FOUNDATION OF THE SOCIETY FOR THE STUDY OF NEUROPROTECTION AND NEUROPLASTICITY

International School of Neurology

UMF IULIU HAȚIEGANU UNIVERSITY OF MEDICINE AND PHARMACY CLUJ-NAPOCA

RoNEURO Institute for Neurological Research and Diagnostic

FUNDATIA JURNALULUI Journal of Medicine and Life

Seminars

Department of Neurosciences University of Medicine and Pharmacy "Iuliu Hatieganu" Cluj-Napoca | Romania

28 APRIL, 2021

VIRTUAL MEETING

Welcome Address

It is a pleasure to welcome you to the 67th edition Seminars - 28 April, 2021. The seminar is hosted by the Department of Neurosciences, Faculty of Medicine, "Iuliu Hatieganu" University of Medicine and Pharmacy, Cluj-Napoca. This seminar aims to establish itself as a highly useful framework that will enable local specialists to benefit from the expertise of our invited speakers who are part of associated international faculty of our Department of Neurosciences Cluj-Napoca, Romania and RoNeuro Science network. Our scope is to flourish over years and set up an educational vector aiming to meet our junior and senior specialists' needs.

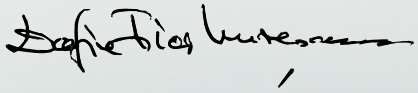
In contrast to large international conferences, the intention behind these seminars is to create an informal and intimate setting, which hopefully will stimulate open discussions.

Due to the uncertainties about the continuing impact of the COVID-19 pandemic, our events will be held in the virtual space, for the time being. As organizers, we would therefore be deeply grateful if you participate and share your time with us.

We are looking forward to your active participation in this educational event!

With consideration,

Prof. Dr. Dafin F. Muresanu,
Chairman Department of Neurosciences, Faculty of Medicine,
"Iuliu Hatieganu" University of Medicine and Pharmacy,
Cluj-Napoca, Romania



Program Coordinator

Dafin F. Mureşanu

President of the European Federation of NeuroRehabilitation Societies (EFNR)

Chairman of EAN Communication and Liaison Committee

Co-Chair EAN Scientific Panel Neurotraumatology

Past President of the Romanian Society of Neurology

Professor of Neurology, Chairman Department of Neurosciences “Iuliu Hatieganu” University of Medicine and Pharmacy, Cluj-Napoca, Romania

Organizers

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Academia de Științe Medicale din România

ACADEMIA ROMANA

UNIVERSITAS MEDICINAE ET PHARMACIAE "CAROLUS DAVILA" BUCURESTIS
DOCENDO DISCITUR
A.D. 1857


SPEAKERS

SPEAKERS

Dafin F. Muresanu
/Romania

Professor of Neurology, Senior Neurologist, Chairman of the Neurosciences Department, Faculty of Medicine, "Iuliu Hatieganu" University of Medicine and Pharmacy Cluj-Napoca, President of the European Federation of Neurorehabilitation Societies (EFNR), Chairman Communication Committee of the European Academy of Neurology (EAN), Past President of the Romanian Society of Neurology, President of the Society for the Study of Neuroprotection and Neuroplasticity (SSNN), Chairman "RoNeuro" Institute for Neurological Research and Diagnostic, Corresponding Member of the Romanian Academy, Member of the Academy of Medical Sciences, Romania and secretary of its Cluj Branch. He is member of 17 scientific international societies (being Member of the American Neurological Association (ANA) - Fellow of ANA (FANA) since 2012) and 10 national ones, being part of the executive board of most of these societies. Professor Dafin F. Muresanu is also a specialist in Leadership and Management of Research and Health Care Systems (specialization in "Management and Leadership, Arthur Anderson Institute, Illinois, USA, 1998"; "MBA – Master of Business Administration - Health Care Systems Management, The Danube University - Krems, Austria, 2003"). He has performed valuable scientific research in high interest fields such as: neurobiology of central nervous system (CNS) lesion mechanisms; neurobiology of neuroprotection and neuroregeneration of CNS; the role of the Blood-brain barrier (BBB) in CNS diseases; developing comorbidities in animal models to be used in testing therapeutic paradigms; nanoparticles neurotoxicity upon CNS; the role of nanoparticles in enhancing the transportation of pharmacological therapeutic agents through the BBB; cerebral vascular diseases; neurodegenerative pathology; traumatic brain injury; neurorehabilitation of the central and peripheral nervous system; clarifying and thoroughgoing study on the classic concepts of Neurotrophicity, Neuroprotection, Neuroplasticity and Neurogenesis by bringing up the Endogenous Defense Activity (EDA) concept, as a continuous nonlinear process, that integrates the four aforementioned concepts, in a biological inseparable manner.

Professor Dafin F. Muresanu is coordinator in international educational programs of European Master (i.e. European Master in Stroke Medicine, University of Krems), organizer and co-organizer of many educational projects: European and international schools and courses (International School of Neurology, European Stroke Organisation Summer School, Danubian Neurological Society Teaching Courses, Seminars - Department of Neurosciences, European Teaching Courses on Neurorehabilitation) and scientific events: congresses, conferences, symposia (International Congresses of the Society for the Study of Neuroprotection and Neuroplasticity (SSNN), International Association of Neurorestoratology (IANR) & Global College for Neuroprotection and Neuroregeneration (GCNN) Conferences, Vascular Dementia Congresses (VaD), World Congresses on Controversies in Neurology (CONy), Danube Society Neurology Congresses, World Academy for Multidisciplinary Neurotraumatolgy (AMN) Congresses, Congresses of European Society for Clinical Neuropharmacology, European Congresses of Neurorehabilitation). His activity includes involvement in many national and international clinical studies and research projects, over 500 scientific participations as "invited speaker" in national and international scientific events, a significant portfolio of scientific articles (231 papers indexed on Web of Science-ISI, H-index: 23) as well as contributions in monographs and books published by prestigious international publishing houses.

SPEAKERS

Prof. Dr. Dafin F. Muresanu has been honoured with: „Dimitrie Cantemir" Medal of the Academy of The Republic of Moldova in 2018, Ana Aslan Award 2018 - "Performance in the study of active aging and neuroscience", for the contribution to the development of Romanian medicine, National Order "Faithful Service" awarded by the President of Romania in 2017; "Iuliu Hatieganu" University of Medicine and Pharmacy Cluj-Napoca, Faculty of Medicine, the "Iuliu Hatieganu Great Award 2016" for the best educational project in the last five years; the Academy of Romanian Scientists, "Carol Davila Award for Medical Sciences / 2011", for the contribution to the Neurosurgery book "Tratat de Neurochirurgie" (vol.2), Editura Medicala, Bucuresti, 2011; the Faculty of Medicine, "Iuliu Hatieganu" University of Medicine and Pharmacy Cluj-Napoca "Octavian Fodor Award" for the best scientific activity of the year 2010 and the 2009 Romanian Academy "Gheorghe Marinescu Award" for advanced contributions in Neuroprotection and Neuroplasticity.

SPEAKERS

Davide Pareyson
/Italy

Education: Degree in Medicine, University of Milan; Board in Neurology, University of Milan, and in Clinical Neurophysiology, University of Pavia, Italy.

Davide Pareyson is a Clinical Neurologist working at the Fondazione IRCCS Istituto Neurologico C.Besta (INCB) of Milan, Italy, where he is currently Head of the Rare Neurodegenerative and Neurometabolic Diseases Unit; he is also Chief of the Functional Department of Rare Neurological Diseases.

His main interest is clinical research on hereditary and acquired peripheral neuropathies and motor neuronopathies, inherited neurological disorders, rare diseases. He performed studies on phenotype-genotype correlation, clinical findings, electrophysiology, neuropathology of hereditary neuropathies (particularly Charcot-Marie-Tooth disease – CMT - and related neuropathies, but also amyloid neuropathy) and other neurogenetic disorders including spinal and bulbar muscle atrophy, hereditary spastic paraplegias, hereditary ataxias, genetic leukodystrophies.

He has been working on the development of outcome measures for hereditary neuropathies and other rare diseases and has coordinated and participated in clinical trials and natural history studies in inherited and acquired neuropathies. He coordinated the international trial on ascorbic acid in CMT1A in Italy and UK (Pareyson et al., Lancet Neurol 2011) and the observational trial in patients with ATTR amyloid neuropathy treated with Tafamidis (Cortese et al., J Neurol 2016). He participated/participates in other interventional trials including the following: CMT (comparing two different rehabilitative approaches in CMT, coordinated by A Schenone, Genoa), CIDP (one coordinated by E Nobile Orazio with IVIG and pulse steroids, E Nobile-Orazio et al., Lancet Neurol 2015, and one international coordinated by RAC Hughes, published on Lancet Neurol 2012), TTR Amyloidosis (Helios-A, ongoing).

He is the Coordinator of the Italian National Registries of Charcot-Marie-Tooth disease and of Spino-Bulbar Muscular Atrophy (www.registronmd.it), and participates as local PI in the TTR-related amyloidosis Italian National Registry.

He has co-authored 245 papers on peer-reviewed Journals (Pubmed) mainly on hereditary disorders and neuromuscular diseases. H-index = 42 (Scopus)

Other Experience, Professional Memberships, Honors:
2016-2019 - Chair of the CMTR, Charcot-Marie-Tooth neuropathy & Related diseases consortium
2019-2021 - Member of the Board (as Past-Chair) of the CMTR
2013- 2017 - Member of the Board of the International Peripheral Nerve Society (PNS)
2013-2018 - Member of the Assembly of the European Academy of Neurology (EAN)
2016-2020 - Co-chair of the EAN Scientific Panel on Neuropathies
2016-2018 - Member of the Management Group of the EAN Scientific Panel on Neurogenetics
2013-2015 - Co-chair of the EAN Scientific Panel on Neurogenetics
2013-2014 - Member of the Election Oversight Committee for the EAN

SPEAKERS

2012-2014 - Member of the Executive Committee of the European Neurological Society (ENS)
2006-2013 - Coordinator of the Clinical Neurogenetics Subcommittee of the ENS

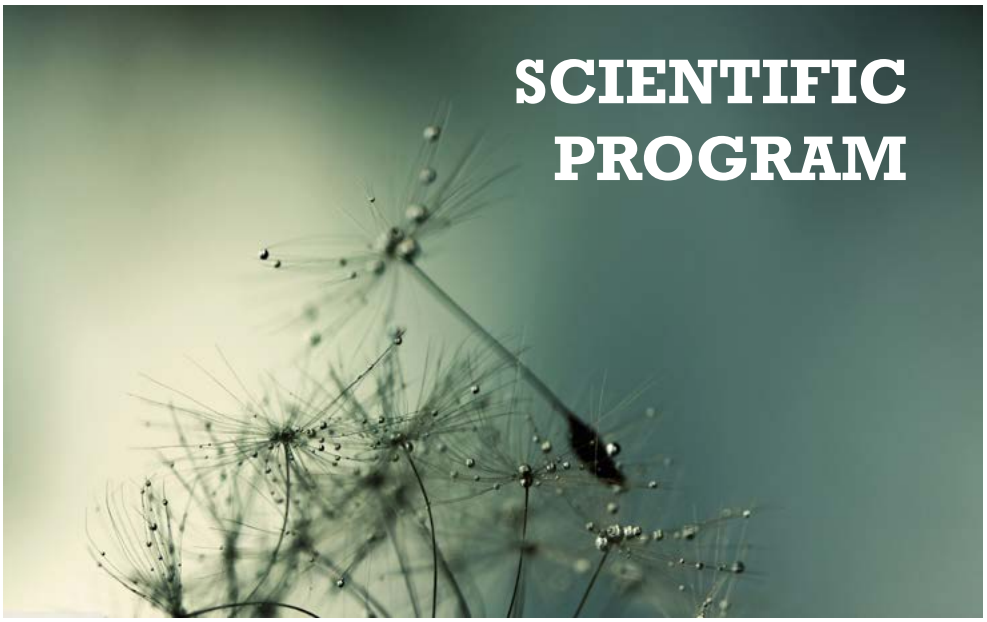
2018- - Chair of the Neuropathy Group of the EURO-NMD ERN (European Reference Network for Neuromuscular Disorders) (since November 2018).
2016-2018 - Deputy Chair of the Neuropathy Group of the EURO-NMD ERN
2020-2022 - Vice-President of the Nervous System Commission of the Scientific Council of AFM- Telethon
2017-2019 - Member of the Nervous System Commission of the Scientific Council of AFM-Telethon
2010-2013 - President of the Italian Peripheral Nerve Society (ASNP)
2013-2016 - Member of the Board of the of the ASNP
2008-2010 - Coordinator the Italian Group for the study of the Peripheral Nervous System (GSSNP)

Member of the Editorial Board of the following Journals: Neurology Genetics (2017-...), Neurological Sciences, Journal of Neuromuscular Diseases; previously J Peripheral Nervous System (until Dec 2016), J Neurology (2008-2012), The Scientific World Journal (2010-2013).

Member of the Italian Neurological Society, European Academy of Neurology (EAN), Peripheral Nerve Society.

Ad Hoc Reviewer for: Nature, Nat Rev Neurol; Brain; Ann Neurol; Neurology; Muscle & Nerve, Neuromuscular Disorders, J Neurol Neurosurg Psychiatry; J Neurol; Hum Mut., European J Neurol, Neurological Sciences, J Peripheral Nervous System, BMC Neurology, Clin Neurol, J Med Genet, J Medical Genetics, Clinical Genetics, Acta Neurologica Scandinavica, J Neurol Sci, Clinical Neurophysiology, Multiple Sclerosis, Clinical Neurology and Neurosurgery, Neurobiology of Disease, Mol Cytogenetics, Journal of Neuromuscular Disorders, Current Opinion in Neurology, etc.; grant reviewer: MDA, AFM, FWO, Wellcome Trust, ABN Clinical Research Training Fellowship, Agence Nationale de la Recherche (ANF), National Institute for Health Research (NIHR, UK).

Research Support
Grants for research activities on CMT, SBMA, hATTR from NIH; AFM-Telethon; Telethon and Telethon-UILMD Italy; AIFA; MDA and CMTA; Regione Lombardia; Ministry of Health


SCIENTIFIC
PROGRAM

Scientific program

28 APRIL, 2021

VIRTUAL MEETING

12:00 - 12:30	Charcot-Marie-Tooth and related neuropathies Davide Pareyson /Italy
12:30 – 13:00	Hereditary TTR-related amyloid neuropathy Davide Pareyson /Italy


ABSTRACTS

Abstracts

CHARCOT-MARIE-TOOTH AND RELATED NEUROPATHIES

DAVIDE PAREYSON
/ITALY

Charcot-Marie-Tooth disease (CMT) is the most frequent hereditary neuromuscular disorder with an estimated prevalence of 10-28:100,000. It is genetically heterogeneous as almost 100 genes and loci have been identified in association with CMT and related neuropathies. It is astonishing to see how diverse are the cellular sub-localisation and the functional roles of the encoded proteins of CMT-associated genes which all lead to similar phenotypes. Myelin formation and maintenance, mitochondrial dynamics, cytoskeleton organization, axonal transport, and vesicular trafficking are the most frequently involved pathways.

CMT is characterised clinically by progressive wasting and weakness of distal limb muscles, variably associated with distal sensory loss, skeletal deformities (ie., pes cavus or planus, less commonly scoliosis), and abnormal deep tendon reflexes. It is classified according to nerve conduction velocities (NCV) and underlying pathology in demyelinating CMT [CMT1 if autosomal dominant (AD), CMT4 if autosomal recessive (AR)], axonal CMT2 (AD or AR), intermediate CMT (which includes the X-linked variety CMTX1 associated with GJB1 mutations, and the less common dominant and recessive intermediate DI-CMT and RI-CMT), and the pure motor forms labelled as distal Hereditary Motor Neuropathies (dHMN). Further subdivision is based on causative genes or identified loci. HNPP (hereditary neuropathy with liability to pressure palsies) and the group of predominantly hereditary sensory (and autonomic) neuropathies (HSAN) are related disorders.

The advent of the novel genetic techniques (NGS) has revolutionized the diagnostic approach improving the diagnostic yield, widening the phenotypic spectrum of several forms, increasing the overlap with other neurogenetic and neuromuscular disorders. Results of NGS needs to be filtered and analysed with great care and the relevance of deep phenotyping is even more important than in the past. A diagnosis may be reached in more than two thirds of the cases and 90% of the mutations are found in 6 genes: PMP22, MPZ, GJB1, MFN2, GDAP1, SH3TC2.

Great advances have been made in defining the natural history of the most prevalent forms, designing outcome measures, studying animal and cellular models to understand the pathomechanisms of the different CMT types, developing and testing possible therapeutic agents. Although we have still no approved pharmacological treatment for CMT and physiotherapy and surgery for skeletal deformities are the only therapeutic intervention available, there is a list of potentially efficacious drugs to be tested in clinical trials in the near future.

Other hereditary neuropathies: we will briefly review less common hereditary disorders where a peripheral neuropathy may be the onset and/or predominant manifestation, including metabolic (mithocondrial, beta-oxidation, haeme, porphyric, peroxisomal, atc) and complex neurodegenerative disorders.

Abstracts

HEREDITARY TTR-RELATED AMYLOID NEUROPATHY

DAVIDE PAREYSON
/ITALY

Hereditary transthyretin amyloidosis (hATTR) is an autosomal dominant disorder due to mutations of the transthyretin (TTR) gene. TTR is synthetized mainly by the liver and released in plasma as a tetrameric transport protein. Mutations in TTR, of which Val30Met is the most common worldwide, cause transthyretin tetramer dissociation, monomer misfolding, and aggregation into insoluble fibrillar proteins in different tissues. Peripheral nerves and heart are the most frequently affected organs, but also eye, leptomeninges and kidneys can be involved. Early diagnosis is fundamental for starting treatment and preventing disease progression in this otherwise lethal disorder; it is easier in familial cases in endemic regions, where Val30Met is by far the predominant mutation, degree of awareness is high, and presentation is typical, i.e., a predominantly small-fibre length-dependent sensory neuropathy with dysautonomia, only later involving motor fibres. Diagnosis is often delayed in non-endemic region where onset occurs much later in life, family history if often negative or misleading, dysautonomia is more subtle, presentation is atypical with a sensory-motor polyneuropathy involving all fibre types (or with other atypical presentations), and progression is definitely faster. The diagnostic pathway and the available treatments will be presented. Orthotopic liver transplantation (OLT) and TTR stabilizers are established treatments. A major breakthrough is represented by the novel gene silencers: Antisense Oligonucleotides (Inotersen) and interfering RNA lipid nanoparticles (Patisiran). Other therapies are under development.

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