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

Programme at a glance

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
14.15 LECTURE
Giampietro Schiavo, Modifying axonal transport as a therapeutic strategy in neuromuscular diseases
15.00-16.15 PLATFORM 1 PATHOMECHANISMS 1
16.15-17.15 PLATFORM 2, PATHOMECHANISMS = ARS
17.15 Coffee break
17.45 ORAL POSTER 1 (12)
18.45 POSTER SESSION 1 (47 posters including 12 oral posters)
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 Coffee break
10.30-11.30 PLATFORM 5 PHENOTYPES AND NEW GENES
11.30 ORAL POSTER 2 (12)
12.30 LUNCH
13.30 POSTER SESSION 2 (47 posters including 12 oral posters)
14.30-15.15 PLATFORM 5 EPIDEMIOLOGY AND GENETIC STUDIES
15.45 TRIP TO VENICE
20.00 GALA DINNER IN SAN SERVOLO

SATURDAY, SEPTEMBER 10, 2016

8.30 LECTURE
Giampaolo Merlini, Unfolding transthyretin amyloidosis
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)
12.30 POSTER SESSION 3 (46 posters including 12 oral posters)
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 (16)
18.35 POSTER SESSION 4 (44 posters including 16 oral posters)
19.35 PRIZES AND CLOSING

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

14.15 - 15.00 LECTURE

1 MODIFYING AXONAL TRANSPORT AS A THERAPEUTIC STRATEGY IN NEUROMUSCULAR DISEASES
Giampietro Schiavo, London UK

15.00 - 16.15 PLATFORM 1. PATHOMECHANISMS

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

30 GDPA1 MUTATIONS IN BRAZILIAN PATIENTS WITH CMT2, CMT2-AR AND CMT4
Fernanda Barbosa Figueiredo, Ribeirão Preto, Brazil

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

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

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

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

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

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

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

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

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

9.00 - 10.00 PLATFORM 4 PATHOMECHANISMS AND MODELS

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

16 GENETIC EPIDEMIOLOGY OF INHERITED PERIPHERAL NEUROPATIES IN BULGARIA
Ivaylo Tarnev, Sofia, Bulgaria

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

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

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

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

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

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

23 ORTHOPAEDIC COMPLICATIONS IN CHARCOT MARIE TOOTH DISEASE: RESULTS OF A PROSPECTIVE STUDY
Matilde Laura, London, United Kingdom

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

15.45 TRIP TO VENICE

20.00 GALA DINNER - SAN SERVOLO, VENICE

SATURDAY, SEPTEMBER 10, 2016

8.30 - 9.15 LECTURE

UNFOLDING TRANSTHYRETIN AMYLOIDOSIS
Giampaolo Merlini, Pavia, Italy

9.15 - 10.15 PLATFORM 7 AMYLOID NEUROPATHY

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

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

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

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

10.15 - 11.00 PLATFORM 8 OTHER NEUROPATHIES

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

4 REHABILITATION OF PATIENTS WITH TRANSTHYRETIN FAMILIAL AMYLOID POLYNEUROPATHY
Agnès Morier, Le Kremlin-Bicêtre, France

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

18.35 - 19.35 POSTER SESSION 4

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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

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, NE1 3BZ, United Kingdom

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

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

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

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

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

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

19.35 PRIZES AND CLOSING