Poster for Wednesday 11th Dec

| Presenting Author | Poster Number | Full title of abstract | Authors | Poster Abstract (400 Words) |
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| Eduardo Tizzano<br><br>FundAME; Department of Clinical and Molecular Genetics and Rare Diseases Unit and Medicine Genetics Group, VHIR, Hospital Valle Hebron, Barcelona, Spain | **20** | Reliability of self-reported registries: The experience of FundAME with the Spanish SMA registry. | Tizzano Eduardo, Cattinari Maria, De Lemus Mencia | The Spanish Registry for Spinal Muscular Atrophy (SMA) is an observational longitudinal registry of SMA. It is a voluntary patient reported data-base led by FundAME, the Spanish SMA Patient Organisation. It covers genetically confirmed SMA patients, both 5q and non 5q SMA. Up to 2015, the Spanish Registry was geneticist reported. Difficulties related to technicalities and updating of data made the organisation take the decision to transform it into a patient reported registry. The two main features given to it where: firstly, to ensure that the information it contained was patient relevant and secondly, to ensure its compatibility with the TREAT-NMD recommended dataset.<br>Among the main challenges that a patient registry of a rare disease faces is guaranteeing the reliability of the data, the periodic update, and enrolling a sufficient number of patients. The data needs to be of excellent quality for which itmust be full, consistent, not ambiguous or discrepant, validated and with a thorough curation procedure.<br>A preliminary evaluation of a sample of 66 patients, indicated that in around 50% of the patients' data was suboptimal for one or more items. Some were missing key information, other contained errors in the interpretation of the genetic report or discrepancies between the genetic and the clinical information. This prompted FundAME to change the dynamics of the registry. The most significant change was the improvement of the software, thus allowing the curator to mark the items considered inconsistent: This leads to an automatic notification of the error to the data manager who would then communicate with the patient, to make the necessary corrections. In addition, the wording of some items was reformulated for better comprehension and the communication system between the data manager and the patient was improved. It is important to note that any item not approved by the curator is not considered for statistical analysis.<br>The value of a registry depends mainly on the quality of the data it contains. In the case of a self-reported registry, this data can be fully reliable, as long as it is validated through a rigorous curating system, supported by a professional team, a data manager that communicates with the patient directly and has a versatile IT platform that enables the necessary corrections to be done. |
| Vanessa dos Reis Ferreira<br><br>Head of Patient Advocacy Europe, Switzerland | **33** | Why Respiratory Health in Duchenne Muscular Dystrophy (DMD) matters? Key learnings from a Patient centric activity in the Development and Lifecycle of Medicines led by Santhera Pharmaceuticals | | Introduction:<br>Duchenne Muscular Dystrophy (DMD) is a rare and fatal muscle disease that causes muscle degeneration. Patients and family members with DMD have a unique and challenging journey. DMD patients and carers become the experts on their unique experiences living with DMD condition. They represent an important stakeholder to engage, not only for the companies that develop new therapies, but also for the authorities that assess, regulate and decide which drugs are effective, well tolerated and cost-effective for patients and the community. Patient Engagement (PE) is an expanding area, elevating patients from solely research subjects to active partners along development and lifecycle of medicines. Santhera Pharmaceuticals has developed a systematic PE framework to engage with DMD patient groups.<br>Materials and Methods: An online questionnaire was carried out with DMD Patient Opinion Leaders (POLs). The questionnaire had a total of almost 50 questions focusing on symptoms, management and care of DMD, as well as on the dissemination of information about the disease. It was administered directly to 14 participants from 12 countries. The questionnaire data were analysed using descriptive statistical, with the assistance of the |

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| | | | | software Survey Monkey.<br>Results: Respiratory problems are one of participants' highest priorities for treatment. The results showed that problems associated with Respiratory Function Decline (RFD) have a high impact on patients with DMD as well as on their carers, which increases over time. Wide differences exist concerning the care available to people affected by DMD, and its quality, between and within European countries. In addition, transition from child to adult health services is not yet a standard procedure within the countries in which it is taking place. Great variability exists concerning the age at which children are first tested for respiratory problems, which ranges between 4 and 12 years. This has potential implications for interventions aimed at reducing lung function decline. Finally, people affected by DMD are not sufficiently informed about respiratory problems.<br>Conclusions: Results point out the need to improve the quality of respiratory care for people affected by DMD. The results offer insights that can be used to develop strategies to improve the dissemination of information about RFD associated with DMD and increase access to interventions focused on respiratory function education for people affected by the disease. |
| Marcus Droege,<br><br>United States | **22** | THE RESTORE REGISTRY: A RESOURCE FOR MEASURING AND IMPROVING SPINAL MUSCULAR ATROPHY OUTCOMES | Laurent Servais1, John W. Day2, Darryl C. De Vivo3, Janbernd Kirschner4, Eugenio Mercuri5, Francesco Muntoni6, Perry B. Shieh7, Eduardo Tizzano8, Isabelle Desguerre9, Susana Quijano-Roy10, Kayoko Saito11, Marcus Droege12, Omar Dabbous12, Farid Khan12, Frederick A. Anderson13, Richard S. Finkel14; 1MDUK Oxford Neuromuscular Centre, University of Oxford, Oxford, United Kingdom; 2Department of Neurology, Stanford University Medical Center, Stanford, CA, United States; 3Departments of Neurology and Pediatrics, Columbia University Irving Medical Center, New York, NY, United States; 4Clinic for Neuropediatrics and Muscle Disease, University Medical Center Freiburg, Freiburg, Germany; 5Department of Paediatric Neurology and Nemo Clinical Centre, Catholic University, Rome, Italy; 6Department of Developmental Neuroscience, University College London, London, UK; 7Department of Neurology, David Geffen School of Medicine at UCLA, Los Angeles, CA, United States; 8Department of Clinical and Molecular Genetics, Hospital Valle Hebron, Barcelona, Spain; 9Hôpital Necker Enfants Malades, APHP, Paris, France; 10Garches Neuromuscular Reference Center (GNMH), APHP Raymond Poincare University Hospital (UVSQ), Garches, | INTRODUCTION: Spinal muscular atrophy type 1 (SMA1) is a rapidly progressing, debilitating disease, caused by biallelic survival motor neuron 1 (SMN1) gene deletion/mutation, leading to subsequent motor neuron loss, muscle weakness, respiratory failure, and early death. Dramatic changes in SMA treatment landscape have altered the outlook of this disease. The RESTORE Registry was created to assess outcomes for patients with genetically confirmed SMA, and to provide information to patients, caregivers, regulatory agencies, and researchers on the effectiveness and long-term safety of approved and emerging treatments, document patient survival, and collect information on healthcare resource utilization, caregiver burden, patient functional status, and quality of life.<br>METHODS: The RESTORE Registry is a prospective, multicenter, multinational, observational study. Participating centers include those involved in existing and evolving SMA registries (e.g., iSMAC, TreatNMD, NeuroNEXT, Cure-SMA, SMArtCARE) and individual SMA treatment centers recruited de novo. Data from existing patients enrolled in partnering registries are transferred to the RESTORE Registry database. Data for newly diagnosed patients are added as they enroll. Patients with SMA treated in the onasemnogene abeparvovec (formerly AVXS-101) managed access program are also being enrolled in the registry. Follow-up is 15 years from enrollment or until death, whichever is earlier. Assessments include SMA history and treatment, pulmonary, nutritional, and motor milestones, healthcare resource utilization, work productivity and activity impairment, adverse events, quality of life, caregiver burden, and survival.<br>RESULTS: The RESTORE Registry has been established, and as of August 2019, 17 sites have been activated and 26 SMA patients have been enrolled in the US. Medical centers in Europe are currently being evaluated for activation into the RESTORE Registry, and patients will be enrolled as product becomes available in each country. The RESTORE registry will be expanding globally throughout North America, Latin America, Asia Pacific, Middle East and European countries.<br>CONCLUSION: The RESTORE Registry has begun recruiting recently diagnosed patients with a genetically confirmed diagnosis of SMA, allowing short- and long-term patient outcomes assessment and extended evaluation of emerging SMA treatments, including gene therapy. RESTORE incorporates all types of patients with SMA and follows them through 15 years of standard care, irrespective of treatment. |

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| | | | France; 11Institute of Medical Genetics, Tokyo Women's Medical University, Tokyo, Japan; 12AveXis, Inc., Bannockburn, IL, United States; 13Center for Outcomes Research, Department of Surgery, University of Massachusetts Medical School, Worcester, MA, United States; 14Division of Neurology, Department of Pediatrics, Nemours Children's Hospital, Orlando, FL, United States | |
| Michela Guglieri<br><br>United Kingdom | **38** | TREAT NMD- development of a LGMD global registry | Jordi Diaz Manera, Heather Hilsden, Helen Walker, Hannah Murray, Nathalie Goemens, Craig Campbell, | TREAT-NMD was established in 2007 as an EU funded network of excellence aiming to advance diagnosis, care and therapies for patients with rare neuromuscular disease (NMD). There is growing interest from all stakeholders in the Limb Girdle Muscular Dystrophies, with new therapeutic approaches currently in development and planned clinical trials. Upcoming natural history studies and clinical trials in LGMDs, give us the opportunity to work together with the main experts and stakeholders in the field to align international initiatives, avoid fragmentation and harmonise data collection, learning from the experience in other neuromuscular diseases such as Duchenne muscular dystrophy and Spinal muscular atrophy. With this aims, we have been working on developing a TREAT NMD global LGMD registry. We recognised that it was important as a first step of this process to gain an overview of existing registries and engaging with main stakeholders, with an open and inclusive rather than selective approach. With this aim, we developed an online survey to disseminate through TREAT NMD and other sources to gather information regarding current registries and databases collecting data on LGMDs. The survey asks the following information: contact details of the principal investigator, coordinator(s) and curator(s); type of the registry (patient reported, clinician reported, clinician and patient reported); language used; targeted population (paediatric and/or adult); types of LGMDs included in the registry and number of patients registered for each sub-type; inclusion of any other neuromuscular disease; ethical approval and inclusion criteria (e.g. genetic versus clinical diagnosis). The possibility to upload the dataset collected and copy of the consent form is given.<br>The results of the survey will be used to establish an international working group, as part of the TREAT NMD LGMD task, to work through reaching consensus on core data set for LGMD registry. Engagement with pharmaceutical companies developing therapeutic strategies for LGMD is ongoing to discuss funding opportunities to support the registries, pilot the core data set and explore options for universal registry platform for LGMD. |
| Melinda Gyenge<br><br>France | **19** | The DM-Scope registry: an innovative framework for Myotonic Dystrophy translational research | M. De Antonio (AP-HP, France), D. Hamroun (CHU Montpellier, France), M. Gyenge (AP-HP, France), B. Eymard (AP-HP, France), J. Puymirat (CHU Quebec, Canada), C. Gagnon (CIUSSS Saguenay-Lac-St-Jean, Canada), F. Myotonic Dystrophy Study group (AP-HP, France), G. Bassez (AP-HP, France) | The relevance of registries as a key component for developing clinical research for rare diseases (RD) has been acknowledged by stakeholders. However, remaining limitations exist. We developed the innovative concept of the DM-Scope registry aiming to promote translational research and care management for Myotonic Dystrophy (DM), a prototypical example of a highly heterogeneous RD. Study objectives were (1) to improve data quality and value; (2) to limit the amount of incomplete data; (3) to improve standardization and data comparability. The revised dataset includes social-demographic data, clinical features, genotype, biomaterial data, and is adjustable for clinical trial data collection. In France, the registry has a nationwide coverage, composed of 55 neuromuscular centres, encompassing the whole clinical and genetic disease spectrum. This widely used platform gathers almost 3000 DM patients (DM1 n=2828, DM2 n=142), in both pediatric (n=322) and adult (n=2648) centres, which accounts for >20% of overall registered DM patients internationally. The registry supported 10 research studies of varied methodology i.e. observational, basic science, interventional studies, and dramatically accelerated patient recruitment for clinical trials. DM-Scope represents the largest collection of standardized data for the DM population so far. Our concept has improved collaboration among healthcare |

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| | | | | professionals by continuously providing tools to monitor cohorts in participating centres. This registry proves to be a powerful device for promoting both research and medical care and is applicable to other countries. In the context of emerging therapies, such an integrated platform contributes to the standardization of DM research and may optimize international multicentre clinical trials. |
| Aynur Ayşe Karaduman<br><br>Turkey | **32** | Pediatric Neuromuscular Diseases Patient Registry System (KUKAS) at Turkey | Ayşe Karaduman, Numan Bulut, İpek Gürbüz, Güllü Aydın, Öznur Yılmaz | Objective: The Pediatric Neuromuscular Diseases Registry System (KUKAS), supported by Association Française contre les Myopathies (AFM) in 2011, was created as a project which aimed to define the profile of Pediatric Neuromuscular Diseases in Turkey, create database, inform families and health professionals, participate in international researches, represent our country, and enable professionals to benefit academically and clinically.<br><br>Method: In January 2011, web page and software for the registry were developed. As the first step of the registry Duchenne Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA) patients were involved and registered to the online system. Then, system has been developed to contain all pediatric neuromuscular diseases with the addition of disease-specific tests to the system, and the information and evaluation data of these patients were recorded in the database. In addition, the interface was added to website for the patient interaction and allowed patients to register themselves to system. Up-to-date information has been provided regularly for health professionals and families of the patients through the web page. SMA Core Data Set was updated in May 2019.<br><br>Results: Two thousand three hundred sixty-five patients and their 6738 assessment data were registered to the system from 2008 to 17th September 2019. The diagnosis of patients recorded to the system were as follows; 1215 DMD, 513 SMA, 120 neuropathies, 91 myopathies, 83 congenital muscular dystrophies, 46 undiagnosed patients and 297 other pediatric neuromuscular diseases. 61.35% of the patients had genetic diagnosis and 25.07% had biopsy results. Up to now, 916 of these patients have reached their own data and updated them by using the patient interaction interface of the website. A total of 15 patients were recruited according to the SMA Expanded Data Set.<br><br>Discussion: The development of questionnaires to record to the system for screening of different types of neuromuscular diseases and the addition of patients up-to-date data have been continuing. We are also developing our cooperation with other national centers. This project has enabled us to be a national and international reference center for pediatric neuromuscular diseases. Updating of DMD Expanded Data Set is under construction. |
| Lindsay Murphy<br><br>United Kingdom | **16** | Global FKRP Registry – a research database for Limb Girdle Muscular Dystrophy R9 (2I) | Lindsay Murphy, Jean-Pierre Laurent, Katherine Mathews, Herb Stevenson, Simone Thiele, John Vissing, Maggie Walter, Lacey Woods, Volker Straub | The Global FKRP Registry is an international registry for individuals with conditions caused by mutation of the Fukutin-Related Protein (FKRP) gene: limb girdle muscular dystrophy R9 (LGMD R9, formerly named LGMD 2I) and the congenital muscular dystrophies MDC1C, Muscle-Eye-Brain Disease and Walker-Warburg Syndrome. The registry seeks to further understanding of the natural history and prevalence of FKRP-related muscular dystrophies (MD). It adheres to the Charter for TREAT-NMD patient registries and contributes to the TREAT-NMD Global Database Oversight Committee (TGDOC).<br>The purpose of the registry is to aid the rapid identification of eligible patients for clinical studies. It disseminates FKRP-relevant information to participants; provides a source of information to academics, industry and healthcare professionals; and supports the FKRP community.<br>Registration is patient-initiated through a secure online portal. Participants give their informed consent and are invited to complete a questionnaire about their condition. Data is reported by both patients and their healthcare professionals and includes: age of onset, presenting symptoms, family history, motor function and muscle strength, respiratory and cardiac function, medication, in addition to information on patient quality of life and pain.<br>Currently, 733 patients (54% female, 46% male) are registered with the Global FKRP Registry, with an age range |

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| | | | | of 1 to 80 years. Registrations are from 43 countries, with greatest numbers from USA (28%), Germany (21%) and UK (11%). Diagnoses are reported as LGMD R9 (86%), MDC1C (2%), other FKRP-related MD (1%), unspecified (11%). Seventy-two percent of patients are reported as being ambulant, 23% as non-ambulant and 5% as unspecified. The FKRP gene mutations reported within the registry are: 65% homozygous for the common mutation (c.826C>A), 28% heterozygous for the common mutation, 5% heterozygous with two unique mutations and 2% homozygous with a unique mutation which is not the common mutation.<br>The Global FKRP Registry is a valuable tool for the collection of patient data which informs academics, healthcare professionals and industry. The core objectives of the registry of patient data collection and assembly of a trial-ready patient cohort are now being realised, demonstrated by recent academic and industry interest in the registry to facilitate patient recruitment. As knowledge of rare neuromuscular conditions increases and advances in the development of potential therapies are made, the Global FKRP Registry is centrally placed to help support the accumulation of natural history data and facilitate recruitment to study potential therapies. |
| Nicole O'Connor<br><br>Netherlands | **21** | Improving Patient Care in Neuromuscular Disease – the TREAT-NMD Education Programmes in Duchenne Muscular Dystrophy (DMD) and Spinal Muscular Atrophy (SMA) | Annemieke Aartsma-Rus, Andoni Urtizberea, Cathy Turner, Clare Bradley, Elizabeth Vroom, Jorge alfredo Bevilacqua, Nathalie Goemans, Nicole O'Connor | Background: As the development of therapies for DMD and SMA continues to accelerate and our understanding of the highest standards of care increases further, there is a need to keep the neuromuscular community informed and educated about the diseases and the very latest treatments, care, knowledge and consensus.<br>Study aims: This project aims to improve life expectancy and quality of life of patients, ensuring information on standards of care are more widely disseminated to health care providers by offering equal learning opportunities, regardless of geographical location. In addition, globally agreed and widely implemented standards of care allow more effective development and delivery of clinical trials.<br>Methods: The TREAT-NMD network has established an Education Committee (TEC) which plans and oversees educational programmes in DMD and SMA. Central to the programmes are masterclasses aimed at researchers and health care providers who work with patients living with SMA and DMD. Masterclasses deliver expert-led medical education with focus on providing multi-disciplinary content, including: current and emerging therapeutics; diagnosis and natural history; standards of care; patient involvement; access to approved treatments. Funding for the programmes comes from several sources including patient organisations and the pharmaceutical industry (via unrestricted education grant) and covers the costs for attendees who are accepted onto the masterclasses. This allows those with little access to their own funding to benefit. Attendees are asked to say how they will cascade their learning to colleagues in order to maximise the benefit to the patient community. Priority is given to geographical regions and centres where the need is greatest.<br>Key findings: Feedback from masterclasses so far has been overwhelmingly positive. The challenges of implementing the latest and highest standards of care, best routes to diagnosis and access to emerging treatments in different healthcare settings are addressed through this project. We have seen that this is a valuable opportunity for practitioners to learn from each other as well as the key opinion leaders.<br>Conclusions and impact: The need to provide education and training for healthcare providers in this rapidly changing field is clear. Standards of care are needed across the world in order to provide patients with the very best quality of life and to ensure that the community is 'trial-ready'. The success of the projects so far mean that the TEC plans to consider other neuromuscular disease areas in the future. |
| Avril Palmeri<br><br>United Kingdom | **31** | Share4Rare: a collaborative platform for rare diseases | Palmeri A, Hernandez Ortega S, Nafria Escalera B, Perera A, Chapi I, Le Corvec A, Brooke N, Ryll B, Athanasiou D, S4R Consortium | The Share4Rare (https://www.share4rare.org/) platform is a safe space for individuals and families living with a rare disease to connect, share knowledge and get involved in scientific research.<br><br>Share4Rare is a collaborative project where rare disease patients and carers bring their unique perspective and expertise to guide the design, functionality and content of the platform. They are also the driving force behind the Share4Rare online community. Share4Rare combines the collective intelligence of the rare disease |

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| | | | | community with the latest technology to advance the cause of rare diseases.<br><br>The Share4Rare platform provides public resources for individuals and families living with a rare disease. Patients and carers form editorial boards and provide oversight for medical chapters written by clinical experts. For neuromuscular diseases, this collaborative approach includes reaching out to, and linking to, other projects and initiatives, including TREAT-NMD and EURO-NMD.<br><br>Adult patients and carers can register to access the secure online platform where they can connect to others affected by the same disease, or experiencing similar symptoms. Based on the symptoms provided during the registration process, Share4Rare will connect them in the first instance to users of the platform who are most like them. They will have the opportunity to ask and answer questions in a global forum. They will also be able to opt in to the direct messaging system with other users, with priority given to those who are most like them.<br><br>The final goal of the project is to advance research into rare diseases. Aggregated data collected from patients will allow respondents to see how their experience of living with a rare disease compares with other users of the platform. It will also provide an opportunity for researchers to connect with the Share4Rare community and specific patients involved in research projects (neuromuscular conditions and paediatric rare tumours). |
| Ben Porter<br><br>United Kingdom | **30** | The UK Myotonic Dystrophy Patient Registry: An Essential Tool in the Facilitation of Translational and Clinical Research | Ben Porter, Phillip Cammish, Chris Turner, Emma Heslop, Darren Monckton, David-Hilton Jones, Margaret Bowler, Margaret Phillips, Mark Roberts, Mark Rogers, Michael Rose, Jacqueline Donachie, Chiara Marini-Bettolo | Objective<br>The UK Myotonic Dystrophy Patient Registry is a patient self-enrolling online database collecting clinical and genetic information about myotonic dystrophy type 1 (DM1) and myotonic dystrophy type 2 (DM2). The registry aims to; facilitate academic and clinical research, better characterise and understand DM, and disseminate information relating to upcoming studies and research advancements.<br><br>Methodology<br>The registry is used to capture longitudinal, self-reported data through an online portal available to patients and clinicians. Where specialised clinical or genetic information is required, the neuromuscular specialist involved in the participant's care can be invited to provide some additional information. The participant is able to select a health care provider from a pre-populated list at registration stage.<br><br>Results<br>Between May 2012 and September 2019, 745 participants registered with the UK DM Patient Registry. On average, 6 new participants register each month. Regarding confirmed clinical diagnosis, 96.7% participants have DM1 and 3.3% have DM2. Overall, 44.4% have genetic confirmation of their condition and the most commonly reported symptoms were fatigue (78%) and myotonia (75%), with 65% of participants reporting both.<br><br>Conclusions<br>The registry has previously supported approximately 17 registry enquiries including; the OPTIMISTIC clinical trial, PHENO-DM1 and the AMO Pharma phase II clinical trial of tideglusib. In the past six months, the registry has also helped facilitate two academic studies, the PREFER study, a clinical ventilation survey and the creation of a dysphagia research advisory group. The registry is an example of an online-based, cost-effective, and patient-driven tool that has been successful in assisting with recruitment for a number of academic and clinical studies since its launch in 2012. |
| Cathy Turner | **5** | The TREAT-NMD Advisory Committee for | Cathy Turner, Volker Straub, Joanne Lee, Annamaria De Luca, Cristina Csimma | Background: Established in 2010 as part of TREAT-NMD, TACT is an expert multidisciplinary expert group that provides the neuromuscular community with independent, confidential guidance on the evaluation of therapeutic development programs (whether novel or repurposed) for rare neuromuscular diseases. TACT is a |

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| United Kingdom | | Therapeutics: A multi-disciplinary expert group to guide neuromuscular drug development | | not-for-profit organization.<br>Aims: The goal of each TACT review is to position the potential therapy along a well-informed, realistic development pathway to clinical development, and eventual registration, by evaluating supporting preclinical data and other critical drug development considerations necessary for objective decision-making. The output is a detailed report with recommendations, leading to the improved non-clinical and clinical study design and conduct to enable generating meaningful data supporting clear Go-No Go decision and the increased potential of longer term funding.<br>Methods: Applications are reviewed by a bespoke panel of world-leading multidisciplinary drug development experts drawn from the TACT committee of around 65 members in response to the specific needs of a particular application. All reviews are conducted under strict confidentiality agreements and conflict declarations. An online and thorough in person review committee review process is followed by a face-to-face meeting between the reviewers and the applicant. A confidential report detailing the committee's recommendations is provided to the applicant within 4-6 weeks after the meeting. TACT conducts two review cycles each year, considering up to four applications each time. Reviews are independent of any funding stream, reviewers are independent, under no conflict of interest, and not compensated for their time other than receipt of a small honorarium.<br>Results: To date TACT has held 20 review meetings, 11 in Europe and 9 in the North America, and has reviewed 59 applications from both academic investigators (32%) and industry (68%). TACT has also worked closely with existing infrastructures established by the National Institute of Health (NIH) and the European Union, and with major patient organizations in multiple regions around the world to ensure the process and guidance reflects the patient perspective and assists the entire neuromuscular community.<br>Conclusion: Feedback shows that TACT has generated recommendations that have greatly helped academic investigators and industry developers, in evaluating and prioritizing their -programs. It has strongly encouraged applicants to consider their development programs in a methodical fashion with clearly actionable steps and decision making and with optimal use of funding and resources. |
| Ivan A Yakovlev<br><br>Russia | **34** | Russian Dysferlinopathy patients register | Ivan A. Yakovlev[1,3,7], Sergei N. Bardakov[2], Mikhail O. Mavlikeev[3], Olga N. Chernova[3], Pierre G. Carlier[4, 5], Andoni J. Urtizberea[6], Artur A. Isaev[1], Roman V. Deev[1,7,8] | Dysferlinopathies are one of the most common forms of girdle-limb muscular dystrophies in the Russian Federation. 54 patients were registered, 29 with the LGMDR2 form (54%) and 25 with the Miyoshi distal form (46%). About one third (35%) of the patients live in isolated mountainous regions of the Russian Federation. Mutations in all patients were confirmed by a new generation sequencing. Registered patients annually undergo clinical assessment, quantitative MRI of muscles (analysis of the fraction of fat and water T2), cardiac function (echocardiography, ECG, cardiac MRI), immunological status (level of cytokines, lymphocyte subpopulations, functional activity of neutrophils/macrophages), laboratory markers of muscular-dystrophic process. There are very rare phenotypes with pronounced contractures of the finger's flexors, Achilles tendons and toes extensors (4/54 cases) among the identified cases of dysferlinopathy. Other specific features of the patients presented in our registry are an early confinement to wheelchair at 47 [45,3-51,1] years and a large number of cases of homozygous mutations in the DYSF c.TG573 / 574AT gene; p. Val67Asp, due to the founder's effect and the high incidence consanguinity. |