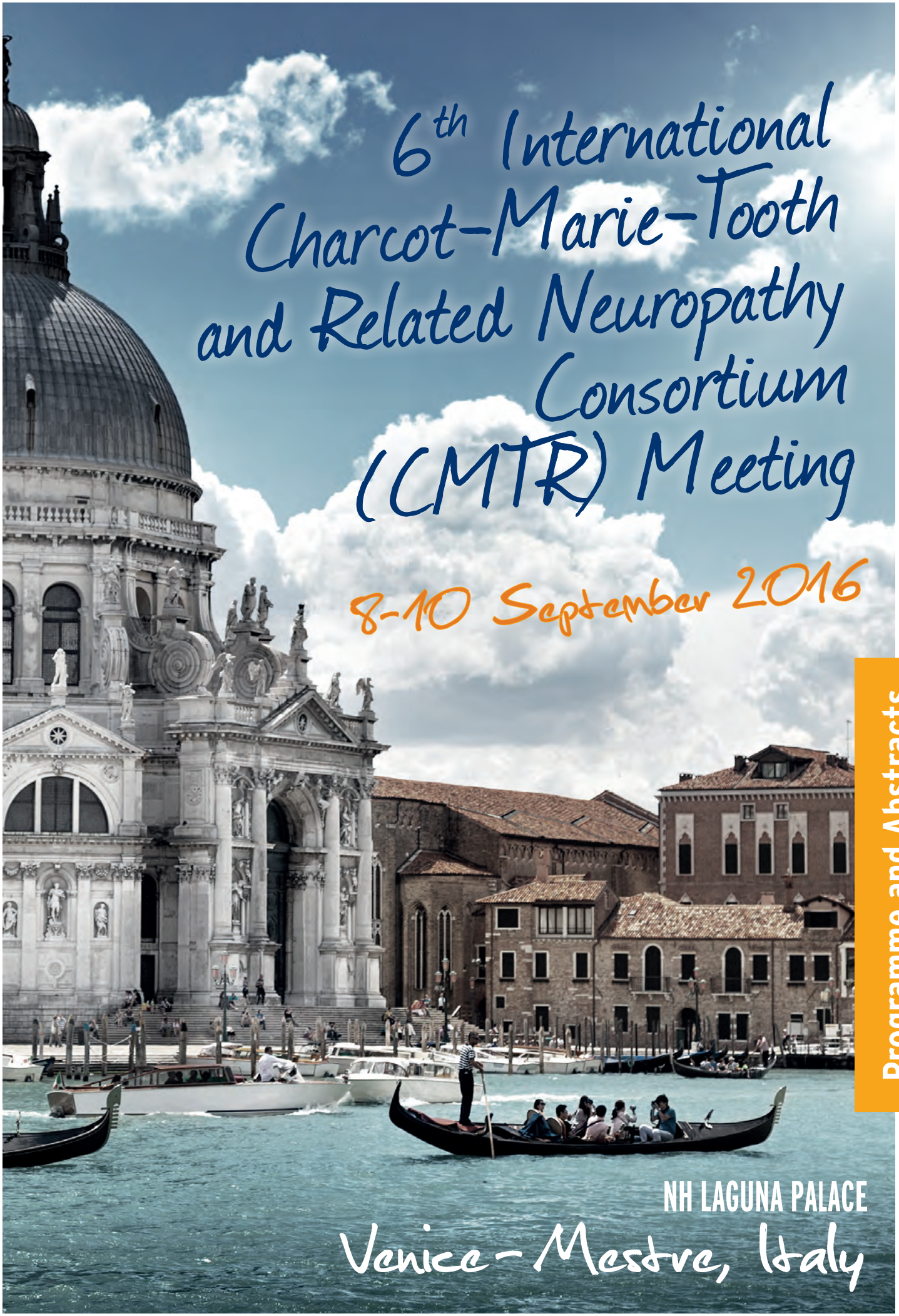


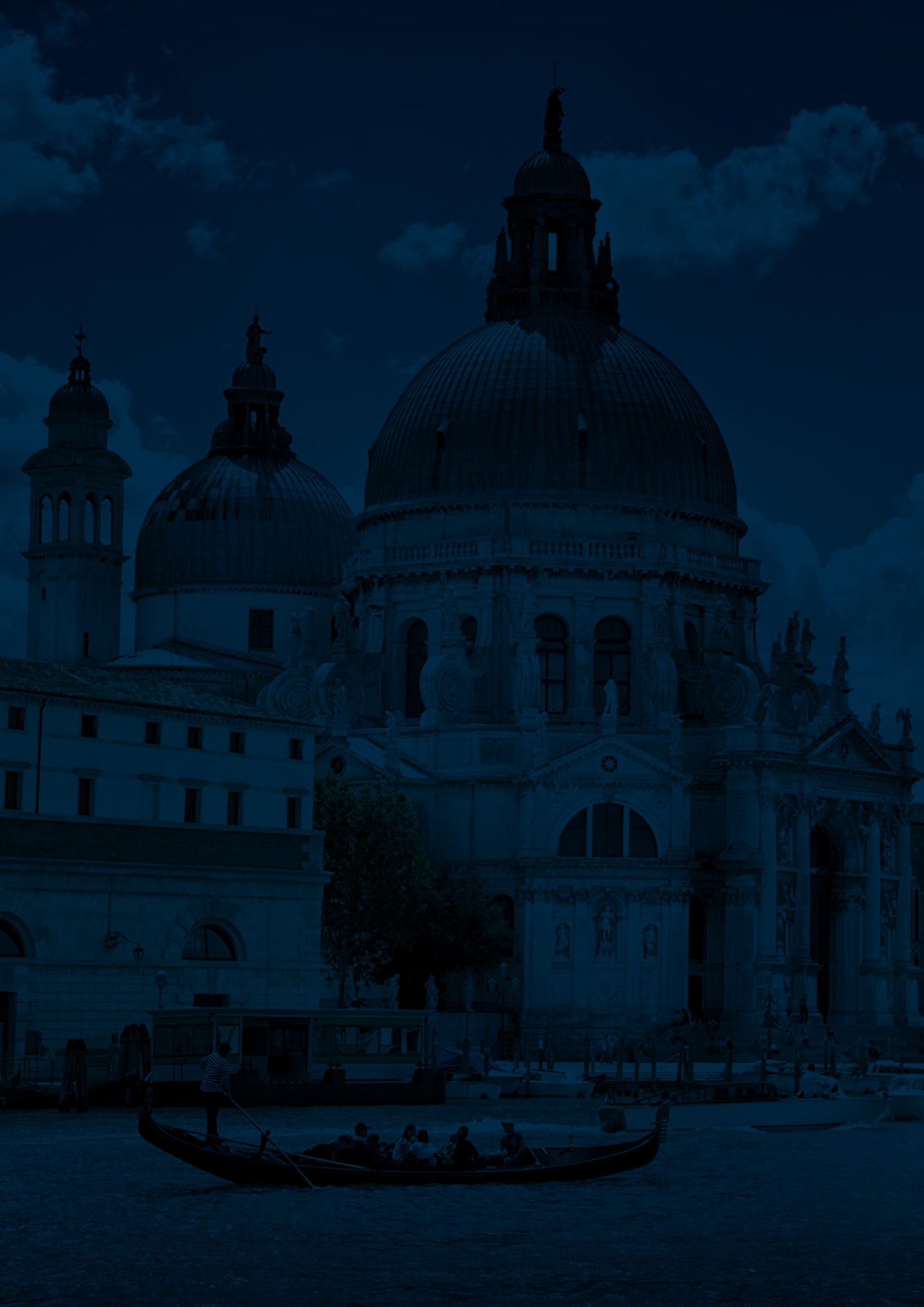
6th International Charcot-Marie-Tooth and Related Neuropathy Consortium (CMTR) Meeting

8-10 September 2016

Programme and Abstracts

NH LAGUNA PALACE

Venice-Mestre, Italy



Programme and Abstracts

6th International Charcot-Marie-Tooth and Related Neuropathy Consortium

(CMTR) Meeting

September 8 – 10, 2016

Venice Mestre, Italy

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Angelo Schenone (Genoa, Italy)
Gian Maria Fabrizi (Verona, Italy)

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Programme at a glance

THURSDAY, SEPTEMBER 8, 2016

10.00	REGISTRATION OPENING
10.30-12.30	CMT-ID MEETING (for CMT-ID sites)
14.00	OPENING OF THE MEETING
14.15-15.00	LECTURE - Modifying axonal transport as a therapeutic strategy in neuromuscular diseases *Giampietro Schiavo*
15.00-16.15	PLATFORM 1 PATHOMECHANISMS
16.15-17.15	PLATFORM 2 PATHOMECHANISMS = ARS
17.15-17.45	Coffee break
17.45-18.45	ORAL POSTER 1
18.45-19.45	POSTER SESSION 1
19.45	WELCOME COCKTAIL

FRIDAY, SEPTEMBER 9, 2016

8.00-9.00	PLATFORM 3 PATHOMECHANISMS AND MODELS
9.00-10.00	PLATFORM 4 PATHOMECHANISMS AND MODELS
10.00-10.30	Coffee break
10.30-11.30	PLATFORM 5 PHENOTYPES AND NEW GENES
11.30-12.30	ORAL POSTER 2
12.30-13.30	Lunch
13.30-14.30	POSTER SESSION 2
14.30-15.15	PLATFORM 6 EPIDEMIOLOGY AND GENETIC STUDIES
15.30	TRIP TO VENICE
20.00	GALA DINNER IN SAN SERVOLO

SATURDAY, SEPTEMBER 10, 2016

8.30-9.15	LECTURE - Unfolding transthyretin amyloidosis *Giampaolo Merlini*
9.15-10.15	PLATFORM 7 AMYLOID NEUROPATHY
10.15-11.00	PLATFORM 8 OTHER NEUROPATHIES
11.00-11.30	Coffee break
11.30-12.30	ORAL POSTER 3
12.30-13.30	POSTER SESSION 3
13.30-14.30	Lunch
14.30-15.15	PLATFORM 9 OUTCOME MEASURES
15.15-16.45	PLATFORM 10 THERAPIES
16.45-17.45	Coffee break
17.15-18.35	ORAL POSTER 4
18.35-19.35	POSTER SESSION 4
19.35	PRIZES AND CLOSING

PROGRAMME

THURSDAY, SEPTEMBER 8, 2016

10.00 **REGISTRATION OPENING**

10.30-12.30 **CMT-ID MEETING (for CMT-ID sites)**

14.00 **OPENING OF THE MEETING**
Mike E. Shy (Iowa City, United States), Davide Pareyson (Milan, Italy),
Gian Maria Fabrizi (Verona, Italy), Mary M. Reilly (London, United Kingdom)

14.15 - 15.00 **LECTURE**
Presented by Mary M. Reilly (London, United Kingdom)
MODIFYING AXONAL TRANSPORT AS A THERAPEUTIC STRATEGY IN NEUROMUSCULAR DISEASES
Giampietro Schiavo, London UK

15.00 - 16.15 **PLATFORM 1 - PATHOMECHANISMS**
Chairs: *Lawrence Wrabetz (Buffalo, United States) - Francesc Palau (Barcelona, Spain)*

01_1 REGULATION OF PERIPHERAL MYELIN PROTEIN 22 TRANSCRIPTION
John Svaren, Madison, United States

01_2 CRYPTIC AMYLOIDOGENIC ELEMENTS IN THE 3' UTR OF THE NEUROFILAMENT HEAVY GENE TRIGGER CHARCOT-MARIE-TOOTH DISEASE
Adriana Rebelo, Miami, United States

01_3 EIF2ALPHA PHOSPHORYLATION: A NOVEL HOMEOSTATIC HUB IN PERIPHERAL NEUROPATHIES
Maurizio D'Antonio, Milan, Italy

01_4 COORDINATION OF GROWTH FACTOR RECEPTOR TRAFFICKING AND CELL PROLIFERATION BY SH3TC2, A PROTEIN INVOLVED IN CHARCOT-MARIE-TOOTH NEUROPATHY
Vietxuan Phan, Dortmund, Germany

01_5 INVESTIGATING THE CELLULAR PATHOGENESIS OF CHARCOT-MARIE-TOOTH DISEASE TYPE 1C USING SKIN-DERIVED PATIENT FIBROBLASTS
Rhys Roberts, Cambridge, United Kingdom

16.15 - 17.15 **PLATFORM 2 - PATHOMECHANISMS: ARS**
Chairs: Anthony Antonellis (Ann Arbor, United States) - Charlotte Sumner (Baltimore, United States)

02_1 EXPANDING THE ALLELIC AND LOCUS HETEROGENEITY OF TRNA SYNTHETASE-RELATED CMT DISEASE
Anthony Antonellis, Ann Arbor, United States

02_2 THE NUCLEAR CONNECTION OF TYROSYL-TRNA SYNTHETASE TO NEURODEGENERATION
Sven Bervoets, Antwerp, Belgium

02_3 MUTATION-INDUCED STRUCTURAL OPENING AND ABERRANT INTERACTION LINK TRNA SYNTHETASES TO CHARCOT-MARIE-TOOTH DISEASE
David Blocquel, La Jolla, United States

02_4 ABERRANT NEUROPILIN 1 INTERACTION AS A BIOMARKER IN DIAGNOSING TRNA SYNTHETASE-LINKED CHARCOT-MARIE-TOOTH DISEASE
Grace Kooi, La Jolla, United States

17.15 - 17.45 Coffee break

17.45 - 18.45 ORAL POSTER 1
Chairs: Jonathan Baets (Edegem, Belgium) - Kelly Monk (Saint Louis, United States)

OP1_1 GLIAL NEUREGULIN-1 REGULATES SCHWANN CELL PATHOLOGY IN CHARCOT-MARIE-TOOTH DISEASE -1A
Ruth M. Stassart, Göttingen, Germany

OP1_2 BIOMARKERS IN CHARCOT-MARIE-TOOTH DISEASE 1A
Michael W. Sereda, Göttingen, Germany

OP1_3 LACK OF GDAP1 IN MOTOR NEURONS REVEALS IMPAIRMENT IN MITOCHONDRIAL DYNAMICS AND CALCIUM HOMEOSTASIS IN THE CHARCOT-MARIE-TOOTH DISEASE PATHOGENESIS
Azahara Civera-Tregón, Barcelona, Spain

OP1_4 DYNC1H1 DIRECTS BOTH AXONAL AND SCHWANN CELL RESPONSE TO NERVE INJURY IN VIVO
Melissa Ducommun, Philadelphia, United States

OP1_5 LOSS OF FUNCTION OF THE CMT-RELATED GENE GDAP1 REDUCES STORE-OPERATED Ca2+ ENTRY (SOCE) AND SOCE-STIMULATION OF RESPIRATION IN INTACT NEURAL CELLS
Francesc Palau, Barcelona, Spain

OP1_6 CHARACTERIZING THE ALLELIC HETEROGENEITY OF GARS-MEDIATED PERIPHERAL NEUROPATHY
Stephanie Oprescu, Ann Arbor, United States

OP1_7 DOMINANT GARS MUTATIONS CAUSE A DEVELOPMENTAL PERTURBATION OF SENSORY NEURON FATE IN CHARCOT-MARIE-TOOTH TYPE 2D MICE
James Sleigh, London, United Kingdom

OP1_8 TWO NOVEL PATHOGENIC MUTATIONS IN THE AARS GENE CAUSE CHARCOT-MARIE-TOOTH DISEASE TYPE 2
Marian Weterman, Amsterdam, The Netherlands

OP1_9 THE ROLE OF HISTONE DEACETYLASE 6 (HDAC6) IN MUTANT GLYCYL-tRNA SYNTHETASE (GARS) AND MUTANT SMALL HEAT SHOCK PROTEIN B1 (HSPB1)-INDUCED AXONAL CHARCOT-MARIE-TOOTH DISEASE (CMT)
Veronick Benoy, Leuven, Belgium

OP1_10 VARIABILITY OF SYMPTOMS ASSOCIATED WITH AMINOACYL-tRNA SYNTHETASE GENES FOR PATIENTS SEEN IN A LARGE CMT CLINIC
Shawna Feely, Iowa City, United States

OP1_11 THREE-DIMENSIONAL STUDY OF NEUROMUSCULAR JUNCTIONS (NMJ) IN HETEROZYGOUS R98C KNOCK-IN CMT1B MOUSE MODEL BY OVEREXPRESSION NEUREGULIN I TYPE III
Yunhong Bai, Iowa City, United States

OP1_12 ENDOPLASMIC-RETICULUM-ASSOCIATED DEGRADATION (ERAD) MODULATES DISEASE SEVERITY IN A CHARCOT-MARIE-TOOTH-1B MOUSE MODEL
Vera Giulia Volpi, Milan, Italy

18.45 - 19.45 POSTER SESSION 1

P1_1 VIRTUAL GRAND ROUNDS IN THE INHERITED NEUROPATHY CONSORTIUM
Lisa Abreu, Miami, United States

P1_2 APPLYING CRISPR/CAS9 TO IN VITRO CELL LINES FOR ACCURATE CMT DISEASE MODELING
Elias Adriaenssens, Antwerp, Belgium

P1_3 EXPLORING THE REGULATION OF TRANSIENT RECEPTOR POTENTIAL VANILLOID 4 (TRPV4) BY THE E3 UBIQUITIN LIGASE NEDD4
William Aisenberg, Baltimore, United States

P1_4 CHARCOT-MARIE-TOOTH DISEASE IN TURKEY: CLINICAL AND GENETIC FINDINGS FROM A SINGLE- CENTRE EXPERIENCE
Halil Ibrahim Akçay, Istanbul, Turkey

P1_5 VOLTAGE-GATED NA+ CHANNEL BLOCKERS ATTENUATE THE TOXICITY OF PROLONGED REPETITIVE ACTIVITY IN A MOUSE MODEL OF CMT1B
Susana Alvarez, Copenhagen, Denmark

P1_6 BANDS OF FONTANA IN MURINE PERIPHERAL NERVES INDICATE AXON LENGTH
Luke Alvey, Dublin, Ireland

P1_7 OPTIMIZATION OF A HIGH-THROUGHPUT SCREENING SYSTEM IN YEAST
Silvia Amor Barris, Antwerp, Belgium

P1_8 DROSOPHILA AS A FUNCTIONAL PLATFORM FOR VALIDATION OF NOVEL GENES CAUSING AUTOSOMAL RESESSIVE CHARCOT-MARIE-TOOTH DISEASE
Derek Atkinson, Antwerp, Belgium

P1_9 NDRG1: EVIDENCE FOR A SECOND FOUNDER MUTATION IN BULGARIA
Derek Atkinson, Antwerp, Belgium

P1_10 THE R373C FBLN5 MUTATION IS ASSOCIATED WITH A PARTICULAR CHARCOT-MARIE-TOOTH TYPE 1 PHENOTYPE
Michaela Auer-Grumbach, Vienna, Austria

P1_11 A COMPREHENSIVE UPDATE OF THE INHERITED NEUROPATHIES CONSORTIUM OF THE RARE DISEASES CLINICAL RESEARCH NETWORK
Chelsea Bacon, Iowa City, United States

P1_12 CORRELATION OF HAND FUNCTION LOSS AND CMTNSv2 SCORES IN CMT1A PATIENTS
Chelsea Bacon, Iowa City, United States

P1_13 GENETIC DISTRIBUTION IN THE SPANISH TREAT-CMT CONSORTIUM
Marisa Barreiro, Valencia, Spain

P1_14 SPG11 IS AN OVERLAPPING GENE BETWEEN CHARCOT-MARIE-TOOTH DISEASE AND HEREDITARY SPASTIC PARAPLEGIA
Esra Battaloglu, Istanbul, Turkey

P1_15 CHARACTERIZATION OF MOTOR AND SENSORY NEURONAL DYSFUNCTION IN BOTH IN VITRO AND IN VIVO MODELS OF CMT2A PATHOLOGY
Nathalie Bernard-Marissal, Lausanne, Switzerland

P1_16 DEVELOPMENT AND OPTIMIZATION OF A PROTOCOL FOR RNA EXTRACTION FROM HUMAN SKIN BIOPSY OF PATIENTS AFFECTED BY PAINFUL AND PAINLESS PERIPHERAL NEUROPATHY
Silvia Santoro, Milan, Italy

P1_17 MUTATIONS IN GLYCYL-TRNA-SYNTHETASE IMPAIR MITOCHONDRIAL FUNCTION IN NEURONS
Veronika Boczonadi, Newcastle upon Tyne, United Kingdom

P1_18 USING WORMS TO SCREEN FOR NOVEL GENE MUTATIONS CAUSING INHERITED PERIPHERAL NEUROPATHY: A VALIDATION STUDY
Megan Brewer, Concord, Australia

P1_19 MITOFUSIN 2 GENE MUTATIONS IN A TURKISH CHARCOT-MARIE-TOOTH DISEASE COHORT
Ayse Candayan, Istanbul, Turkey

P1_20 NULL MUTATIONS IN THE DESERT HEDGEHOG GENE MAY CAUSE A MINIFASCICULE NEUROPATHY OUTSIDE THE 46,
XY GONADAL DYSGENESIS SYNDROME
Gian Maria Fabrizi, Verona, Italy

P1_21 PHENOTYPIC HETEROGENEITIES AND CENTRAL NERVOUS SYSTEM INVOLVEMENTS IN CHARCOT- MARIE-TOOTH DISEASE WITH NEFL MUTATIONS
Geon Kwak, Seoul, South Korea

P1_22 ALTERATIONS OF AUTOPHAGIC FLUX IN CHARCOT-MARIE-TOOTH 2B DISEASE
Mariangela Stasi, Lecce, Italy

P1_23 DYNAMIC BALANCE: RELATING FUNCTIONAL REACH TESTS TO FALLS AND IMPAIRMENT
Gita Ramdharry, London, United Kingdom

P1_24 HEREDITARY NEUROPATHY WITH LIABILITY TO PRESSURE PALSIES SURVEY - UTILIZING SOCIAL MEDIA TO UNCOVER OVERLOOKED SYMPTOMS
Ayse Deniz Elmali, Istanbul, Turkey

P1_25 IMPACT OF FOOT ALIGNMENT IN PEDIATRIC CHARCOT-MARIE-TOOTH-DISEASE
Timothy Estilow, Philadelphia, United States

P1_26 BALANCE IMPAIRMENT IN PEDIATRIC CHARCOT-MARIE-TOOTH-DISEASE
Timothy Estilow, Philadelphia, United States

P1_27 IMPACT OF VISUAL INPUT ON BALANCE IN CHILDREN WITH CHARCOT-MARIE-TOOTH DISEASE
Timothy Estilow, Philadelphia, United States

P1_28 THE ROLE OF INFLAMMATION IN NEURODEGENERATION ASSOCIATED WITH LACK OF GDAP1 IN CHARCOT-MARIE-TOOTH DISEASE
Francesc Palau, Barcelona, Spain

P1_29 DIAGNOSTIC SCREENING OF EIGTHY CHARCOT-MARIE-TOOTH TYPE2 PATIENTS USING ION TORRENT PLATFORM BY CUSTOMIZED PANEL
Moreno Ferrarini, Verona, Italy

P1_30 GDPA1 MUTATIONS IN BRAZILIAN PATIENTS WITH CMT2, CMT2-AR AND CMT4
Wilson Marques Jr., Ribeirão Preto, Brazil

P1_31 A CHARCOT-MARIE-TOOTH TYPE 2 FAMILY CARRYING THE PRO7ARG MUTATION IN THE IMMEDIATE N-TERMINAL REGION OF THE HEAT-SHOCK 27-KDA PROTEIN
Francesca Gualandi, Ferrara, Italy

P1_32 DESCRIPTION OF A CLUSTER OF PATIENTS WITH THE HSPB1 p.R140G MUTATION
Rafael Sivera, Gandia, Spain

P1_33 SUPERIMPOSED INFLAMMATORY NEUROPATHY IN PATIENTS AFFECTED BY CHARCOT-MARIE-TOOTH NEUROPATHY
Chiara Gemelli, Genova, Italy

P1_34 TWO NOVEL "DOUBLE" POINT MUTATIONS IN MFN2 (MITOFUSIN2) GENE IN TWO UNRELATED AXONAL CMT PATIENTS
Alessandro Geroldi, Genova, Italy

P1_35 REGULATION OF THE NRG1/ERBB SYSTEM IN CMT1A PERIPHERAL NERVES
Giovanna Gambarotta, Torino, Italy

19.45 WELCOME COCKTAIL

FRIDAY, SEPTEMBER 9, 2016

8.00 - 9.00 PLATFORM 3 - PATHOMECHANISMS AND MODELS
Chairs: Mario Saporta (Miami, United States) - Alessandra Bolino (Milan, Italy)

03_1 CHARACTERIZATION OF THE CMT1B-P0T124M MOUSE MODEL
Ghjuvan'Ghjacumu Shackleford, Buffalo, United States

03_2 NERVE EXCITABILITY CHANGES AFTER NaV1.8 CHANNEL BLOCKER TREATMENT IN MICE DEFICIENT OF MYELIN PROTEIN P0
Mihai Moldovan, Copenhagen, Denmark

03_3 AXONAL RNA PROFILING OF HUMAN MOTOR NEURONS FROM PATIENTS WITH CHARCOT-MARIE-TOOTH DISEASE AS A NOVEL APPROACH TO STUDY AXON DEGENERATION
Renata de Moraes Maciel, Doral, United States

03_4 MECHANISMS OF TRPV4-MEDIATED HEREDITARY AXONAL NEUROPATHY IN DROSOPHILA
Thomas Lloyd, Baltimore, United States

9.00 - 10.00 PLATFORM 4 - PATHOMECHANISMS AND MODELS
Chairs: Vincent Timmerman (Antwerp, Belgium) - Alex Rossor (London, United Kingdom)

04_1 EXPRESSION OF HSPB8_K141N MUTANT LEADS TO AXONOPATHY AND MOTOR DEFICITS IN A NEW TRANSGENIC MOUSE MODEL OF CMT2L
Delphine Bouhy, Antwerp, Belgium

04_2 CHARACTERIZATION OF AN ATP7AT985I CONDITIONAL KNOCK-IN MOUSE MODEL FOR X-LINKED DISTAL HEREDITARY MOTOR NEUROPATHY
Gonzalo Perez Siles, Sydney, Australia

04_3 GENOME-WIDE ASSOCIATION STUDY IDENTIFIES POTENTIAL GENETIC MODIFIERS IN CHARCOT-MARIE- TOOTH DISEASE TYPE 1A
Stephan Züchner, Miami, United States

04_4 IMPAIRED NERVE CONDUCTION RELATING TO THE ALTERED RATIO OF NON-COMPACT REGION OVER COMPACT REGION OF MYELIN
Jun Li, Nashville, United States

10.00 - 10.30 Coffee break

10.30 - 11.30 PLATFORM 5 - PHENOTYPES AND NEW GENES
Chairs: Stephan Züchner (Miami, United States) - Byung-Ok Choi (Seoul, South Korea)

05_1 GENOTYPE-PHENOTYPE CHARACTERISTICS AND BASELINE NATURAL HISTORY OF CMT2A CAUSED BY MUTATIONS IN THE MFN2 GENE
Alexander Rossor, London, United Kingdom

05_2 MUTATIONS IN MORC2 GENE CAUSE AXONAL CHARCOT-MARIE-TOOTH DISEASE
Paula Sancho, Valencia, Spain

05_3 INVESTIGATING THE FUNCTIONAL CONSEQUENCES OF MICRORCHIDIA 2 (MORC2) MUTATIONS CAUSING AXONAL CMT (CMT2Z)
Marina Kennerson, Concord, Australia

05_4 CONTACTIN-ASSOCIATED PROTEIN 1 MUTATIONS CAUSE CHARACTERISTIC ULTRASTRUCTURAL LESIONS IN THE PARANODAL REGION OF HUMAN PERIPHERAL NERVES
Jean-Michel Vallat, Limoges, France

11.30 - 12.30 ORAL POSTER 2
Chairs: Marina Grandis (Genoa, Italy) - Pavel Seeman (Prague, Czech Republic)

OP2_1 NEW MISSENSE MUTATIONS IN VRK1 ARE ASSOCIATED WITH AUTOSOMAL RECESSIVE AXONAL CHARCOT-MARIE-TOOTH DISEASE
Lara El Bazzal, Marseille, France

OP2_2 THE VARIANT p.G66V IN CHCHD10 CAUSES TYPE 2 CHARCOT-MARIE-TOOTH DISEASE
Emil Ylikallio, Helsinki, Finland

OP2_3 GAIT ABNORMALITIES AND ASSOCIATED CHANGES IN SKELETAL MUSCLE BIOLOGY IN TREMBLER J NEUROPATHIC MICE
Lucia Notterpek, Gainesville, United States

OP2_4 A NOVEL CMT2P MISSENSE MUTATION IN THE RING DOMAIN OF LRSAM1 IMPAIRS TRANSCRIPTOME FORMATION
Sezgi Arpag, Nashville, United States

OP2_5 COMPOUND HETEROZYGOUS MUTATION IN SGPL1 CAUSE AN AUTOSOMAL RECESSIVE CHARCOT- MARIE-TOOTH DISEASE TYPE 2 IN A SERBIAN FAMILY
Derek Atkinson, Antwerp, Belgium

OP2_6 A PROPOSAL FOR UPDATING THE CLASSIFICATION OF CHARCOT-MARIE-TOOTH DISEASES AND RELATED DISORDERS
Laurent Magy, Limoges, France

OP2_7 MODELING OF TRIM2, TRIPARTITE MOTIF CONTAINING 2, AND OTHER CMT2, CHARCOT-MARIE-TOOTH NEUROPATHY TYPE 2, MUTATIONS IN PATIENT-SPECIFIC MOTOR NEURONS
Markus Sainio, Helsinki, Finland

OP2_8 MULTIFOCAL MOTOR NEUROPATHY CAUSED BY PHOSPHATASE AND TENSIN HOMOLOUGE (PTEN) MUTATION
Boglarka Bansagi, Newcastle upon Tyne, United Kingdom

OP2_9 TDP43-DEPENDENT ALTERATION OF RNA METABOLISM IN HSPB8-RELATED AUTOSOMAL DOMINANT DISTAL HEREDITARY MOTOR NEUROPATHY AND MYOFIBRILLAR MYOPATHY: A FAMILY STUDY
Andrea Cortese, Pavia, Italy

OP2_10 RARE CODING VARIANTS IN THE MME GENE, ENCODING THE METALLOPROTEASE NEPRILYSIN, ARE LINKED TO LATE-ONSET AXONAL NEUROPATHIES
Michaela Auer-Grumbach, Vienna, Austria

OP2_11 IPSC-DERIVED MOTOR NEURONS FROM CMT2A PATIENTS WITH MFN2 MUTATIONS HAVE MITOCHONDRIAL DEFECTS
Yueqin Zhou, Los Angeles, United States

OP2_12 A LIPID BASED APPROACH TO IMPACT ON CMT1A PHENOTYPE
Lucilla Nobbio, Genoa, Italy

12.30 - 13.30 Lunch

13.30 - 14.30 POSTER SESSION 2

P2_1 VENTRAL ABDOMINAL SENSORY LOSS IS COMMON IN LENGTH DEPENDENT SENSORIMOTOR PERIPHERAL NEUROPATHY OF INHERITED AND OTHER ETIOLOGIES
Benn Smith, Scottsdale, United States

P2_2 THE INVOLVEMENT OF AN RNA BINDING PROTEIN IN CHARCOT-MARIE-TOOTH DISEASE
Thomas Geuens, Antwerp, Belgium

P2_3 A PHASE 1 HEALTHY VOLUNTEER STUDY OF ACE-083, A NOVEL, LOCALLY-ACTING MUSCLE AGENT
Chad Glasser, Cambridge, United States

P2_4 HEARING LOSS IN CHARCOT-MARIE-TOOTH
Tiffany Grider, Iowa City, United States

P2_5 CLINICAL NEXT GENERATION SEQUENCING GENE PANEL IDENTIFIED A NOVEL ATP7A MUTATION IN TWO BROTHERS WITH DISTAL HEREDITARY MOTOR NEUROPATHY AND AUTONOMIC DYSFUNCTION
Francesca Gualandi, Ferrara, Italy

P2_6 SPINAL MUSCLE ATROPHY WITH LOWER EXTREMITY PREDOMINANCE (SMA-LED) ASSOCIATED TO A NOVEL DYNC1H1 MUTATION: THE RELEVANCE OF MUSCLE MRI
Raquel Guimarães-Costa, Paris, France

P2_7 IMPAIRMENT OF AUTOPHAGY AS A POSSIBLE PATHOMECHANISM FOR CMT CAUSING MUTATIONS IN HSPB1
Mansour Haidar, Wilrijk, Belgium

P2_8 A NOVEL MISSENSE MUTATION IN THE E3 UBIQUITIN LIGASE LRSAM1 CAUSES CHARCOT-MARIE-TOOTH DISEASE TYPE 2
Johanna Hakonen, Amsterdam, Netherlands

P2_9 CLINICAL AND GENETIC PROFILES IN KOREAN PATIENTS WITH X-LINKED DOMINANT CHARCOT-MARIE-TOOTH DISEASE TYPE 1
Hyun Myung Doo, Seoul, South Korea

P2_10 SIGMAR1 MUTATION ASSOCIATED WITH AUTOSOMAL RECESSIVE SILVER-LIKE SYNDROME
Alejandro Horga, London, United Kingdom

P2_11 RELATIONSHIP OF PLANTAR SENSATION, FOOT LOADING AND WALKING ABILITY IN PATIENTS WITH CHARCOT-MARIE-TOOTH DISEASE (CMT)
Daphne Hüttemann, Münster, Germany

P2_12 EFFECTS OF HIP ASSIST ROBOT ON CHARCOT-MARIE-TOOTH PATIENTS
Sun Hee Hwang, Seoul, South Korea

P2_13 PROTEIN NETWORK ANALYSIS TO IDENTIFY NOVEL DRUG TARGETS FOR ALS
Jon Klein, Rochester, MN, United States

P2_14 IDENTIFICATION OF COMMON PATHOMECHANISMS INVOLVED IN THE PATHOGENESIS OF AXONAL CMT SUBTYPES
Manisha Juneja, Antwerp, Belgium

P2_15 WHOLE EXOME SEQUENCING ANALYSIS IN EIGHT POLISH HSN FAMILIES
Dagmara Kabzińska, Warsaw, Poland

P2_16 GENETIC EPIDEMIOLOGY OF INHERITED PERIPHERAL NEUROPATIES IN BULGARIA
Ivaylo Tournev, Sofia, Bulgaria

P2_17 A CASE OF CMT4H RESPONSIVE TO STEROID TREATMENT
Elizabeth Kichula, Philadelphia, United States

P2_18 A SEVERE SPINAL MUSCULAR ATROPHY PHENOTYPE ASSOCIATED WITH A NOVEL BICD2 MUTATION
Elizabeth Kichula, Philadelphia, United States

P2_19 MUTATIONS IN ATL3 CAUSING HSAN DISRUPT ER-DYNAMICS AND CROSSTALK WITH MITOCHONDRIA
Vincent Timmerman, Antwerp, Belgium

P2_20 INTERNATIONAL, MULTI-CENTER, RANDOMIZED, DOUBLE-BLIND, PLACEBO-CONTROLLED PHASE 3 STUDY ASSESSING IN PARALLEL GROUPS THE EFFICACY AND SAFETY OF 2 DOSES OF PXT3003 IN PATIENTS WITH CHARCOT-MARIE-TOOTH DISEASE TYPE 1A TREATED FOR 15 MONTHS
René Goedkoop, Issy-les-Moulineaux, France

P2_21 BASELINE ANALYSIS OF PXT3003 PHASE 2 DATA REVEALS TWO BLOOD EARLY CANDIDATE BIOMARKERS FOR THERAPEUTIC RESPONSE IN CHARCOT-MARIE-TOOTH DISEASE TYPE 1A
Julien Laffaire, Issy-Les-Moulineaux, France

P2_22 NOVEL DE-NOVO MUTATION IN THE GNB4 GENE IN A CZECH PATIENT WITH CHARCOT-MARIE-TOOTH DISEASE CONFIRMS THE CAUSALITY OF THIS GENE
Petra Laššuthová, Prague, Czech Republic

P2_23 ORTHOPAEDIC COMPLICATIONS IN CHARCOT-MARIE-TOOTH DISEASE: RESULTS OF A PROSPECTIVE STUDY
Matilde Laurà, London, United Kingdom

P2_24 A MULTIDISCIPLINARY APPROACH TO MONITOR PREGNANCY IN CHARCOT-MARIE-TOOTH DISEASE
Matilde Laurà, London, United Kingdom

P2_25 ARE PREGNANCIES THE CAUSE OF CHARCOT-MARIE-TOOTH TYPE 1 GENDER DIFFERENCES?
Rita de Cassia Carvalho Leal, Ribeirão Preto, Brazil

P2_26 SMALL HEAT SHOCK PROTEIN B3 (HSPB3) MUTATION IN A LATE-ONSET CMT2 FAMILY
Su Jung Lee, Gongju, South Korea

P2_27 PMP22 MUTANT ALLELE-SPECIFIC SIRNA ALLEVIATES DEMYELINATING NEUROPATHIC PHENOTYPE IN VIVO
Ji-Su Lee, Seoul, South Korea

P2_28 CHARCOT-MARIE-TOOTH DISEASE: FREQUENCY OF GENETIC SUBTYPES IN SARDINIAN POPULATION
Lorena Lorefice, Cagliari, Italy

P2_29 NOVEL AIFM1 MUTATION CAUSE AN EARLY CHILDHOOD-ONSET POLYNEUROPATHY WITH EXCLUSIVE MOTOR INVOLVEMENT
Vincenzo Lupo, Valencia, Spain

P2_30 CHILDHOOD HEREDITARY NEUROPATHY IN THAILAND
Oranee Sanmaneechai, Bangkok, Thailand

P2_31 SENSORY NEUROPATHY IN CHILDREN PRESENTING WITH BEHR SYNDROME DUE TO OPA1 MUTATIONS
Yann Pereon, Nantes, France

P2_32 NERVE CONDUCTION VELOCITY IN CMT1A: WHAT ELSE CAN WE TELL?
Fiore Manganelli, Naples, Italy

P2_33 THE AARS-RELATED NEUROPATHY IN FOUR CZECH PATIENTS- CLINICAL AND ELECTROPHYSIOLOGICAL STUDY
Radim Mazanec, Prague, Czech Republic

P2_34 A PERSONALIZED GENE THERAPY APPROACH FOR CHARCOT-MARIE-TOOTH DISEASE TYPE 2D
Kathryn Morelli, Bar Harbor, United States

P2_35 DEFINING CELLULAR PHENOTYPES OF RECESSIVE AND DOMINANT GARS-MEDIATED DISEASE
Rebecca Meyer, Ann Arbor, United States

14.30 - 15.15 PLATFORM 6 - EPIDEMIOLOGY AND GENETIC STUDIES
Chairs: Franco Taroni (Milan, Italy) - Marina Kennerson (Concord, Australia)

06_1 EPIDEMIOLOGIC ASPECTS OF THE CHARCOT-MARIE-TOOTH DIAGNOSIS IN DENMARK; A NATIONWIDE STUDY
Signe Vaeth, Aarhus, Denmark

06_2 AN NGS TARGETED-RESEQUENCING APPROACH FOR THE GENETIC DIAGNOSIS OF INHERITED PERIPHERAL DEMYELINATING NEUROPATHIES
Stefania Magri, Milan, Italy

06_3 THE INHERITED NEUROPATHY VARIANT BROWSER
Stephan Züchner, Miami, United States

15.30 TRIP TO VENICE

20.00 GALA DINNER - SAN SERVOLO, VENICE

SATURDAY, SEPTEMBER 10, 2016

8.30 - 9.15 LECTURE

Presented by David Adams (Le Kremlin-Bicêtre, France)

UNFOLDING TRANSTHYRETIN AMYLOIDOSIS
Giampaolo Merlini, Pavia, Italy

9.15 - 10.15 PLATFORM 7 - AMYLOID NEUROPATHY

Chairs: Teresa Coelho (Lisbona Portugal) - Andrea Cortese (London, United Kingdom)

07_1 EFFECT OF AMYLOIDOSIS ON SMALL SENSORY NERVE FIBERS AND PERIPHERAL NERVE FUNCTION IN DISTAL LEG OF PATIENTS WITH TRANSTHYRETIN FAMILIAL AMYLOID POLYNEUROPATHY
Gigi Ebenezer, Baltimore, United States

07_2 TAFAMIDIS FOR THE TREATMENT OF FAMILIAL AMYLOID POLYNEUROPATHY: EFFICACY AND SAFETY DATA FROM A GROUP OF PORTUGUESE PATIENTS WITH THREE YEARS FOLLOW-UP
Teresa Coelho, Porto, Portugal

07_3 NOVEL ANTIBODIES AGAINST AMYLOIDOGENIC FORMS OF TRANSTHYRETIN BIND SPECIFICALLY TO DEPOSITS PRESENT IN TISSUES FROM ATTR AMYLOIDOSIS PATIENTS
Jeffrey Higaki, San Francisco, United States

07_4 PERIPHERAL POLYNEUROPATHY IN WILD TYPE TRANSTHYRETIN CARDIAC AMYLOIDOSIS: INITIAL OBSERVATONS AND SIMILARITY TO FAMILIAL AMYLOID POLYNEUROPATHY
Peter James B. Dyck, Rochester, United States

10.15 - 11.00 PLATFORM 8 - OTHER NEUROPATHIES

Chairs: Giuseppe Lauria (Milan, Italy) - Peter James B. Dyck (Rochester, United States)

08_1 MOLECULAR INVERSION PROBE-TARGETED GENERATION SEQUENCING TO IDENTIFY GENETIC MARKERS IN PAINFUL NEUROPATHIES - THE PROPANE STUDY
Monique Gerrits, Maastricht, The Netherlands

08_2 LOWER LIMB MRI-DETERMINED FAT FRACTION IS HIGHLY RESPONSIVE OVER 12 MONTHS IN PATIENTS WITH HEREDITARY SENSORY NEUROPATHY TYPE 1
Matthew Evans, London, United Kingdom

08_3 AN INTRA-CHROMOSOMAL TRANSLOCATION INSERTS A 1.35 MEGABASE DNA FRAGMENT INTO THE CHROMOSOME 7q34-q36.2 DHMN1 LOCUS
Alexander Drew, Sydney, Australia

11.00 - 11.30 Coffee break

11.30 - 12.30 ORAL POSTER 3

Chairs: Michaela Auer-Grumbach (Vienna, Austria) - Wilson Marques Jr. (Ribeirão Preto, Brazil)

OP3_1 NOVEL PHE210LEU MISSENSE MUTATION IN AIFM1 GENE IS ASSOCIATED WITH AN AXONAL POLYNEUROPATHY
Megan Simmons, Nashville, United States

OP3_2 HNRNPA1 MUTATIONS EXPAND THE SPECTRUM OF MOTOR NEURON DISEASES
Inès Mademan, Antwerp, Belgium

OP3_3 SENSITIVITY OF MRI AS A BIOMARKER OF DISEASE SEVERITY IN CHILDREN WITH CHARCOT-MARIE- TOOTH DISEASE
Kayla Cornett, Sydney, Australia

OP3_4 ASSESSMENT OF NERVE MRI AS A BIOMARKER OF CHARCOT-MARIE-TOOTH DISEASES
Richard Dortch, Nashville, United States

OP3_5 DIAGNOSTIC PITFALLS OF TRANSTHYRETIN AMYLOIDOSIS: AVOIDING MISDIAGNOSIS OF A TREATABLE HEREDITARY NEUROPATHY
Andrea Cortese, Pavia, Italy

OP3_6 PATIENT CENTRICITY: SURVIVING TRANSTHYRETIN FAMILIAL AMYLOID POLYNEUROPATHY
Teresa Coelho, Porto, Portugal

OP3_7 NEUROTOXIC 1-DEOXYSPHINGOLIPIDS ARE NATIVELY DEGRADED BY A CYP4F DEPENDENT PATHWAY
Thorsten Hornemann, Zurich, Switzerland

OP3_8 A ZEBRAFISH MODEL FOR SMALL-FIBER NEUROPATHY
Ivo Eijkenboom, Maastricht, The Netherlands

OP3_9 A DOG SPONTANEOUS MODEL FOR HUMAN SENSORY NEUROPATHIES: IDENTIFICATION OF A MUTATION IN THE UPSTREAM REGION OF A NEUROTROPHIC FACTOR
Catherine André, Rennes, France

OP3_10 CELLULAR PATHOMECHANISMS OF HEREDITARY SENSORY NEUROPATHY TYPE I (HSN-1) IN MAMMALIAN MOTOR NEURONS
Emma Wilson, London, United Kingdom

OP3_11 CLINICAL AND NEUROPHYSIOLOGICAL PROFILE OF PERIPHERAL NEUROPATHY IN AICARDI-GOUTIÈRES SYNDROME
Manoj Menezes, Sydney, Australia

OP3_12 ESTIMATE PENETRANCE IN HEREDITARY DISORDERS USING A NON-PARAMETRIC APPROACH: NEW INSIGHTS IN VAL30MET TRANSTHYRETIN (TTR) FAMILIAL AMYLOID POLYNEUROPATHY (FAP)
Violaine Plante-Bordeneuve, Creteil, France

12.30 - 13.30 POSTER SESSION 3

P3_1 THE NATURAL HISTORY OF TRANSTHYRETIN FAMILIAL AMYLOID POLYNEUROPATHY: AN ANALYSIS FROM THE TRANSTHYRETIN AMYLOIDOSIS OUTCOMES SURVEY
Teresa Coelho, Porto, Portugal

P3_2 TRANSITION FROM ASYMPTOMATIC TO SYMPTOMATIC TRANSTHYRETIN FAMILIAL AMYLOID POLYNEUROPATHY: AN ANALYSIS FROM THE TRANSTHYRETIN AMYLOIDOSIS OUTCOMES SURVEY
Teresa Coelho, Porto, Portugal

P3_3 TRANSTHYRETIN-RELATED AMYLOIDOSIS IN THE MEDITERRANEAN AND BALKAN AREA: FOCUS ON THE GLU89GLN MUTATION
Anna Mazzeo, Messina, Italy

P3_4 REHABILITATION OF PATIENTS WITH TRANSTHYRETIN FAMILIAL AMYLOID POLYNEUROPATHY
David Adams, Le Kremlin-Bicêtre, France

P3_5 CLINICAL AND NEUROPHYSIOLOGICAL CHARACTERIZATION OF TTRVAL30MET FAMILIAL AMYLOID POLYNEUROPATHY IN A BRAZILIAN TERTIARY CENTER OF PERIPHERAL NEUROPATHIES
Wilson Marques Júnior, Ribeirão Preto, Brazil

P3_6 GENETIC EPIDEMIOLOGY OF TRANSTHYRETIN FAMILIAL AMYLOID POLYNEUROPATHY IN A BRAZILIAN TERTIARY CENTER OF PERIPHERAL NEUROPATHIES
Wilson Marques Júnior, Ribeirão Preto, Brazil

P3_7 DIFLUNISAL COMPASSIVE USE IN TRANSTHYRETIN FAMILIAL AMYLOIDOTIC POLYNEUROPATHY: REPORT OF A FIRST SPANISH EXPERIENCE
Sebastian Azorin, Barcelona, Spain

P3_8 SYMPTOM PRESENTATION OF PATIENTS WITH TTR MUTATIONS IN IOWA
Shawna Feely, Iowa City, United States

P3_9 A COHORT OF ITALIAN FAMILIAL AMYLOID POLYNEUROPATY PATIENTS: TRANSTHYRETIN MUTATIONAL SPECTRUM
Paola Rimessi, Ferrara, Italy

P3_10 MOLECULAR GENETICS BACKGROUND AND CLINICAL FEATURES OF INHERITED POLYNEUROPATHY PLUS SYNDROMES-STUDY OF 8 FAMILIES
Andrzej Kochaski, Warsaw, Poland

P3_11 NATURAL HISTORY STUDY IN HEREDITARY SENSORY NEUROPATHY TYPE 1 (HSN1)
Umaiyal Kugathasan, London, United Kingdom

P3_12 PAINFUL SMALL FIBRE NEUROPATHY IN TYPE 1 GAUCHER DISEASE
Grazia Devigili, Udine, Italy

P3_13 HEREDITARY GELSOLIN AMYLOIDOSIS (HGA) IN AN ITALIAN FAMILY: CLINICAL, ELECTROPHYSIOLOGICAL AND GENETIC FEATURES
Anna Sagnelli, Milan, Italy

P3_14 CLINICAL AND GENETIC CHARACTERIZATION IN A LARGE CASE SERIES OF CHILDHOOD ONSET HEREDITARY PERIPHERAL NEUROPATHIES
Isabella Moroni, Milan, Italy

P3_15 DE NOVO PMP2 MUTATIONS IN FAMILIES WITH TYPE 1 CHARCOT-MARIE-TOOTH DISEASE
Steven Scherer, Philadelphia, United States

P3_16 HEREDITARY SENSORY ATAXIC NEUROPATHY ASSOCIATED WITH PROXIMAL MUSCLE WEAKNESS IN THE LOWER EXTREMITIES: A NEW CLINICAL ENTITY?
Tatsufumi Murakami, Kurashiki, Japan

P3_17 AXONAL PERIPHERAL NEUROPATHY PREDOMINANT PATIENTS WITH KIF5A MUTATIONS
Da Eun Nam, Gongju, South Korea

P3_18 AN ONLINE SURVEY OF NEUROLOGISTS ABOUT CHARCOT-MARIE-TOOTH DISEASE TYPE 1A
Xavier Paoli, Issy Les Moulineaux, France

P3_19 A 10-YEAR CLINICO-ELECTROPHYSIOLOGICAL AND LOWER-LIMB MUSCLE MRI LONGITUDINAL STUDY IN CHARCOT-MARIE-TOOTH DISEASE TYPE 1A DUPLICATION
Ana Lara Pelayo-Negro, Santander, Spain

P3_20 DIAGNOSTIC EXPERIENCE FROM A LARGE CHARCOT-MARIE-TOOTH CLINIC
Janel Phetteplace, Iowa City, United States

P3_21 A NOVEL PATHOGENIC RAB7 MUTATION CAUSING PREDOMINANTLY MOTOR CMT2B
Paola Saveri, Milan, Italy

P3_22 CLINICAL FINDINGS FROM A LARGE SERIES OF CMT2I PATIENTS WITH MPZ P70S MUTATION
Chiara Pisciotta, Milan, Italy

P3_23 NOVEL HSJ1 MUTATION IN AN ITALIAN CMT2 FAMILY WITH HEARING LOSS
Chiara Pisciotta, Milan, Italy

P3_24 SUBCLINICAL SMALL FIBER INVOLVEMENT IN CMT4D
Giuseppe Piscosquito, Telese Terme (BN), Italy

P3_25 LONGITUDINAL EVALUATION OF THE HAND FUNCTION IN PATIENTS AFFECTED BY CHARCOT-MARIE-TOOTH (CMT) NEUROPATHY WITH A SENSOR ENGINEERED GLOVE TEST (SEGT)
Valeria Prada, Genoa, Italy

P3_26 TESTING OVERWORK WEAKNESS IN CHARCOT-MARIE-TOOTH (CMT) DISEASE: IS IT TRUE OR FALSE?
Valeria Prada, Genoa, Italy

P3_27 A GENOMIC APPROACH TO IDENTIFY NEW GENES RESPONSIBLE FOR INHERITED MOTOR AND CMT2 NEUROPATHIES: A COLLABORATIVE STUDY
Stefano Carlo Previtali, Milan, Italy

P3_28 INVESTIGATION OF AXONAL TRANSPORT AND MYELINATION DEFECTS IN TWO IN VITRO SYSTEMS OF CHARCOT-MARIE-TOOTH DISEASE TYPE 1A AND MODIFICATION THROUGH SELECTIVE HISTONE DEACETYLASE 6 INHIBITION
Robert Prior, Leuven, Belgium

P3_29 AN EXPERIMENTAL TRIAL OF AN EARLY ONSET SHORT-TERM TREATMENT WITH A COMBINATIONAL DRUG (PXT3003) IN THE CHARCOT-MARIE-TOOTH 1A RAT MODEL
Thomas Prukop, Göttingen, Germany

P3_30 PATIENT & PUBLIC INVOLVEMENT: HOW SERVICE USER ENGAGEMENT HAS INFORMED RESEARCH INTO FALLS INTERVENTIONS IN PEOPLE WITH CHARCOT-MARIE-TOOTH DISEASE
Gita Ramdharry, London, United Kingdom

P3_31 ALTERATIONS OF INTRALYSOSOMAL PH IN FIG4-DEFICIENT CELLS
Vignesh Ravi, Nashville, United States

P3_32 FUNCTIONAL ANALYSIS AND GENOME-WIDE RNA-SEQ OF HUMAN MOTOR NEURONS IMPLICATE SELECTIVE MITOCHONDRIAL DEPLETION, RESISTANCE TO APOPTOSIS AND INCREASED MITOPHAGY IN CHARCOT-MARIE-TOOTH 2A
Federica Rizzo, Milan, Italy

P3_33 A HOMOZYGOUS RETICULON 2 MUTATION IS A CAUSE OF DHMN WITH PYRAMIDAL SIGNS
Alexander Rossor, London, United Kingdom

P3_34 A MISSENSE MUTATION IN THE MITOCHONDRIAL ENCODED TRNA SERINE 2 (AGY); A POTENTIAL GENETIC MODIFIER IN CMT2
Alexander Rossor, London, United Kingdom

13.30 - 14.30 Lunch

14.30 - 15.15 PLATFORM 9 - OUTCOME MEASURES
Chairs: Joshua Burns (Westmead, Australia) - Matilde Laurà (London, United Kingdom)

09_1 RESPONSIVENESS OF GAIT ANALYSIS PARAMETERS IN A COHORT OF 71 CMT SUBJECTS
Giuseppe Piscosquito, Telese Terme (BN), Italy

09_2 QUANTIFICATION OF INTRAMUSCULAR FAT ACCUMULATION IN CMT1A USING MRI: AN INTERNATIONAL LONGITUDINAL STUDY
Jasper Morrow, London, United Kingdom

09_3 CHARCOT-MARIE-TOOTH DISEASE INFANT SCALE: REPORT ON PROGRESS AND FINAL VERSION FOR VALIDATION
Melissa Mandarakas, Sydney, Australia

15.15 - 16.45 PLATFORM 10 - THERAPIES
Chairs: John Svaren (Madison, United States) - Maurizio D'Antonio (Milan, Italy)

010_1 NIACIN-MEDIATED TACE ACTIVATION AMELIORATES CMT NEUROPATHIES WITH FOCAL HYPERMYELINATION
Alessandra Bolino, Milan, Italy

010_2 INTRATHECAL GENE THERAPY IN A NEUROPATHY MODEL EXPRESSING A CMT1X MUTATION
Alexia Kagiava, Nicosia, Cyprus

O10_3 IFB-088 A POTENTIAL NEW THERAPEUTIC OPTION TO TREAT DEMYELINATING CHARCOT-MARIE-TOOTH DISEASES
Philippe Guedat, Nantes, France

O10_4 LECITHIN THERAPY IMPROVES DISEASE PROGRESSION IN A RAT MODEL OF CHARCOT-MARIE-TOOTH DISEASE 1A
Robert Fledrich, Göttingen, Germany

O10_5 AN EXPERIMENTAL TRIAL OF AN LATE ONSET LONG-TERM TREATMENT WITH TUMERIC AND MERIVA® CURCUMIN IN THE CHARCOT-MARIE-TOOTH 1A RAT MODEL
Thomas Prukop, Göttingen, Germany

O10_6 AEROBIC EXERCISE IN PATIENS AFFECTED BY CHARCOT-MARIE-TOOTH (CMT) NEUROPATHY: RESULTS OF A RANDOMIZED, SINGLE BLIND, CONTROLLED STUDY
Laura Mori, Genoa, Italy

16.45 - 17.15 Coffee break

17.15 - 18.35 ORAL POSTER 4
Chairs: Michael Sereda (Goettingen, Germany) – Fiore Manganelli (Naples, Italy)

OP4_1 FUNCTIONAL STUDIES OF DCTN2 – PROBABLY A NEW INTERMEDIATE CHARCOT-MARIE-TOOTH GENE
Geir Julius Braathen, Skien, Norway

OP4_2 GANGLIOSIDE INDUCED DIFFERENTIATION ASSOCIATED PROTEIN 1 MUTATIONS IN SPAIN, A NATIONWIDE STUDY
Rafael Sivera, Valencia, Spain

OP4_3 DISEASE PROGRESSION IN CHARCOT-MARIE-TOOTH DISEASE TYPE 1A: A LONGITUDINAL STUDY USING RASCH ANALYSIS-BASED WEIGHTED CMT NEUROPATHY SCORES
Vera Fridman, Boston, United States

OP4_4 DETERIORATION IN GAIT AND FUNCTIONAL AMBULATION IN CHILDREN AND ADOLESCENTS WITH CHARCOT-MARIE-TOOTH DISEASE: A LONGITUDINAL STUDY
Rachel Kennedy, Parkville, Australia

OP4_5 IN SEARCH OF MODIFIERS OF CMT1A AND HNPP
Frank Baas, Amsterdam, The Netherlands

OP4_6 CMT4G: A LARGE SERIES OF FRENCH PATIENTS
Raul Juntas Morales, Montpellier, France

OP4_7 BLINK REFLEX ROLE IN ALGORITHMIC GENETIC TESTING OF INHERITED POLYNEUROPATHIES
Christopher Klein, Rochester, United States

OP4_8 NOVEL OUTCOME MEASURES FOR CHARCOT-MARIE-TOOTH DISEASE: VALIDATION, RELIABILITY AND SENSITIVITY TO CHANGES OF 6-MINUTE WALK TEST AND STEPWATCHTM ACTIVITY MONITOR AND IDENTIFICATION OF THE WALKING FEATURES MORE RELATED TO A BETTER QUALITY OF LIFE
Luca Padua, Rome, Italy

OP4_9 EVALUATING THE BENEFITS OF COMMUNITY BASED AEROBIC TRAINING ON THE PHYSICAL HEALTH AND WELL-BEING OF PEOPLE WITH CHARCOT-MARIE-TOOTH DISEASE TYPE 1A
Gita Ramdharry, London, United Kingdom

OP4_10 EFFICACY OF FOCAL MECHANIC VIBRATION TREATMENT ON BALANCE IN CHARCOT- MARIE-TOOTH 1A DISEASE:
A PILOT STUDY
Costanza Pazzaglia, Milan, Italy

OP4_11 THE AGEING OF CMT1A PATIENTS
Stefano Tozza, Naples, Italy

OP4_12 TESTING THE PHARMACOLOGICAL EFFECTS ON CMT1A FIBER STRUCTURES: A COMPREHENSIVE EVALUATION OF IN VITRO MYELINATION
Davide Visigalli, Genoa, Italy

OP4_13 NORMATIVE AEROBIC EXERCISE VALUES IN CMT
Gita Ramdharry, London, United Kingdom

OP4_14 SCREENING FOR INTERACTIONS BETWEEN VIRALLY DELIVERED CX32 AND NEUROPATHY-ASSOCIATED MUTANTS: TOWARDS A GENE THERAPY FOR CMT1X
Styliana Kyriakoudi, Nicosia, Cyprus

OP4_15 A DYNC1H1 MUTATION IN AUTOSOMAL DOMINANT SPINAL MUSCULAR ATROPHY SHOWS THE POTENTIAL OF PHARMACOLOGICAL INHIBITION OF HISTONE DEACETYLASE 6 AS A TREATMENT FOR DISEASE ASSOCIATED CELLULAR PHENOTYPES
Fabio Simoes, Brighton, United Kingdom

OP4_16 TUNING ACTIN POLYMERIZATION TO RESCUE ABNORMAL MYELIN PERMEABILITY IN HNPP
Bo Hu, Nashville, United States

18.35 - 19.35 POSTER SESSION 4

P4_1 FOUND A NEEDLE IN A HAYSTSCK! DIAGNOSTIC PATHWAY IN OUR NEUROPHYSIOLOGICAL OUTPATIENT CLINIC FROM A GENERAL SUSPICTION OF CARPAL TUNNEL SYNDROME TO THE CONFIRMATION OF HEREDITARY NEUROPATHY
Tiziana Rosso, Castelfranco Veneto, Italy

P4_2 NERVE ULTRASOUND IN DIFFERENT CMT TYPES
Daniele Coraci, Rome, Italy

P4_3 A NERVE ULTRASOUND EVALUATION IN PATIENTS WITH FRIEDREICH'S ATAXIA
Alessandro Salvalaggio, Padova, Italy

P4_4 PERIPHERAL NERVE ULTRASOUND IN CHILDREN WITH DÉJÉRINE-SOTTAS DISEASE
Monique Ryan, Parkville, Australia

P4_5 NERVE ULTRASOUND FINDINGS IN A COHORT OF PATIENTS WITH MPZ-RELATED CHARCOT-MARIE- TOOTH NEUROPATHIES
Stefano Tamburin, Verona, Italy

P4_6 ARE GABA-B LIGANDS OF THERAPEUTIC INTEREST FOR CMT1A? NEW INSIGHTS FOR DECIPHERING THEIR MECHANISMS OF ACTION
Valerio Magnaghi, Milan, Italy

P4_7 TARGETED MULTI-GENE PANELS AS A TOOL FOR DIAGNOSTICS IN CMT: FIRST RESULTS
Anja Schirmacher, Muenster, Germany

P4_8 CLINICAL AND NEUROPHYSIOLOGICAL CHARACTERISTICS OF THE ASSOCIATION BETWEEN CHARCOT-MARIE-TOOTH 1A AND PRE DIABETES OR DIABETES MELLITUS IN A BRAZILIAN POPULATION
Juliana Secchin, Cachoeiro de Itapemirim, Brazil

P4_9 NOVEL INF2 GENE MUTATIONS IN CZECH PATIENTS WITH SPORADIC HMSN DETECTED BY GENE PANEL TESTING
Pavel Seeman, Prague, Czech Republic

P4_10 MRI OR MUSCLE ULTRASOUND FOR DIAGNOSING CHARCOT MARIE DISEASE?
Orest Semeryak, Lviv, Ukraine

P4_11 LACK OF FATIGABILITY IN 6 MINUTE WALK TEST FOR CHILDREN WITH CHARCOT-MARIE-TOOTH DISEASE
Rosemary Shy, Iowa City, United States

P4_12 MONITORING PREGNANCY IN CHARCOT-MARIE-TOOTH DISEASE: RESULTS OF A SURVEY
Mariola Skorupinska, London, United Kingdom

P4_13 PLASMA-METABOLITE AND SKIN-PROTEIN SIGNATURES OF CHARCOT-MARIE-TOOTH 1A PROVIDE MOLECULAR MARKERS OF DISEASE AND SUGGEST FUTURE THERAPEUTIC INTERVENTIONS
Francesc Palau, Barcelona, Spain

P4_14 TRANSLATIONAL PROFILING OF MOTOR NEURONS IN TWO MOUSE MODELS OF CHARCOT-MARIE- TOOTH DISEASE TYPE 2D
Emily Spaulding, Bar Harbor, United States

P4_15 POTOCKI-LUPSKI SYNDROME AND CHARCOT-MARIE-TOOTH 1A DISEASE: A RARE ASSOCIATION
Anna Mazzeo, Messina, Italy

P4_16 A NEW MORC2 MUTATION IN A LARGE FAMILY WITH GENDER-RELATED PHENOTYPE VARIABILITY
Tanya Stojkovic, Paris, France

P4_17 DOMINANT TRPV4 MUTATIONS IN HEREDITARY AXONAL NEUROPATHIES
Jeremy Sullivan, Baltimore, United States

P4_18 NOVEL GENES INVOLVED IN NEUROPATHIC PAIN IN PATIENTS
Radek Szklarczyk, Maastricht, The Netherlands

P4_19 A NOVEL MUTATION IN THE 5' UNTRANSLATED REGION OF GAP JUNCTION PROTEIN BETA 1 ASSOCIATED WITH X-LINKED CHARCOT-MARIE-TOOTH IN TWO UNRELATED FAMILY
Federica Taioli, Verona, Italy

P4_20 A COMPOUND HETEROZYGOUS MUTATION IN THE VACCINIA RELATED KINASE-1 GENE IS A CAUSE OF HEREDITARY MOTOR NEUROPATHY WHITH UPPER MOTOR NEURON SIGNS
Pedro J Tomaselli, London, United Kingdom

P4_21 ROLE OF X-BOX BINDING PROTEIN 1 PATHWAY IN CHARCOT-MARIE-TOOTH DISEASE TYPE 1B
Thierry Touvier, Milan, Italy

P4_22 MOLECULAR AND MORPHOLOGICAL SIGNATURE OF SCHWANN CELLS ADHERED TO A NERVE GUIDE: A CLOSER LOOK ON BIOCHEMICAL PROCESSES DURING NERVE REGENERATION
Andreas Roos, Newcastle upon Tyne, United Kingdom

P4_23 SPORT ACTIVITY IN CHARCOT-MARIE-TOOTH DISEASE: FROM A CASE OF A PARALYMPIC SWIMMER TO A PROPOSAL OF SURVEY STUDY ON SPORT BENEFIT PERCEPTION
Giuseppe Vita, Messina, Italy

P4_24 TARGET-ENRICHMENT SEQUENCING AND COPY NUMBER EVALUATION IN INHERITED POLYNEUROPATHY
Christopher Klein, Rochester, United States

P4_25 DEVELOPMENT OF BEST PRACTICE GUIDELINES FOR PAEDIATRIC CHARCOT-MARIE-TOOTH DISEASE
Joshua Burns, Westmead, Australia

P4_26 POLG MUTATIONS IN RECESSIVE CMT2 AND DOMINANT PROGRESSIVE EXTERNAL OPHTHAMOPLEGIA
DaHye Yoo, Gongju, South Korea

P4_27 VOCAL CORD PARALYSIS IN CHARCOT-MARIE-TOOTH TYPE 4B1 DISEASE ASSOCIATED WITH A NOVEL MUTATION IN THE MYOTUBULARIN-RELATED PROTEIN 2 GENE: A CASE REPORT AND REVIEW OF THE LITERATURE
Alberto Andrea Zambon, Milan, Italy

P4_28 MFN2-R94Q TRANSGENIC MICE DEVELOP SENSORIMOTOR DEFECTS AND MITOCHONDRIAL DYSFUNCTION
Yueqin Zhou, Los Angeles, United States

19.35 **PRIZES AND CLOSING**
Vincent Timmerman, Antwerp, Belgium
Steve Scherer, Philadelphia, United States - Angelo Schenone, Genoa, Italy - Davide Pareyson Milan, Italy

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GENERAL INFO

VENUE

The 6th International Charcot-Marie-Tooth and Related Neuropathy Consortium (CMTR) Meeting will take place at NH Laguna Palace (Viale Ancona, 2, 30172 Venice - Mestre).

The Meeting rooms are located in the Congress Centre (ground floor).

The poster sessions will take place in the same area.

ORGANIZING SECRETARIAT

the office

The Meeting Secretariat will be opened as follows:

Thursday 8 September	10.00 - 19.45
Friday 9 September	7.45 - 15.30
Saturday 10 September	8.00 - 19.00

During the Meeting you can reach Veronica Simeone - Meeting Secretariat directly dialing mob +39 335 1249818.

REGISTRATION FEES

Registration fees	**after June 15**
Standard registration (*)	Euro 390,40
Young resident (**)	Euro 390,40
Accompanying person (***)	Euro 170,00

(*) the fee includes: attendance to the scientific sessions, the conference kit, 4 coffee breaks, 2 lunches, welcome cocktail (September 8, 2016), certificate of attendance

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(***) the fee includes: the welcome cocktail on September 8 and gala dinner September on 9 (including transfer and afternoon trip to Venice)

ID BADGE

Your personal ID badge will be ready for you at the Meeting Registration Desk.

For security reasons, delegates, accompanying persons and exhibitors will be asked to wear their ID badges during the whole Meeting and at all social events.

CERTIFICATES OF ATTENDANCE

Certificate should be requested at the end of the Meeting. They will be sent by email one week after the Meeting.

COFFEE BREAKS & LUNCHES - **NH Laguna Palace**

	Coffee break	Lunch	Coffee break
Thursday 8 September	/	/	17.15 - 17.45
Friday 9 September	10.00 - 10.30	12.30 - 13.30	/
Saturday 10 September	11.00 - 11.30	13.30 - 14.30	16.45 - 17.15

WELCOME COCKTAIL - **NH Laguna Palace**

Thursday, 8 September: 19.45

TRIP TO VENICE & GALA DINNER IN SAN SERVOLO (9 September 2016 - SOLD OUT)

A detailed programme will be handed out to all those who have pre-registered to this social event.

PRESENTATIONS

Oral presentation (platform)

The time allotted for Oral presentations **(O)** is 10 minutes + 5 minutes for questions for a total of 15 minutes. Only **PowerPoint presentations** are accepted.

Oral posters (OP)

Oral presentation during the oral poster session: time allotted is 3 minutes to briefly present the main message of your poster + 2 minutes for questions.

Posters (P)

The number on each poster board corresponds to the number assigned in the abstract book.

Poster panels size: 90 cm width and 180 cm height. Pin-heads for mounting the posters will be available at the Secretariat desk.

Posters & Oral Posters

Poster and Oral Poster	Poster viewing
Sessions 1 and 2	Thursday 8 September: 15.00 - Friday 9 September: 14.30
Sessions 3 and 4	Friday 9 September: 15.30 - Saturday 10 September: 19.35

EXHIBITION

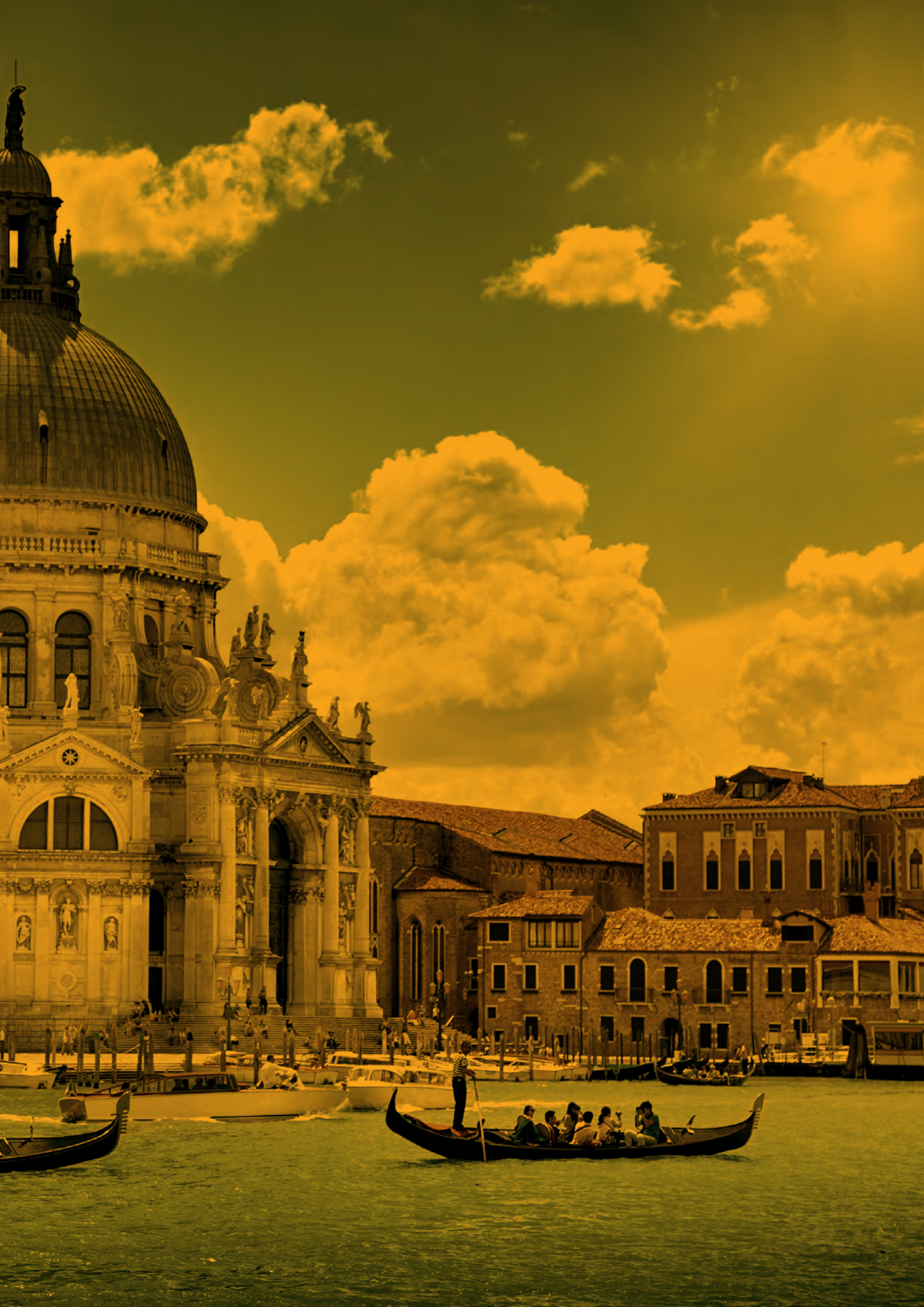
A Technical Exhibition will take place during CMTR 2016.

OPENING HOURS

Thursday 8 September 2016	14.00 - 19.30
Friday 9 September 2016	8.30 - 15.00
Saturday 10 September 2016	8.30 - 18.00

LIABILITY & INSURANCE

The Meeting Secretariat and Organizers accept no responsibility whatsoever for any injury or damage involving persons and property during the Meeting.



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