include 76 patients with MD1, 116 patients with MD2, 25 peoples with CICN1 mutations, and 14 persons with SCN4A mutations. The majority (85%) of records are from two centers (Prague and Brno), five centers have completed less than 10 records.

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S.P.35
The Human Variome Project – Sharing data – Reducing disease
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The Human Variome Project (www.humanvariomeproject.org), a Consultative Partner of the United Nations Educational, Scientific and Cultural Organisation, is the international initiative to collect, curate, interpret and share information on all human genetic variation. The Project establishes and maintains the necessary standards, systems and infrastructure for genetic knowledge sharing, offers training and education for clinicians, researchers and the general public and works with individual countries to build their medical genetics and genomics capacity. These activities promote the development of better genetic services and will lead to the improvement of genetic treatment and diagnostic abilities worldwide. The Human Variome Project has established a Global Collection Architecture to ensure comprehensive collection of genetic data worldwide and encourages collection via Human Variome Project Country Nodes and Gene/Disease Specific Databases. Examples of each of these include the Human Variome Project Australian Node (www.hvpaustralia.org.au) and InSiGHT database (InSiGHT, www.insight-group.org). An International Confederation of Countries has also been initiated with Australia, Austria, Belgium, China, Cyprus, Egypt, Greece, Kuwait, Malaysia, Spain, Vietnam and Nepal in the application process. We believe that there will be synergy between the myogenetics initiative and the Human Variome Project. Potential collaboration on data collection between the Human Variome Project Australian Node (www.hvpaustralia.org.au) and the Australian Neuromuscular Network is currently under discussion.

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S.P.36
The six months performance of neuromuscular diseases registry system of Turkey (Kukas)
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KUKAS is a database and website which is especially developed for registries of Duchenne Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA) patients in Turkey. The aims of the system are to register initially DMD and SMA patients and secondly, other rare seen neuromuscular diseases; and to inform patients/caregivers or professionals about neuromuscular diseases with updated datas on website. This study, general usage traffic of www.kukas.info which is used by patients/caregivers or professionals and hasta.kukas.info which is used by researchers between the dates 01/09/2011–13/03/2012 are given by using Google Analytics. Researchers made 401 visits to the website with average 9.9 and totally 3.696 pages visits. Three hundred and eighty six visits have been made from Turkey and 15 visits from other countries during 6 months. The average time spent by researchers on the site was 9.29 min. 1.166 website visits and 5.535 pages visits were made between these dates. 1.118 visits were made from Turkey and 48 visits from nine different countries. It is reported that visitors spent average 3.07 min in web site. Thirty patients made online registration. Neuromuscular Diseases Unit wellcomes patients from all around the country. Despite different education levels and social statuses of families, the higher numbers of online registrations and patients benefitting the website than expected shows that, the study have been developing in accordance with the aim of the system.

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S.P.37
The New Zealand Neuromuscular Disease Registry turns one – Data from the first year
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The New Zealand Neuromuscular Disease Registry was launched in September 2011. One year on we present data from participants recruited to the registry in its first year of operation. Patient registries for rare diseases have become increasingly recognised as an important step in facilitating research and propagating standards of care. The Muscular Dystrophy Association (MDA) of New Zealand, working in consort with interested clinicians and in collaboration with the Office for Population Health and Genomics in Western Australia has established the New Zealand Neuromuscular Disease Registry, which was launched in September 2011. Unlike many other registries in the rare disease field the NZ Neuromuscular disease Registry is not disease-specific, instead it covers a range of both paediatric and adult neuromuscular disorders including the muscular dystrophies, spinal muscular atrophies, hereditary neuropathies, congenital myopathies, myasthenia, myotonic syndromes, metabolic myopathies, inherited ataxias and inflammatory myopathies. This has allowed us to efficiently obtain ethics committee approval for all these conditions with one application. The nation-wide registry links into reputable international anonymised disease-specific registries including the Global TREAT NMD databases for DMD and SMA and the data collected from each participant is dependent upon the neuromuscular condition they have and the requirements of the international registry to which the NZ Registry is linked. In the case of participants who have conditions for which we have not identified an international database, the minimum data collected on each participant by the registry is basic demographic information as well as confirmation of diagnosis by genetic test. Here we report on the full range of data collected including clinical and molecular information for the first year of the registry.

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S.P.38
Infrastructure for new drug development to treat muscular dystrophy – Current status of patient registration in japan: REMUDY
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Clinical trials for new therapeutic strategies are currently being planned for Duchenne and Becker muscular dystrophies (DMD/BMD); however, many challenges exist in the planning and conduct of a clinical trial for rare diseases. The epidemiological data, total number of patients, natural history, and clinical outcome measures are mostly
unclar. Adequate numbers of patients are needed to achieve significant results in clinical trials. As solutions to these problems, patient registries are an important infrastructure worldwide, especially in the case of rare diseases such as DMD/BMD. Both inter- and out-side of Europe, TREAT-NMD, a clinical research network for neuromuscular disorders, developed a global database for dystrophinopathy patients. We have developed a national registry of Japanese DMD/BMD patients in collabora-
tion with TREAT-NMD. The database includes clinical and molecular genetic data as well as all required items for the TREAT-NMD global patient registry. As of February 2012, 690 patients were registered in this database. The purpose of this registry is the effective recruitment of eligible patients for clinical trials, and it may also provide timely information to regi-stants about upcoming trials. This registry data also provides more detailed knowledge about natural history, epidemiology, and clinical care. In recent years, drug development has become dramatically globalized, and global clinical trials (GCTs) are being conducted in Japan as well. It is appropriate, particularly with regard to orphan diseases, to include Japanese patients in GCTs to increase evidence for evaluation, because such large-scale trials would be difficult to conduct solely within one country. GCTs enable the synchronization of clinical drug development in Japan with that in other countries, minimizing drug approval delays.

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S.P.39
BIO- NMD: An update on the discovery and validation of biomarkers for Neuromuscular Diseases (NMDs) – An EU funded FP7 project
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BIO-NMD is an EU-funded translational research project which aims to identify and validate new biomarkers for neuromuscular diseases. It focuses on DMD, BMD and Collagen-6 myopathies and is a collaboration of 12 partners, coordinated by the University of Ferrara. Nearing the end of its three year funding, this poster reports on progress made, including the identifica-
tion of several new biomarkers correlated to aspects of NMDs. For example, macrophages as cell biomarkers mirroring skeletal muscle (Gualandi et al., 2011); MMP-9 as a serum biomarker which serves to mon-
tor disease progression in DMD (Nadarajah et al., 2011); components involved in autophagy as exploratory novel biomarkers for COLV1 disease (Grumati et al., 2010). Based on the novel miRNAs previously identified as originating from the dystrophin locus, partners are working to establish their effect on dystrophin isoform expression regulation and role in control-
lung dystrophin transcription (paper submitted). Potential implications are detailed for future research into NMDs and for treatment monitoring. Tools developed during the course of the project are described, such as a set of methods developed for high-throughput analysis of animal and patient samples including by whole exome sequencing, targeted resequenc-
ing of candidate genes, whole transcriptome, short RNA analysis, and high-
throughput immunoassay testing. An integrative platform has been de-
veloped to analyse and mine the flow of generated data in the context of NMD. Ariadne MedScan technology has been used to build a literature-
derived biological knowledge base focusing on neuromuscular diseases.

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S.P.40
BIO- NMD: Identifying serum miRNAs as biomarkers for diagnosis and monitoring therapeutic interventions in Duchenne Muscular Dystrophy
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MicroRNAs are small RNA molecules (~22 nt in size) that regulate gene expression at a post-transcriptional level. Several reasons define miR-
NAs as an excellent class of blood-based biomarkers for DMD: miRNAs are present in a stable form in serum and plasma samples; a recent publica-
tion, based on a small group of DMD patents, reported that the level of miR-1, miR-133 and miR-206 in serum correlate with the severity of the disease; the same miRNAs were found abundant in the serum of mdx mice and their level returned to those observed in wild-type mice after exon skip-
ing therapy. Finally, evaluation of miRNAs level in blood-derived prod-
ucts represents a less invasive method compared to their analysis in muscle biopsies. With the aim to validate the finding that the level of specific muscle miRNAs (dystromiRs) in serum correlate with the disease severity and the possibility of utilizing dystromiRs as biomarkers for monitoring the outcome from therapeutic interventions, we are quantifying miR-1, miR-
133, miR-206 and miR-31 in DMD, DMD patients treated with exon skip-
ing therapy (AVI-4658- study 28; Cirak et al., 2011) and controls samples. The data analysis indicate increased levels of miR-1, miR-133a, miR-133b and miR-31 in DMD serum compared to healthy control samples with the largest difference detected for miR-31. We are currently evaluating the lev-
els of these miRNA in serum of AVI treated patients. The preliminary data indicates a difference in some patients for the level of one miRNA between pre- and post-treatment samples and more extensive analysis to confirm this finding are currently being performed. The initial results obtained in this study indicate that dystromiRs have the potential to serve as biomark-
ers for Duchenne muscular dystrophy and also as a marker for assessing the outcome of exon skipping therapies.

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S.P.41
BIO-NMD: Identifying genomic pre-clinical biomarkers for diagnostics and therapeutics of Duchenne muscular dystrophy
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Duchenne muscular dystrophy (DMD) is an inherited disease caused by mutations in the dystrophin gene that disrupt the open reading frame. Despite all muta-
tions abolish the possibility to produce dystrophin protein, there is significant variability in the clinical course and response to corticosteroids in individual patients, suggesting that other factors such as modifying genes play a role in determining the phenotype. In order to find candidate biomarkers for DMD we are performing whole exome sequencing in DMD patients with varying severity defined by the age of loss of ambulation. Five DNA samples from patients on steroids with early loss of ambulation (<8.5 years) and five DNA samples from patients on steroids with late loss of ambulation (>12 years) were sequenced and produced on average 2.8 Gb sequencing data with mean coverage of the sequenced regions of 50×. The identified variants (SNV) with the same genotype between the patients with early loss of ambulation and late loss of ambulation were filtered out and only the exotic non-synonymous SNVs found in three or more patients in each of these two groups were analysed. The preliminary analysis on gene ontology groups using all genes with SNVs in at least three patients in the early loss of ambulation group showed statistical significance for genes involved in extracellular matrix organization (p = 0.0000001), cell adhesion (0.000009) and calcium ion binding (p = 0.0007). For the late loss of ambulation group statistical