













Welcome Address

It is a pleasure to welcome you to the 106th edition Seminars - 17 May, 2023. 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

Dafie Ties huremen

Program Coordinator



Dafin F. Mureşanu

President of the European Federation of NeuroRehabilitation Societies (EFNR)

Secretary General AMN (Academy for Multidisciplinary 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

















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 Neurotraumatology (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 (260 papers indexed on Web of Science-ISI, H-index: 25) as well as contributions in monographs and books published by prestigious international publishing houses. Prof. Dr. Dafin F. Muresanu has been honoured with: "Dimitrie Cantemir" Medal of the Academy of The Republic of



Dafin F.
Muresanu
/Romania

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.

Prof. Antonio Federico, born in Polla (Sa) on the 25.08.48, from 1990 is full professor of Neurology at the University of Siena , Director of the Unit Clinical Neurology and Neurometabolic Disease.

He was Director of the Department of Neurological, Neurosurgical and Behavioural Sciences, University of Siena (2002-2008).

He received the degree in Medicine and specialization in Nervous and Mental Diseases, summa cum laude, at the University of Naples in 1972 and 1975 respectively. He received the Lepetit Award for the best degree dissertation in 1972.

His biological training was in the Institute of Biochemistry as student and after in Physiology of the University of Naples, and in the Centre de Neurochimie of CNRS, in Strasbourg, directed by prof. Mandel where he worked in the years 1973-75. He also collaborated with many international research groups, in different countries where he spent in the past years some times: in Montreal (Prof. Andermann, Karpati and Shoudgbridge), in London (dr A. Harding and prof. Morgan-Hughes), in Toronto (dr.Robinson), in Bonn (prof. von Bergmann), in Paris (dr.Baumann), in Baltimore (proff. Moser and Naidu), in Oxford (prof. Matthews), etc. His clinical formation was made at the Medical School of the University of Naples, in the Dept, Neurology, and after in Siena, where he moved on 1980 with his mentor, prof. G.C. Guazzi. Associated professor in Neurology in 1982, since 1990 he is full professor of Neurology, Medical School, University of Siena. In 2013, he received honoris causa degree in Medicine at University Carol Davila, Bucharest, Rumania.

In the years 1990-96 he was Secretary of the Italian Society of Neurology. In the years 2006-08 was President of the Italian Society of Neurology. He coordinated the Study Group on Clinical Neurogenetics of the Italian Society of Neurology. He has been referee for projects evaluation in the area of Orphan drugs and Orphan diseases for Biomed Projects from EU, for MURST, CNR and Istituto Superiore di Sanita, and other national and international funding agencies, etc.

He is member of the Second Opinion Group of the American Leucodistrophy Association. Associated editor of Neurological Sciences, Springer-Verlag Editor from 2000. From 2012, he is Editor-in Chief.

He is author of more than 500 article quoted by Pubmed. He is author of a chapter on Cerebrotendinous Xanthomatosis, Vinken and Bruyn Edts, Handbook of Clincal Neurology, vol 49, Neurodystrophies and Neurolipidoses.

On the book McKusick's Mendelian Inheritance in Man,. Ed.1992, Catalog of Autosomal Dominant and Recessive Phenotypes he is cited for 3 different diseases. He was editor of the book Late Onset Neurometabolic diseases (A.Federico, K. Suzuki and N.Baumann Edts), Karger 1991, and many other books from Italian and international

Publishing Companies. Recently he published (2015) Manuale di Neurologia Pratica and Neurologia and Assistenza infermieristica, for students.

His main field of interest is related to neurometabolic, neurodegenerative and rare diseases, investigated from a genetic, metabolic, neuroimaging and clinical point of vue.



Antonio Federico

Summary of the academic involvements: - Director of the Section Neurological Sciences, Dept Neurological, Neurosurgical and Behavioural Sciences (2000-2012) - Director of the Research Center for the Diagnosis, Therapy and Prevention of the Neurohandicap and Rare Neurological Diseases, until the 2010 - Vice-Dine of the Medical School, University of Siena (2003-2006) - Director of the Postgraduate School of Neurology, University of Siena, from 2006 up to 2014. - Director of the PhD School in Cognitive and Neurological Sciences, University of Siena (from 2000 up to date) - Coordinator of the Section of the Univ. Siena of the PhD Program Neurosciences, Univ. Florence. - Research delegate for the Dept Medicine, Surgery and Neurosciences (2013-2018) - Vice-Rector of the University of Siena, from 1st april 2016 to november 2017.

Medical Involvements – Until November 2018 (date of retirement) Director of the OU Clinical Neurology and Neurometabolic Diseases, University Hospital of Siena Medical School. –He is still Director of the Regional Reference Center for Rare Diseases - Regional Coordinator of the Network for Rare Neurological Diseases, Tuscany Region. - Member of several Ministry of Health and Regional Committees National and International Commitments - President of the Italian Society of Neurology (2009-11) - Italian delegate to the World Federation of Neurology - Italian Delegate to the European Union of Medical Specialists (Section Neurology) - Italian Delegate and Chairman of the Neuromediterraneum Forum and President - Consultive Member of the European Brain Council - Editor – in – Chief of Neurological Sciences, Springer Verlag Editor. He is in the Editorial Board of many national and international journals. - Member of the American Panel United Leucodystrophies. – Member of the Scientific Committee of AISM (Associazione Italiana Sclerosi Multipla) - Chairman of the Scientific Committee of the European Academy of Neurology (2014-2018) - Chairman of Neuromediterraneum Forum - Co-Chairman of Research group of WFN Migration Neurology.

Member of the Scientific Societies: - Societa Italiana di Neurologia (Past Secretary, President, Past-President and Member of the Committee) - Society for the Inborn Errors of Metabolism - Italian Association of Neuropathology - SINDEM (Italian Association of Dementias) - Italian Association for Parkinson's disease - Italian Association of Neurogeriatrics (Member of the Scientific Committee) - Italian Stroke Forum - European Academy of Neurology (Member of the Board and Chairman of the Scientific Committee) - American Academy of Neurology - World Federation of Neurology (Co-Chair Section of Migration Neurology) - Neuromediterraneum Forum (President).





Scientific program

17 MAY, 2023 VIRTUAL MEETING

13:00 – 13:30	Update of Clinical and pathogenic aspects of inherited small vessel diseases Antonio Federico /Italy	
13:30 – 14:10	Syndromes of mineral accumulation into the brain Antonio Federico /Italy	
14:10 – 14:50	Leucoencephalopathies as a model of inherited primary glial cell degeneration	





Abstracts

SYNDROMES OF MINERAL ACCUMULATION INTO THE BRAIN: CLINICAL AND PATHOGENETIC ASPECTS

ANTONIO FEDERICO /ITALY

We will report several clinical conditions in which the main characteristic is the mineral accumulation into the brain, mainly in basal nuclei, clinically characterized by different severity of parkinsonism, mental deterioration, psychiatric abnormalities.

Mineral accumulation is due to cooper, in Wilson's disease, a well known epatolenticular degeneration, iron in a recently described syndrome with dystonia/parkinsonism and in patothenate kinase deficiency (Hallervorden-Spatz disease), calcium in several mitochondrial diseases, in the so called Fahr syndrome, now better known as Primary familial brain calcification. We will describe the different clinical presentations, the pathogenetic aspects and the recent data on the molecular diagnosis.

We will also report several other more rare conditions, useful for the differential diagnosis and we will describe a diagnostic algorithm for diagnosis.

UPDATE OF CLINICAL AND PATHOGENIC ASPECTS OF INHERITED SMALL VESSEL DISEASES

Genetic ischemic cerebral subcortical small vessel diseases (SSVD) are rare, usually autosomal dominant conditions related to impairment of proteins mainly involved in small vessel functions. Symptoms are characterized by combinations of migraine with aura, ischemic events (transient ischemic attacks, lacunar strokes) and progressively worsening ischemic lesion load in brain imaging, vascular cognitive impairment (usually of the frontal-subcortical type) with behavioral-psychiatric symptoms and bilateral pyramidal and pseudobulbar signs leading to severe disability and premature death. In some patients, microbleeds and hemorrhagic strokes may be evident. A large clinical heterogeneity is usually present.

Between the different forms the most frequent is CADASIL, due to mutations of the NOTCH3 gene, followed by COL4A1/A2-related disease, autosomal dominant forms of HTRA1-related disease and leucoencephalopathies with calcifications and cysts. CARASIL, with an autosomal recessive HTRA1 mutation, is less frequent. A new form has been recently described, named CARASAL.

Here we will report our experience with these patients describing recent data on their pathogenesis and some guideline on the diagnosis and therapeutic options. ANTONIO FEDERICO /ITALY

Abstracts

GENETIC LEUCODYSTROPHIES AS A MODEL OF OLIGODENDROCYTE DYSFUNCTION

ANTONIO FEDERICO
/ITALY

Leukodystrophies are a group of orphan genetic diseases that primarily affect the white matter (WM) of the brain. Glialcellsplayamajorrole in the structural, metabolic and trophic support of axons.

Diversity of the genetically determined defects that interfere with glial cell functions explain the large heterogeneity of leucodystrophies that may be classified:

- According to neuropathology (staining: ortochromatic, metachromatic, sudanophilic; site of demyelination: sparing U fibres,etc; associated findings)
- According with clinical aspects (peripheral nerve, muscle, eye involvement, macrocephaly, tendinous xanthomas, premature aging,, skin and bone changes, endocrine involvement: adrenocortical or ovarian insufficiency, diabetes, etc)
- According to biochemical abnormalities
- According to molecular genetic abnormalities.

We will report the main well known forms (Adrenoleucodystrophy, Metachromatic Leucodystrophy, Krabbe Disease) and some rarer conditions as Vanishing White Matter disease, Vacuolating Leucodystrophy, Alexander disease, Spheroid leukoencephalopathy, etc, and also some recently identified forms, describing the clinical findings for clinical suspicion and the pathogenetic mechanisms.





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