for each scale. ROM for ankle dorsiflexion was moderately correlated with VAS (ρ = 0.42, p < 0.001), WBFS (ρ = 0.37, p = 0.002), and LMFS (ρ = 0.34, p = 0.004). ROM for knee extension was moderately correlated with WBFS (ρ = 0.30, p = 0.027), and weakly correlated with LMFS (ρ = 0.28, p = 0.041). There was no correlation between knee extension and VAS (ρ = 0.25, p = 0.072). ROM of hip extension was moderately correlated with WBFS (ρ = 0.32, p = 0.024) and LMFS (ρ = 0.36, p = 0.011), and weakly correlated with VAS (ρ = 0.297, p = 0.036). The optimal cut-off values for continued standing with KAFOs were 80 mm (100%) for VAS, 4 (100%) for WBFS, and 17 (100%) for LMFS. Our findings revealed correlations between pain and joint contracture, suggesting that these subjective pain scales are useful for assessments of continued standing with KAFOs.

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CARE-NMD aims to disseminate and implement best-practice standards of care for Duchenne muscular dystrophy (DMD) in Europe. Patient registries offer a valuable approach to engaging with the patient community, both to disseminate information and to survey their experiences. Registries permit the identification of a patient population with a precise genetic diagnosis, and are thus essential to the development of novel, mutation-specific therapeutic approaches such as exon-skipping. A core driving factor in their development has often therefore been clinical trial readiness: e.g. determining trial viability for a specific genetic mutation. However, as registries enable contact with a patient population, they also offer research opportunities outside the clinical trial domain. These include surveying availability of high-quality care and quality of life issues. Furthermore, registries permit the distribution of care information aimed at that particular audience. CARE-NMD has utilised patient registries in both of these contexts. The project has promoted knowledge of best-practice care via the translation and dissemination of the Family Guide to the care standards. This is now available in 22 languages via the CARE-NMD and TREAT-NMD websites, with 200 monthly downloads. The project has also conducted, via national patient registries in seven countries (Bulgaria, Czech Republic, Denmark, Germany, Hungary, Poland, and the UK), the largest ever survey of care and quality of life for DMD. The overall response rate is 66%, with 1100 responses received (April 2012), and national response rates of 48–89%. The data gathered provide unparalleled information on the experience of patients and families living with DMD across Europe. The use of registries also enables the return of information to the patient community, enhancing patient-led advocacy for the availability of better care, and strengthening mutual understanding between rare disease researchers and the patient community.

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S.P.31
What was the age and cause of death in patients with Duchenne muscular dystrophy in Sweden during 2000–2010?
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The aim of this project was to study the age and cause of death in patients with Duchenne muscular dystrophy (DMD) in Sweden. DMD is the most common neuromuscular disorder in childhood with an incidence of between 1:3500 to 1:4500 male births. The disorder is X-linked recessive and caused by mutations in the dystrophin gene. The gene product dystrophin has been localized to the sarcolemma. Absent dystrophin leads to progressive muscle wasting with degeneration and necrosis of muscle fibers. The disorder mainly affects the skeletal muscle and the myocardium. Onset usually occurs before the age of three years with progressive muscle weakness and loss of ambulation between seven and 13 years of age. Respiratory insufficiency occurs at 16–18 years of age. No cure is available, but corticosteroid treatment has been reported to result in improvement of muscle strength and delay loss of motor function. Home
mechanical ventilation has been introduced during the last decades for patients with DMD and has been reported to result in increased survival. The cause of death in patients with DMD from Sweden was studied for the time period 2000–2010. Information about the date of death and cause of death (multiple causes if relevant) was retrieved from the Cause of Death Registry which is managed by the Swedish National Board of Health and Welfare. Cause of death was further studied in the patients’ medical journals. During 2000–2010 64 patients with DMD have been localised so far in the Cause of Death Registry. The mean age of death was 25 years, with a range of 10–46 years. The main cause of death was related to cardiac failure in 40% and to respiratory failure in 35%. Cardiac and/or respiratory failure are still the main causes of death in DMD in Sweden. The age of death reported in this study is similar to that reported in recent studies from other countries.

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S.P.32
The HIBM Patient Monitoring Program (HIBM-PMP): Registry and natural history study to advance research and clinical management in Hereditary Inclusion Body Myopathy (HIBM or GNE Myopathy)
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HIBM is an autosomal recessive, non-inflammatory, neuromuscular disorder caused by a defect in the biosynthetic pathway for sialic acid with no approved treatment. Given the rarity of the disease and the potential for novel therapies, there is a need to characterize the clinical presentation and progression of disease to support treatment development. This program proposes the use of a single web-based electronic data capture and management platform to conduct two integrated and connected data collection efforts: a HIBM disease registry and a natural history study. The registry will solicit information on medical/diagnostic history, and clinical symptoms/progression from patients and/or their physicians using an online questionnaire. Patient-reported data would be confirmed with medical professionals, providing an intermediate level of data verification. Registry patients may be recruited by their physicians to participate in the natural history study which will be conducted at specialized centers. The study involves annual visits to assess biochemical markers of sialylation, muscle strength and function and patient-reported outcomes. Data from both studies would be monitored against medical records to create a GCP-compliant dataset. The hybrid design maximizes the ease of registry participation yet promotes the development of rigorous datasets to support the sharing of verified data with regulatory agencies, clinicians, patients and academia. A governance board with a defined charter and representation from public, private and academic sectors will establish and enforce policies and procedures for access to and use of HIBM-PMP data. Physicians will have real-time access to data from their patients and can request additional data from the governance board for use in research, presentations and publications. The PMP for HIBM will be launched in 2012 at multiple sites in the US, Europe and Israel. Expansion of the platform into other diseases is planned in 2013.

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S.P.33
Australasian neuromuscular disease registry
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Disease registries provide opportunity for clinical trials and improved service provision. The most effective registries are those designed in partnerships to operate within a harmonised global network. The Australasian Neuromuscular Disease Registry (ANMDR) has been created in collaboration with Treat–NMD, WA Health, the Centre of Comparative Genomics and clinical and patient stakeholder groups. The purpose of ANMDR is to serve the needs of the neuromuscular disease community by (1) improving health service planning and (2) identifying patients for follow-up, on the basis of their demographic, clinical or genetic profile, who may benefit from access to emerging diagnostic tools or therapeutics. ANMDR houses specific disease registries including muscular dystrophy, myotonic dystrophy, spinal muscular atrophy, with capacity to house registries for other specific diseases in the future, such as fascioscapular humeral dystrophy and congenital muscular dystrophy. The registry collects clinical data in a secure manner that allows interoperability with other registries. Upon approval by an independent Advisory Board, the registry sends anonymised data to those registries and appropriate researchers. ANMDR is internet based and provides for the ability to control and restrict access to the information (as determined by the Registry Curator), and to interpret data outputs. The registry enables participation in integrated and unified approaches for research in service provision and planning, information sharing, best practices, biobanking, harmonised data collection, analyses and reporting, and creating more robust ethical and legal frameworks. The ANMDR is one element of a larger plan to offer local opportunities to access globally consolidated shared resources, infrastructure and policy. Policy and planning to contextualise needs and identify opportunities of the ANMDR requires both local and international input.

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S.P.34
ReaDY: The Czech national registry of myotonic disorders
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The patient registries are the key activities which can help us in planning of the effective health care, assessing standards of diagnosis and care, and answer the questions concerning of the prevalence of neuromuscular disorders. Preliminary survey in the nine neuromuscular centers and four genetic laboratories in Czech Republic (CR) revealed about 400 patients with myotonic disorders. The majority seems to be the patients with myotonic dystrophy type 2 (MD2), but the exact figures have not been available up to date. Therefore Czech neuromuscular society decided to establish the national myotonic registry which can answer these questions. 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. The registry was launched in the June 2011 and up to February 2012 contains 235 records from eight Czech neuromuscular centers. The database