ePosters Schedule

Wednesday 6 July, 2022

Wednesday, Screen 1

Chair: Stefano Previtali

eP01.01.01
Genetic, Proteomic and Morphological Characterisation of Two Children With Novel Nonsense Mutations of BVES (POPDC1)
Anne Schänzer

eP01.01.02
Cardiac Analysis Reveals Morphological Alterations in an Intermediate Mouse Model of Spinal Muscular Atrophy
Nithya Nair

eP01.01.03
Respiratory Follow-Up In Children With Spinal Muscular Atrophy - A Descriptive Study
Inês Pereira

eP01.01.04
Worldwide Prevalence of Home Mechanical Ventilation in Neuromuscular Disorders
Michel Toussaint

eP01.01.05
De Novo and Dominantly Inherited SPTAN1 Mutations Cause Spastic Paraplegia and Cerebellar Ataxia
Jonathan De Winter

eP01.01.06
Use of NGS for Diagnosis of Asymptomatic Hyperckemia in Childhood
Pilar Marti

eP01.01.07
Prevalence of Titinopathy in India
Aishwarya Dhall

eP01.01.08
Optimisation of a Cell-Based Strategy for Rapid Evaluation of Compounds in Myotonic Dystrophy Type I
Andrea López-Martínez

eP01.01.09
Spectrum of Muscular Dystrophies in India
Mehar Chand Sharma

eP01.01.10
Median Nerve Ultrasound in Carpal Tunnel Syndrome
Imen Kacem

Wednesday, Screen 2

Chair: Maike Dohrn

eP01.02.01
Clinical Features and the Novel p.K357E Mutation in a Cohort of Patients With mfn2-Related Neuropathy
Elena Abati

eP01.02.02
Patient-Reported Symptom Burden of Charcot-Marie-Tooth Disease Type 1A (CMT1A): Findings from a Real-World Digital Study
Youcef Boutalbi

eP01.02.03
One Cause, Many Courses: Leveraging Whole-Genome Sequencing for Comprehensive Modifier Studies in CMT1A
Maike Dohrn

eP01.02.04
Work Impacts in Charcot-Marie-Tooth Disease Type 1A (CMT1A): Findings from a Real-World Digital Study
Youcef Boutalbi

eP01.02.05
R298C LMNA Mutation Can Cause either Peripheral Neuropathy, Cardiomyopathy or Both: A Case Series Study
Leila Tamaoui

ePosters Schedule

eP01.02.06
Overlap Between Hereditary Sensory-Motor Neuropathy (HSMN) And Chronic Inflammatory Demyelinating Polyradiculoneuropathy (CIDP)
Manon Hustinx

eP01.02.07
Plasma and Skin Biomarkers for Charcot Marie Tooth Disease
Michael Shy

eP01.02.08
Depression in Patients with Charcot-Marie-Tooth Disease Type 1A (CMT1A): Findings from a Real-World Digital Study
Youcef Boutalbi

eP01.02.09
Novel Variant in the Stalk Domain of KIF5A in a Patient With CMT2-Like Phenotype
Eleni Liouta

Wednesday, Screen 3

Chair: James Russell

eP01.03.01
Deacetylation of E3 Ubiquitin Ligase NEDD4-1 by Sirtuin1 Regulates Axonal Growth and Treats Diabetic Neuropathy
James Russell

eP01.03.02
CuidAME Registry: Using Process Automation and Machine Learning Technology to Build SMA Data Analytics Repository
Sonia Segovia

eP01.03.03
Spinal Muscular Atrophy Disease Registries: Overview and Recent Progress
Stephanie Raynaud

eP01.03.04
Cognitive Assessment of Spinal Muscular Atrophy
Sanae Akodad

eP01.03.05
SMA-REACH-UK, Adult-SMA-REACH and UK SMA-Patient-Registry an Integrated Model: Transition of Data and Longitudinal Data Collection.
Sonia Segovia

eP01.03.06
Proposal of New Functional Motor Scale to Evaluate Muscle Fatigue in Adult SMA Patients
Gabriele Siciliano

eP01.03.07
CuidAME: A New Registry for Longitudinal Data Collection of Spanish SMA Patients
Sonia Segovia

eP01.03.08
Swallowing Evaluation in Treated SMA patients – A Pilot Prospective Study
Nicolas Deconinck

eP01.03.09
Safety and Effectiveness of Onasemnogene Abeparvovec Alone or with Other Disease-Modifying Therapies: Findings from RESTORE
Laurent Servais

eP01.03.10
Brown-Vialetto-Van Laere Syndrome, Temporary Clinical Stabilization with Intravenous Immunoglobulin (IVIg) and MRI Abnormalities
Edna Julieth Bobadilla

eP01.03.11
A Compound Heterozygous Mutationin Calpain-1 Identifies a New Gene for Spinal Muscular Atrophytype-4
Gonzalo Perez Siles

eP04.05.01
Improved Quality of Life in Patients with X-Linked Myotubular Myopathy (XLMTM) Treated with Resamirigene Bilparvovec
Laurent Servais

ePosters Schedule

Wednesday, Screen 4

Chair: Anthony Amato

eP01.04.01
COVID-19: Retrospective Analysis in Neuromuscular Disease Patient's Impact on Healthcare, Quality of Life and Anxiety
Laura Buscemi

eP01.04.02
COVID-19-Related Neuropathy in Colombia: The Experience During the First 23 Months Of Pandemic
Laura Peña Guzmán

eP01.04.03
Myositis And Myocarditis With Anti-Jo-1 Antibodies Following SARS-CoV-2 mRNA Vaccination Or COVID-19 Infection
Myrthe Willems

eP01.04.04
Guillain-Barre Syndrome in 220 Patients with COVID-19
Josef Finsterer

eP01.04.05
IVIg Treatment in Chronic Inflammatory Neuropathies During the COIVD19 Pandemic
JUDYTA Barańska

eP01.04.06
Quality of Life of Myasthenia Gravis Patients During COVID-19 Pandemic – One Year Follow Up
Aleksandar Stojanov

eP01.04.07
COVID Spike Antibodies in Neuromuscular Conditions: A KU Experience
Mamatha Pasnoor

eP01.04.08
Parsonage-Turner Syndrome after COVID-19 Vaccination
Elisabeth Van Boxstael

eP01.04.09
Multi-Centre Study to Assess the Safety of Alglucosidasi and of Laronidasi in Home Infusion Setting
Maurizio Scarpa

eP01.04.10
Clinical Course of Four Neuromuscular Disease Patients Infected COVID-19
Akiko Ishii

Wednesday, Screen 5

Chair: Anneke van der Kooi

eP01.05.01
Muscle Inflammation Drives Mitochondrial Dysfunction in Inclusion Body Myositis
Stefanie Meyer

eP01.05.02
B Cell Receptor Profiling before and after IVIg Treatment in Idiopathic Inflammatory Myopathies
Anneke van der Kooi

eP01.05.03
Multi-Muscle Pathology Assessment in Inclusion Body Myositis: Post-mortem Study in Two Cases
Stefanie Glaubitz

eP01.05.04
Design of a Global Phase 2/3 Randomized, Placebo-Controlled Trial of Ravulizumab in Adult Dermatomyositis
Rohit Aggarwal

eP01.05.05
A Prospective Diagnostic Accuracy Study of Multi-Modality Testing in Patients Suspected of a Treatable IIM
Renske Kamperman

ePosters Schedule

eP01.05.06
Fibroblast Model Unveils New Molecular Insights in Inclusion Body Myositis
Judith Cantó-Santos

eP01.05.07
Prevalence and Clinical Correlation of Myositis-Specific and Myositis-Associated Autoantibodies in Indian Myositis Cohort
Bandana Jassal

eP01.05.08
Immune-Mediated Necrotizing Myopathy: An Emerging Disorder
Sofía Portela Sánchez

eP01.05.09
COVID-19 and Vaccination Against Sars-CoV-2 in Patients With Myasthenia Gravis From Belgrade, Serbia
Stojan Peric

Wednesday, Screen 6

Chair: Volker Straub

eP01.06.01
Two-Years Prospective Natural History Study in 24 Adult LGMDR12 Patients: Clinical and Radiological Outcome Measures
Bram De Wel

eP01.06.02
Assessing the Relationship of Patient Reported Outcome Measures with Functional Status in Dysferlinopathy
Heather Hilsden

eP01.06.03
Clinical and Genetic Features in Two Families Carrying Novel and Reported DYSF Variants
Birute Burnyte

eP01.06.04
Description of Motor Function in Duchenne Muscular Dystrophy in a Center of Expertise in Colombia
Sandra Castellar

eP01.06.05
Preliminary Results from MLB-01-003: An Open Label Phase 2 Study of BBP-418 in LGMD2I
Douglas Sproule

eP01.06.06
The Founder Mutation TRAPPC11 c.1287+5G>A is a Frequent Cause of Limb-girdle Muscular Dystrophy in Roma Population
Carlos Ortez

eP01.06.07
A Familial Case with Phenotypic Differences in a CAV3 Pathogenic Variant
Seung-ah Lee

eP01.06.09
Molecular Diagnosis of Muscular Dystrophy Using the LGMD Gene Panel in Adult Neurology
Seok-Jin Choi

eP01.06.10
Observational Study: the Quality of Life in Patients with Alpha-Sarcoglycan, Beta-Sarcoglycan and Gamma-Sarcoglycan Gene Mutation
Serena Paniga

ePosters Schedule

Thursday 7 July, 2022

Thursday, Screen 1

Chair: Sabrina Sacconi

eP02.01.01
Value of Muscle Ultrasound in Pediatric Neuromuscular Patients
Hanna Kuepper

eP02.01.02
Place of Muscle Ultrasound in the Diagnosis and Follow-up of Pediatricneuromuscular Diseases
Susana Quijano-Roy

eP02.01.03
Machine Learning in Ultrasound-Guided Differentiation of Myopathic From Neurogenic Patterns: A Pilot Study
Elisabeth Chroni

eP02.01.04
Novel Titin Mutation Responsible for Hereditary Myopathy With Early Respiratory Failure in Adult Moroccan Man
Leila Tamaoui

eP02.01.05
Deep Learning-Based Electrodiagnosis of Needle-Electromyography
Ilhan Yoo

eP02.01.06
Normative Values for Commonly Used Nerve Conduction Studies in Russian Population
Maria Kovalchuk

eP02.01.07
Cardiac MRI in Duchenne and Becker Muscular Dystrophy
Manu S G

eP02.01.08
Artificial Intelligence Based Automatic Muscle MRI Segmentation: Towards a Generalized Solution for Quantitative Imaging
Eduard Snezhko

eP02.01.09
Neuropathies Amidst the Pandemic: Remote Phenotype Validation and Assessment of Patient Needs
Maike Dohrn

eP02.01.10
Subclinical Status of Dysferlinopathy
Sergey Bardakov

Thursday, Screen 2

Chair: Imem Kacem

eP02.02.01
Overview of Patients With Chronic Inflammatory Demyelinating Polyneuropathy of the Neuromuscular Reference Center of LIèGE
Margaux Poleur

eP02.02.02
Intravenous Immunoglobulin Therapy in Patients with Chronic Inflammatory Demyelinating Polyneuropathy: A Systematic Literature Review
Colin Anderson-Smits

eP02.02.03
Chronic Inflammatory Demyelinating Polyradiculoneuropathy in Patients in Diabetic and Non-Diabetic Patients: A Comparative Study
Imen Kacem

eP02.02.04
Identical Late Responses in Early Stages of Guillain-Barré Syndrome: A-Waves or Repeater F-Waves
Dimitra Veltsista

eP02.02.05
An Unusual Clinical Evolution in Anti-contactin-1 Positive CIDP
Yann Vivier

ePosters Schedule

Thursday 7 July, 2022

eP02.02.07
Characteristics and Epidemiology of Patients with Multifocal Motor Neuropathy in Latvia
Marija Roddate

eP02.02.08
Herpes Zoster May Be a Trigger for Lumbosacral Radiculoplexus Neuropathy
Catarina Aragon Pinto

eP02.02.09
Acute Worsening of Anti-mag Neuropathy Following Treatment With Rituximab
Suraj Muley

eP02.02.10
Clinical and Prognostic Characteristics of Guillain-Barre Syndrome Associated With COVID-19, Is This Coincidental?
Behnaz Ansari

Thursday, Screen 3

Chair: Wolfgang Loscher

eP02.03.01
Risdiplam in Children With Spinal Muscular Atrophy: Real-World Experience After One Year of Treatment
Marta Gomez Garcia De La Banda

eP02.03.02
Cost-Utility Analysis of Risdiplam Compared with Onasemnogene Abeparvovec in Spinal Muscular Atrophy Type 1
Matthias Bischof

eP02.03.03
SAPPHIRE: Efficacy and Safety of Apitegromab in Later-Onset SMA; Phase 3 Trial in Progress
Thomas O. Crawford

eP02.03.04
Safety of Onasemnogene Abeparvovec in Patients With SMA in Real Clinical Practice
Kristina Nevmerzhitskaya

eP02.03.05
Treatment of Spinal Muscular Atrophy with Onasemnogene Abeparvovec in Switzerland
Georg M. Stettner

Thursday, Screen 4

Chair: TBC

eP02.04.01
Tocilizumab in Therapy Myasthenia Gravis Patients With COVID-19
Vesna Martic

eP02.04.02
Myasthenia Gravis Demographics Re-Visited
Nanna Witting

eP02.04.03
Incidence of Skin Changes in Patients with Myasthenia Gravis Prescribed Mycophenolate or Azathioprine
Anahit Mehrabyan

eP02.04.04
Quality of Life in Myasthenia Gravis and Correlation of MG-QOL15 With Other Functional Scales
Laura Porras

eP02.04.05
Barriers and Facilitators to Exercise in Auto-Immune Myasthenia Gravis
Simone Birnbaum

eP02.04.06
Clinical Characteristics of Patients With Seronegative Myasthenia Gravis
Rebecca Kjær Andersen

ePosters Schedule

eP02.04.07
TREAT-NMD Global Registry Network: Facilitating 12 Years of Neuromuscular Drug Development
Cathy Turner

eP02.04.08
Immunofluorescence Signal Intensity Measurements as a Semi-quantitative Tool to Assess Sarcoglycan Expression in Muscle Biopsy
Simona Zanotti

eP02.04.09
Three Cases of Congenital Fibre Disproportion With Etiology Other Than Congenital Myopathy
Emna Farhat

Thursday, Screen 5

Chair: Kristl Claeys

eP02.05.01
One-year ENDEAVOR Data (Ambulatory, ≥4 to <8-year-olds): Phase 1b Trial of Delandistrogene Moxeparvovec in DMD
Craig Zaidman

eP02.05.02
Whole Genome Sequencing in a Pair of Duchenne Muscular Dystrophy Siblings with Discordant Cognitive Phenotype
Luca Bello

eP02.05.03
Growth Patterns and Loss of Ambulation in Boys with Duchenne Muscular Dystrophy (DMD)
Georgia Stimpson

eP02.05.04
An Alu-Mediated Insertion in the DMD Gene Canceled Out by Exon 15 Splicing
Alberto Budillon

eP02.05.05
FU-5Cv: Inducible Muscle-Specific Downregulation of Utrophin in Dystrophic Mice to Better Mimic Duchenne Muscular Dystrophy
Laurence Anne Neff

eP02.05.06
Preclinical Assessment of Therapeutic Cocktails in Dystrophic Mice: Tamoxifen Combined to Metformin, Citrulline and Steroids
Laurence Anne Neff

eP02.05.07
Roles of Ltbp4 and Abcc6 on the Phenotype of Mouse Models of Duchenne Muscular Dystrophy
Laurence Anne Neff

eP02.05.08
Integrated Analyses of Data from Clinical Trials of Delandistrogene Moxeparvovec in DMD
Stefanie Mason

eP02.05.09
Phenotypic Variability of Becker Muscular Dystrophy: A Detailed Clinical Characterization Protocol Towards Trial Readiness
Gabrielle Siciliano

eP02.05.10
Non-neuromuscular Manifestations in a Colombian Patient with Megaconial Congenital Muscular Dystrophy
Edna Julieth Bobadilla

Thursday, Screen 6

Chair: Stojan Peric

eP02.06.01
DYNE-101 Corrects the DM1 Splicing Phenotype of hTfR1/DMSXL Mice and Is Well Tolerated in NHPs
Aaron Novack

eP02.06.02
Myotonic Dystrophy Type 2 and Autoimmune Diseases
Stojan Peric

eP02.06.03
Nuclear Envelope Dysfunction in Myotonic Dystrophy Type 1
Sandra Rebelo

eP02.06.04
Does Small Fiber Neuropathy Contribute to Chronic Muscle Pain in Patients with Myotonic Dystrophy?
Viviane Chantal Schmitt

eP02.06.05
Frequently Used Outcome Measures to Evaluate Muscle Strength in Patients with Myotonic Dystrophy Type 1
Sandra Rebelo

eP02.06.06
Clinical Features of the UK Myotonic Dystrophy Patient Registry
Helen Walker

eP02.06.07
Myofibrillar Myopathy: Clinico-Genetic Spectrum from an Indian Neuromuscular Center
Abel Thomas Oommen

eP02.06.08
Muscular Dystrophies Due to Collagen VI Mutations
Joana Coelho

ePosters Schedule

Friday 8 July, 2022

Friday, Screen 1

Chair: Marianne de Visser

eP03.01.01
Validation of the Individualized Neuromuscular Quality of Life Questionnaire in Korean Patients with Neuromuscular Diseases
Seung-ah Lee

eP03.01.02
Improving Clinical Trials in Neuromuscular Diseases: The TREAT-NMD Advisory Committee for Therapeutics (TACT)
Cathy Turner

eP03.01.03
Impact of Independence in Mobility on Independence Outdoors in People With Neuromuscular Conditions
Karen Wong

eP03.01.04
Neuropsychological and Behavioral Profile in a Cohort of Becker Muscular Dystrophy Pediatric Patients
Michele Tosi

eP03.01.05
Evolution of Gait Parameters and Influence of Orthotics on Gait in a Patient With Dysferlinopathy
Meredith James

eP03.01.06
Predictors of Gait in Patients with Late-Onset Pompe disease
Théo Maulet

eP03.01.07
Simplified and Non-invasive Optical Motion Tracking of Respiratory Dynamics in the MDX Mouse Model.
Jana Zschüntzsch

eP03.01.08
Serum Neurofilament Light Chain as a Marker of Nervous System Damage in Myopathies
Jochen Schaefer

eP03.01.09
Troponin T in Spinal and Bulbar Muscular Atrophy
Luca Bello

eP03.01.10
Urinary Titin as a Biomarker of Myotonic Dystrophy Type 1
Endre Pál

eP03.01.11
InGene 2.0: An Intelligent Technological Approach to Genotype-Phenotype Relationship Study in Rare Neuromuscular Diseases
Gabriele Siciliano

Friday, Screen 2

Chair: Edyardo Nobile Orazio

eP03.02.01
Coexistence of Charcot-Marie-Tooth Disease and Chronic Inflammatory Demyelinating Polyradiculoneuropathy
Zoran Vukojevic

eP03.02.02
HINT1-Related Autosomal Recessive Axonal Neuropathy With Neuromyotonia (ARAN-NM). Presentation of a Greek Pedigree
Maria Moschou

eP03.02.03
The Importance of Functional Assessment In Hereditary Sensory and Autonomic Neuropathy
Luisa Fernanda Castaño Herrera

ePosters Schedule

ePO3.02.04
Dose-Exposure-Efficacy Response Relationships for Intravenous Immunoglobulin,10% in Patients with Multifocal Motor Neuropathy
Zhaoyang Li

ePO3.02.05
Long-Term Course of Chronic Inflammatory Demyelinating Polyneuropathy: Clinical and Neurophysiological Outcomes
Evgeniya Melnik

ePO3.02.06
Hematological Effects of Intravenous Immunoglobulin Therapy in Patients with Neuromuscular Diseases – A Retrospective Analysis
Pieter Olivier

ePO3.02.07
Man-In-The-Barrel Syndrome as a Manifestation of Multiple Myeloma Relapsed
Dionis Vallejo

ePO3.02.08
Sensory Polyneuropathy Associated With Vitamin D Deficiency
Sa-yoon Kang

ePO3.02.09
Diagnosing Small Fiber Neuropathy Remains Challenging in Sarcoidosis - Preliminary Data
Lisette Raasing

ePO3.02.11
In Vitro Comparison Between Different 10% Intravenous Immunoglobulin Preparations
Rabye Ouaja

Friday 8 July, 2022

Friday, Screen 3

Chair: TBC

ePO3.03.01
MANATEE: GYM329 (RO7204239) in Combination with Risdiplam Treatment in Ambulant Children with Spinal Muscular Atrophy
Francesco Muntoni

ePO3.03.02
Progression ofthe Revised Hammersmith Scale items in patients with Spinal Muscular Atrophy treated with Nusinersen
Sandra Castellar

ePO3.03.03
Critical Review of the Spanish Pharmacoclinic Protocol for the Monitoring of Nusinersen Treatment
Laura Carrera

ePO3.03.04
Experience in the Treatment With Pyridostigmine Monotherapy, in Patients With Spinal Muscular Atrophy in Colombia
Sandra Mesa

ePO3.03.05
Onasemnogene Abeparvovec Treatment Outcomes by Patient Weight at Infusion: Initial Findings from the RESTORE Registry
Laurent Servais

ePO3.03.06
Treatments and Outcomes for Patients with Spinal Muscular Atrophy Type 2: Findings from RESTORE Registry
Laurent Servais

ePO3.03.07
Characteristics and Epidemiology of Amyotrophic Lateral Sclerosis in a Health-Care Area in Northwestern Spain
Alejandra Espinosa Trujillo

ePosters Schedule

Friday 8 July, 2022

eP03.03.08
Characterization of the ALS patients' population in a large Italian Centre
Francesca Bianchi

eP03.03.09
Descriptive Analysis of Bulbar Amyotrophic Lateral Sclerosis (ALS) In the Northern Area of Tenerife
Helena Pérez Pérez

eP03.03.10
Atypical Onset of Amyotrophic Lateral Sclerosis: Seven Cases Report
Leila Tamaoui

eP03.03.11
Cognitive Decline in ALS Patients. MoCA Score and CSF Biomarkers (T-Tau, P-Tau, ß 42)
Sofía Portela Sánchez

Friday, Screen 4

Chairs: Isabel Leite & Pushpa Narayanswami

eP03.04.01
Effectiveness of Eculizumab Treatment for Generalized Myasthenia Gravis in Us Clinical Practice: gMG Registry Data
Srikanth Muppidi

eP03.04.02
Pharmacokinetics and Pharmacodynamics of Nipocalimab in Healthy Participants and Patients with Generalized Myasthenia Gravis
Anne-Gaëlle Dosne

eP03.04.03
Zilucoplan in Myasthenia Gravis: Safety and Tolerability Results From the Phase 3 Randomised RAISE Study
Maria Isabel Leite

eP03.04.04
Baseline Characteristics of Patients with Myasthenia Gravis Enrolled in an Expanded Access Programme for Efgartigimod
Jan L. De Bleecker

eP03.04.05
Promise-MG: Results of a Multicenter Comparative Effectiveness Study of Myasthenia Gravis Treatments
Pushpa Narayanaswami

eP03.04.06
The Association Between QMG Scores and Health-related Quality of Life in Myasthenia Gravis Patients
Cynthia Qi

eP03.04.07
Oral Tobacco, but Not Smoking, Is Associated With an Increased Risk of Myasthenia Gravis
Malin Petersson

eP03.04.08
Productivity Losses for Generalized Myasthenia Gravis Patients and their Caregivers: Association with Disease Severity
Saiju Jacob

eP03.04.09
Efficacy and Safety of Tolebrutinib in Adults with Generalized Myasthenia Gravis: Phase 3 Study Design
Sana Syed

eP03.04.10
Serological Diagnostics of MuSK Myasthenia Gravis in South Korea: Comparison of ELISA, RIPA and CBA
Young Nam Kwon

eP03.04.11
Clinical Differences Between Ocular and Generalized Myasthenia Gravis
Kasper Holst Axelsen

ePosters Schedule

Friday, Screen 5

Chair: Corrado Angelini

eP03.05.01
Effects of ERT on Cardiac Function in Classic Infantile Pompe Disease- 19 Years of Follow-up
Linda Scheffers

eP03.05.02
Safety of Home-Based Infusion of Alglucosidase Alfa in Adults With Late-Onset Pompe Disease
Imke Ditters

eP03.05.03
Minimal Clinical Important Difference for Lung Function and Walking Ability in Adult Pompe Patients
Aglina Lika

eP03.05.04
Effect of Alglucosidase Alfa Dosage on Survival and Walking Ability in Classic Infantile Pompe Disease
Imke Ditters

eP03.05.05
Czech Nationwide Screening of Pompe Disease – Case Reports
Livie Mensova

eP03.05.06
Involvement of Muscle Capillaries in Late Onset Pompe Disease (LOPD) With Childhood Onset
Corrado Angelini

eP03.05.07
Neuromuscular Symptoms and Gene Variants From a Long-Chain Fatty Acid Oxidation Disorder Gene Panel Program
Vanessa Rangel Miller

eP03.05.08
Natural History of Muscular Forms of Fatty Acid Beta-Oxidation Disorder: Description of 44 Patients
Alice Rouyer

eP03.05.09
A Case of VCP Mutation Featuring With Lobulated Myofiber, Motor Neuron Disease and Frontotemporal Dementia
Ju-hee Chae

eP03.05.10
Head Drop and Hyperckemia Associated With a Carnitine Palmitoyltransferase II (CPT2) Deficiency
Michela Bisciglia

eP03.05.11
Mitochondrial Network Disruption in Skeletal Muscles of the McArdle Mouse Model
Mónica Villarreal-Salazar

Friday, Screen 6

Chair: Josef Finsterer

eP03.06.01
Etiology, Genetics and Prevalence of Myopathies in the Population of Alicante (Spain)
Pablo Ros-Arlanzón

eP03.06.02
Far More than Vacuoles? Proteomic Profiling of Danon Disease Reveals a Striking Mitochondrial Phenotype
Felix Kleefeld

eP03.06.03
Global FKRP Registry - The Research Database for Limb Girdle Muscular Dystrophyase R9 (2I)
Volker Straub

eP03.06.04
First Description of a Caveolin-3-Related Rippling Disease in a Peruvian Family
Edna Bobadilla

eP03.06.05
Paucisymptomatic Hyperckemia as a Phenotype of Myopathy Associated with ano5: Case Report
Cristian Correa

eP03.06.06
Artificial Intelligence Electrocardiogram-Derived Age Detects Accelerated Aging in LMNA Patients
Shahar Shelly

eP03.06.07
Design of 'Time Is Muscle': RCT on IVIg Add-on to Prednisone in Newly Diagnosed Myositis
Renske Kamperman

eP03.06.08
The PACE-DM1 tool: An Adapted Home-Based Physical Activity Program for DM1 to Counter Physical Deficiencies
Anne-marie Fortin

eP03.06.09
Photosensitive Epilepsy and Polycystic Ovary Syndrome as Manifestations Of MERRF
Josef Finsterer

ePosters Schedule

Saturday 9 July, 2022

Saturday, Screen 1

Chair: TBC

eP04.01.01
The Burden of Duchenne Muscular Dystrophy in Belgium: A Registry-Based Study
Marjan Cosyns

eP04.01.02
Improving and Harmonising Care Standards for Duchenne Muscular Dystrophy in the UK
Cathy Turner

eP04.01.03
Illness Perceptions And Quality Of Life In Adolescents With Neuromuscular Disorders And Caregivers
Sam Geuens

eP04.01.04
A Pilot T1-Weighted MRI Study to Evaluate Chronic Corticosteroid-Use in Duchenne Muscular Dystrophy on Brain
Sam Geuens

eP04.01.05
Immune Function Indices in Patients with Amyotrophic Lateral Sclerosis
Oksana Kononets

eP04.01.06
Duchenne Muscular Dystrophy Diagnostic Gaps in Primary Medical Chain
Tatiana Gremiakova

Saturday, Screen 2

Chair: Kristl Claeys

eP04.02.01
Nothing to Laugh About
Frédéric Supiot

eP04.02.02
Acute Arsenic Intoxication Presentation Guillain-Barre Syndrome Mimic in Oncohematological Patient
Antonio J. Gutiérrez Martínez

eP04.02.03
Six Months of Electroneumyography in Ouagadougou, a Great Experience
Olivier Kapto

eP04.02.04
A Case Report on Orbital Myositis
Eni Reka

eP04.02.05
Belgian Retrospective Survey of Hereditary Transthyretin-Mediated (hATTR) Amyloidosis Patients Treated With Patisiran in Real-World Practice
Jan de Bleecker

eP04.02.06
Predictive Factors of Response to Tafamidis in a Cohort of Non-endemic ATTRv Patients
Laura Martínez-Vicente

eP04.02.07
Quantitative Sensory Testing in patients diagnosed with ATTR Amyloidosis in Colombia: A Case Series
Edicson Ruiz

ePosters Schedule

Saturday, Screen 3

Chair: TBC

eP04.03.01
Respiratory-Onset of ALS in a Pregnant Woman With a Novel SOD1 Mutation
Pegah Masrori

eP04.03.02
Association of APOE ε4 Allele With Survival in Amyotrophic Lateral Sclerosis Among Tunisian Cases
Imen Kacem

PS02.1
Comparison of Inflammation and Neurodegeneration Markers in CSF as Predictors of Survival in ALS Patients
Pegah Masrori

eP04.03.04
Systemic Genetic Screening of Korean Patients With Amyotrophic Lateral Sclerosis
Jin-ah Kim

eP04.03.05
New Mutation (Val31Gly) In PFN1 Gene Responsible for the Development of ALS18 in Bulgarian Pedigree
Teodor Angelov

eP04.03.06
Clinical Characteristics of c.63C>G (p.Phe21Leu) Variant in SOD1 Gene in Colombian Patients with ALS
Christian Correa

eP04.03.07
Serum Creatine Kinase and Creatinine in the Diagnosis and Prognostic Prediction of Amyotrophic Lateral Sclerosis
Imen Kacem

eP04.03.08
Potential Role of Mitochondrial Dysfunction in an Unusual Co-occurrence of ALS and Primary Biliary Cirrhosis
Pritikanta Paul

eP04.03.09
Hereditary Spastic Paraparesis Type 9A Mimicking ALS: A Case Report
Cristian Correa

eP04.03.10
International Phase 3 Trial Evaluating Sodium Phenylbutyrate/Taurursodiol in Amyotrophic Lateral Sclerosis (PHOENIX): Enrollment Update
Leonard H. van den Berg

Saturday, Screen 4

Chair: TBC

eP04.04.01
Long Term Preliminary Safety and Efficacy Outcomes for X-Linked Myotubular Myopathy with Gene Replacement Therapy
Nancy L. Kuntz

eP04.04.02
FORCETM Platform Demonstrates Durable Dystrophin Expression in Mdx Mice and Favorable Safety Profile in NHPs
Aaron Novack

eP04.04.03
Givinostat for the Management of Becker Muscular Dystrophy: A Randomised, Placebo-Controlled, Double-Blind Study
Daniele Velardo

eP04.04.04
Post-Authorisation Safety Study of Mexiletine Treatment in Patients with Non-Dystrophic Myotonia: Methodology Overview
Alla Zozulya-Weidenfeller

eP04.04.05
An Open-Label, Non-Comparative Study of Mexiletine in Children and Adolescents with Myotonic Disorders: Methodology Overview
Alla Zozulya-Weidenfeller

ePosters Schedule

eP04.04.06
Readily Available Low-Cost Highly Effective Treatment for Inherited Muscle Disorders Diagnosed by Whole Exome Sequencing
Ioannis Tsiverdis

eP04.04.08
Diagnosing Necrotizing Myopathy With Hmgcr Antibodies – A Quest Proving Time is Muscle
Zoltan Zsigmond Major

eP04.04.09
Anti-SRP-Positive Immune-Mediated Necrotizing Myopathy Accompanied by Hashimoto Thyroiditis
Seol-Hee Baek

eP04.04.10
Necrotizing Autoimmune Myopathy Anti-HMGCR Antibodies and Demyelinating Polyneuropathy
Dionis Vallejo

eP04.04.11
Hereditary Inclusion Body Myopathy (HIBM) as a Rare Clinical Entity: A Case Report
Ivan Barbov

Saturday, Screen 5

Chair: Jeniffer Shoskes

eP04.05.02
Clinical and Genetic Features of the Myotubular and Centronuclear Myopathy Patient Registry Cohort
Joanne Bullivant

eP04.05.03
Novel Splicing Mutation in MTM1 Leading To Two Abnormal Transcripts Causes Severe Myotubular Myopathy
Luca Bosco

eP04.05.04
Severe Congenital Myopathy With Type II Fibers Atrophy Due to MYL1
Jesica Maria Exposito Escudero

eP04.05.05
Clinical and Pathologic Findings of Korean Patients With Selenon-Related Myopathy
Seung-ah Lee

eP04.05.07
RYR1-Related Congenital Myopathy in a Cohort of Peruvian Patients
Edna Bobadilla

eP04.05.08
Risk of Malignant Hyperthermia in Patients Carrying a Variant in the Ryanodine Receptor 1 Gene
Sarah Herdewyn

eP04.05.09
Congenital Myopathy Caused by Mutations in the Neubulin Gene Associated to Schizophrenia: A Case Report
Sofía Portela Sánchez

eP04.05.10
Clinical Features of the UK FSHD Patient Registry
Helen Walker

eP04.05.11
Safety and Tolerability of Losmapimod for the Treatment of FSHD
Jennifer Shoskes

Saturday, Screen 6

Chair: TBC

eP04.06.01
Automated Integrative Splicing Predictor Tool: Focus on Deep Intronic Variants Prioritization for the DMD Gene
Mariateresa Zanobio

eP04.06.02
The Epidemiology of Mutations of Dystrophin in the Hungarian Population
Szabolcs Udvari

eP04.06.03
Targeting NAPDH oxidases in Duchenne Muscular Dystrophy: Diapocynin Therapeutic Effect on Adult MDX Mice
Ghali Guedira

eP04.06.04
Eteplirsen Safety, Tolerability, and Pharmacokinetics in Young Patients with DMD Amenable to Exon 51 Skipping
Eugenio Mercuri

eP04.06.05
Description of Osmolyte Pathways in Maturing MDX Mice Reveals Altered Taurine and Sodium/Myo-Inositol Co-transporter Levels
Caroline Merckx

eP04.06.06
Non-dystrophic Myotonia; The Patient Journey to Diagnosis
Tim Aldwinckle

eP04.06.07
Sensory Polyneuropathy in Oculopharyngeal Muscular Dystrophy, It This a Novel Phenotypical Findings?
Gauthier Remiche

eP04.06.08
Effects of FGF21 Supplementation in Muscle Cells from Mitochondrial Disease Patients
Margarita Chudenkova

eP04.06.09
Clinical Characterization of Familial Hyperkalemic Periodic Paralysis with a SCN4A Met1592Val Mutation
Sa-yoon Kang

eP04.06.11
Evaluation of the Relationship between Genotype and Phenotype of Dystrophinopathy in Iranian Race
Behnaz Ansari