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

September 8 – 10, 2016
Venice Mestre, Italy
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ORGANIZING SECRETARIAT
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Via San Nicola 14, 34121 Trieste - Italy
cmtr2016@theoffice.it
www.theoffice.it/cmtr2016
# Programme at a glance

## THURSDAY, SEPTEMBER 8, 2016

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<td>10.30-12.30</td>
<td>CMT-ID MEETING (for CMT-ID sites)</td>
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<td>10.00</td>
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<td>14.00</td>
<td>OPENING OF THE MEETING</td>
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| 14.15  | 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  | Coffee break                                                          |
| 17.45  | ORAL POSTER 1                                                         |
| 18.45  | POSTER SESSION 1                                                      |
| 19.45  | WELCOME COCKTAIL                                                      |

## FRIDAY, SEPTEMBER 9, 2016

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<td>PLATFORM 3 PATHOMECHANISMS AND MODELS</td>
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<td>9.00-10.00</td>
<td>PLATFORM 4 PATHOMECHANISMS AND MODELS</td>
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<td>10.30-11.30</td>
<td>PLATFORM 5 PHENOTYPES AND NEW GENES</td>
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<td>13.30</td>
<td>POSTER SESSION 2</td>
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<td>14.30-15.15</td>
<td>PLATFORM 6 EPIDEMIOLOGY AND GENETIC STUDIES</td>
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<td>15.30</td>
<td>TRIP TO VENICE</td>
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<td>20.00</td>
<td>GALA DINNER IN SAN SERVOLO</td>
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## SATURDAY, SEPTEMBER 10, 2016

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

THURSDAY, SEPTEMBER 8, 2016

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

10.00  REGISTRATION OPENING

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

14.15 - 15.00  LECTURE
Presented by Mary 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)

O1_1  REGULATION OF PERIPHERAL MYELIN PROTEIN 22 TRANSCRIPTION
John Svaren, Madison, United States

O1_2  CRYPTIC AMYLOIDGENIC ELEMENTS IN THE 3’ UTR OF THE NEUROFILAMENT HEAVY GENE
TRIGGER CHARCOT-MARIE-TOOTH DISEASE
Adriana Rebelo, Miami, United States

O1_3  EIF2ALPHA PHOSPHORYLATION: A NOVEL HOMEOSTATIC HUB IN PERIPHERAL NEUROPATHIES
Maurizio D’Antonio, Milan, Italy

O1_4  COORDINATION OF GROWTH FACTOR RECEPTOR TRAFFICKING AND CELL PROLIFERATION BY SH3TC2, A PROTEIN
INVOLVED IN CHARCOT-MARIE-TOOTH NEUROPATHY
Vietxuan Phan, Dortmund, Germany

O1_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)

O2_1  EXPANDING THE ALLELIC AND LOCUS HETEROGENEITY OF TRNA SYNTHETASE-RELATED CMT DISEASE
Anthony Antonellis, Ann Arbor, United States
O2_2 THE NUCLEAR CONNECTION OF TYROSYL-TRNA SYNTHETASE TO NEURODEGENERATION
Sven Bervoets, Antwerp, Belgium

O2_3 MUTATION-INDUCED STRUCTURAL OPENING AND ABERRANT INTERACTION LINK TRNA SYNTHETASES TO CHARCOT-MARIE-TOOTH DISEASE
David Blocquel, La Jolla, United States

O2_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, Esplugues del Llobregat, Barcelona, Spain

OP1_4 DYNCH1 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, Florida, 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 RECESSIVE 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, Milano, 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 PALSY 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 GDAPI IN CHARCOT-MARIE-TOOTH DISEASE  
Francesc Palau, Barcelona, Spain

P1_29  DIAGNOSTIC SCREENING OF EIGHTY 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)

O3_1  CHARACTERIZATION OF THE CMT1B-POT124M MOUSE MODEL
Ghjuvan’Ghjacumu Shackleford, Buffalo, United States

O3_2  NERVE EXCITABILITY CHANGES AFTER NaV1.8 CHANNEL BLOCKER TREATMENT IN MICE DEFICIENT OF MYELIN PROTEIN P0
Mihai Moldovan, Copenhagen, Denmark

O3_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

O3_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 (Antwerpen, Belgium) - Alex Rossor (London, United Kingdom)

O4_1  EXPRESSION OF HSPB8.K141N MUTANT LEADS TO AXONOPATHY AND MOTOR DEFICITS IN A NEW TRANSGENIC MOUSE MODEL OF CMT2L
Delphine Bouhy, Antwerp, Belgium

O4_2  CHARACTERIZATION OF AN ATP7AT985I CONDITIONAL KNOCK-IN MOUSE MODEL FOR X-LINKED DISTAL HEREDITARY MOTOR NEUROPATHY
Gonzalo Perez Siles, Sydney, Australia

O4_3  GENOME-WIDE ASSOCIATION STUDY IDENTIFIES POTENTIAL GENETIC MODIFIERS IN CHARCOT-MARIE-TOOTH DISEASE TYPE 1A
Stephan Zuchner, Miami, United States

O4_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 Zuchner (Miami, United States) - Byung-Ok Choi (Seoul, South Korea)

O5_1  GENOTYPE-PHENOTYPE CHARACTERISTICS AND BASELINE NATURAL HISTORY OF CMT2A CAUSED BY MUTATIONS IN THE MFN2 GENE
Alexander Rossor, London, United Kingdom
O5_2 MUTATIONS IN MORC2 GENE CAUSE AXONAL CHARCOT-MARIE-TOOTH DISEASE
Paula Sancho, Valencia, Spain

O5_3 INVESTIGATING THE FUNCTIONAL CONSEQUENCES OF MICRORCHIDIA 2 (MORC2) MUTATIONS CAUSING AXONAL CMT (CMT2Z)
Marina Kennerson, Concord, Australia

O5_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 CHHIO1 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  SPIINAL 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 UBQUITIN 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 NEUROPATHIES 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, Antwerpen, 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, Ribeirao 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 Lorelice, 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 (Milano, Italy) - Marina Kennerson (Concord, Australia)

O6_1 EPIDEMIOLOGIC ASPECTS OF THE CHARCOT-MARIE-TOOTH DIAGNOSIS IN DENMARK; A NATIONWIDE STUDY
Signe Vaaeth, Aarhus, Denmark

O6_2 AN NGS TARGETED-RESEQUENCING APPROACH FOR THE GENETIC DIAGNOSIS OF INHERITED PERIPHERAL DEMYELINATING NEUROPATHIES
Stefania Magri, Milano, Italy

O6_3 THE INHERITED NEUROPATHY VARIANT BROWSER
Stephan Zuchner, Miami, United States

15.30 TRIP TO VENICE

20.00 GALA DINNER - SAN SERVOLO, VENICE
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 7AMYLOID 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, South San Francisco, United States

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

10.15 - 11.00  PLATFORM 8 OTHER NEUROPATHIES
Chairs: Giuseppe Lauria (Milan, Italy) - Peter James 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 DHMNI 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 (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 compassionate 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 polyneuropathy 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)
Umayyal 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 HSJI 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, Milano, 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 Rosser, London, United Kingdom

P3_34 A MISSENSE MUTATION IN THE MITOCHONDRIAL ENCODED TRNA SERINE 2 (AGY): A POTENTIAL GENETIC MODIFIER IN CMT2
Alexander Rosser, 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)

O9_1 RESPONSIVENESS OF GAIT ANALYSIS PARAMETERS IN A COHORT OF 71 CMT SUBJECTS
Giuseppe Piscosquito, Telese Terme (BN), Italy

O9_2 QUANTIFICATION OF INTRAMUSCULAR FAT ACCUMULATION IN CMT1A USING MRI: AN INTERNATIONAL LONGITUDINAL STUDY
Jasper Morrow, London, United Kingdom

O9_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)

O10_1 NIACIN-MEDIATED TACE ACTIVATION AMELIORATES CMT NEUROPATHIES WITH FOCAL HYPERMYELINATION
Alessandra Bolino, Milan, Italy

O10_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
16.45 - 17.15 Coffee break

17.15 - 18.35 ORAL POSTER 4
   Chairs: Michael Sereda (Göttingen, 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 STEPWATCH™ 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 haystack! Diagnostic pathway in our neurophysiological outpatient clinic from a general suspicion 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 Déjerine-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 OPHTHAMOPEGIA
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, Antwerpen, Belgium
Steve Scherer, Philadelphia, United States - Angelo Schenone, Genoa, Italy - Davide Pareyson Milan, Italy
THANKS TO CMTR MEETING SPONSORS

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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 Meeting Secretariat will be opened as follows:

<table>
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<tr>
<th>Date</th>
<th>Time</th>
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<tr>
<td>Thursday 8 September</td>
<td>10.00 – 19.45</td>
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<tr>
<td>Friday 9 September</td>
<td>7.45 – 15.30</td>
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<td>Saturday 10 September</td>
<td>8.00 – 19.00</td>
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During the Meeting you can reach Veronica Simeone - Meeting Secretariat directly dialing mob +39 335 1249818.

REGISTRATION FEES

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<tr>
<td>Standard registration (*)</td>
<td>Euro 390,40</td>
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<td>Young resident (**)</td>
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<td>Accompanying person (***)</td>
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(*)& the fee includes: attendance to the scientific sessions, the conference kit, 4 coffee breaks, 2 lunches, welcome cocktail (September 8, 2016), certificate of attendance

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

(***) 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

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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

<table>
<thead>
<tr>
<th></th>
<th>Poster viewing</th>
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<tbody>
<tr>
<td>Sessions 1 and 2</td>
<td>Thursday 8 September: 15.00 - Friday 9 September: 14.30</td>
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<tr>
<td>Sessions 3 and 4</td>
<td>Friday 9 September: 15.30 - Saturday 10 September: 19.35</td>
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EXHIBITION

A Technical Exhibition will take place during CMTR 2016.

OPENING HOURS

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<tbody>
<tr>
<td>Thursday 8 September</td>
<td>14.00 - 19.30</td>
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<tr>
<td>Friday 9 September</td>
<td>8.30 - 15.00</td>
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<tr>
<td>Saturday 10 September</td>
<td>8.30 - 18.00</td>
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LIABILITY & INSURANCE
The Meeting Secretariat and Organizers accept no responsibility whatsoever for any injury or damage involving persons and property during the Meeting.