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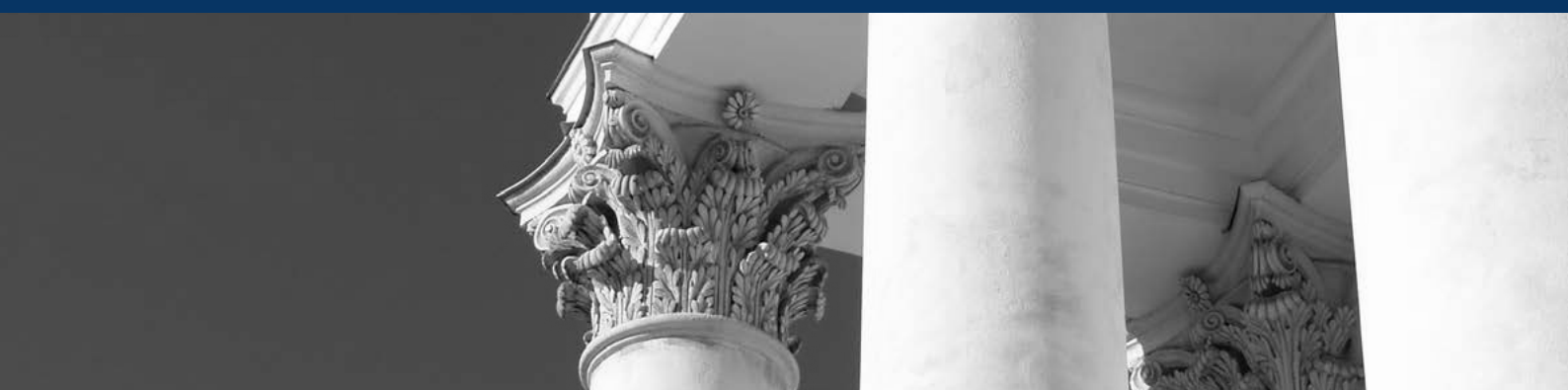
IULIU HAȚIEGANU
UNIVERSITY OF
MEDICINE AND PHARMACY
CLUJ-NAPOCA
ROMANIA

"IULIU HATIEGANU" UNIVERSITY OF MEDICINE AND PHARMACY DOCTORAL SCHOOL

NEUROSCIENCE

PROGRAM

2015-2016 | SECTION 4 | MAY 6TH, 2016



PhD NEUROSCIENCE PROGRAM COORDINATOR

Dafin F. Mureşanu

President of the Romanian Society of Neurology

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

President of the Society for the Study of Neuroprotection and
Neuroplasticity (SSNN)

INTERNATIONAL GUEST LECTURER

Antonio Federico

Department of Medicine, Surgery and Neurosciences,
Medical School, University of Siena
Siena, Italy

Vice-Rector of the University of Siena

Chairman of the Scientific Committee and Member of
the Board of the European Academy of Neurology

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2015-2016

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University of Medicine and Pharmacy
"Iuliu Hatieganu", Cluj Napoca, Romania
www.umfcluj.ro

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COURSE PROGRAM

COURSE PROGRAM

MAY 6TH, 2016

"PROF. DR. MIRCEA DOROFTEIU" AUDITORIUM

23 GHEORGHE MARINESCU STREET, 1ST FLOOR | CLUJ-NAPOCA | ROMANIA

10:00 – 10:10	Dafin F. Muresanu /Romania Welcome Address
10:10 – 11:00	Antonio Federico /Italy Genetic leucodystrophies as a model of oligodendrocyte dysfunction
11:00 – 11:50	Antonio Federico /Italy What it is new on neurometabolic extrapyramidal diseases?
11:50 – 12:40	Antonio Federico /Italy The role on the European Neurological Society in the promotion of research and care of Rare Neurologic Diseases, a Pandora box for Neurology and Neurosciences
12:40 – 13:10	Coffee Break
13:10 – 14:00	Antonio Federico /Italy Update on treatment of neurometabolic genetic diseases
14:00 – 14:50	Antonio Federico /Italy Update of Inherited small vessel diseases
14:50 – 15:00	Discussions

INTERNATIONAL GUEST LECTURER

ANTONIO FEDERICO
ITALY

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 Sanità, 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 in the past 3 years. 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.

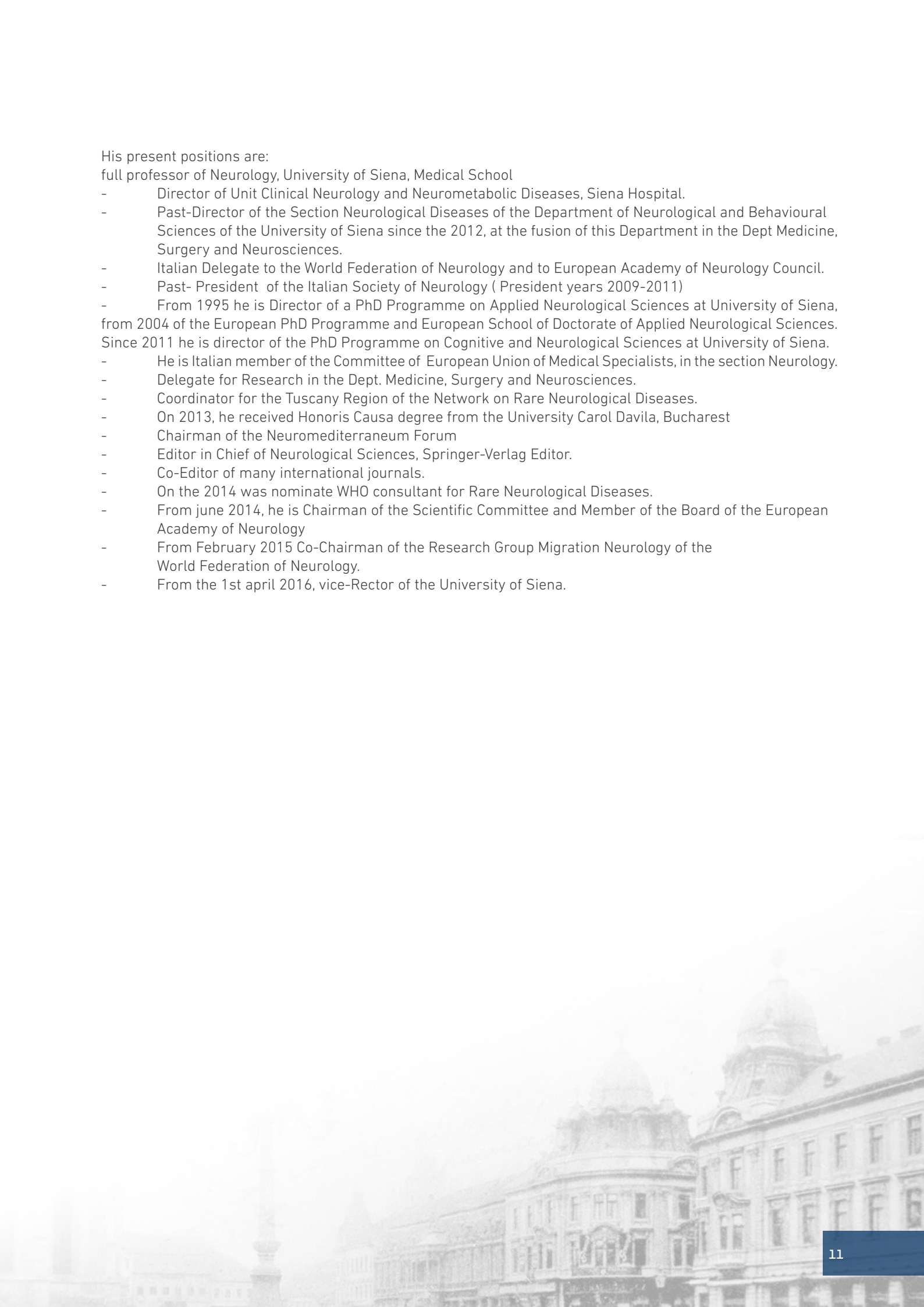
Summary of the academic involvements:
- Director of the Section Neurological Sciences, Dept Neurological , Neurosurgical and Behavioural Sciences (2000-2012)
- Director of the Research Center for the Diagnosis, Therapy and Prevention of the Neurohandicap and Rare Neurological Diseases, until the 2010
- 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-)
- Vice-Rector of the University of Siena, from 1st april 2016.

Medical Involvements
- Director of the OU Clinical Neurology and Neurometabolic Diseases, University Hospital of Siena Medical School.
- 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
- Chairman of Neuromediterraneum Forum
- Co-Chairman of Research group of WFN Migration Neurology

Member of the Scientific Societies:
- Società 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)

His present positions are:
full professor of Neurology, University of Siena, Medical School

- Director of Unit Clinical Neurology and Neurometabolic Diseases, Siena Hospital.
- Past-Director of the Section Neurological Diseases of the Department of Neurological and Behavioural Sciences of the University of Siena since the 2012, at the fusion of this Department in the Dept Medicine, Surgery and Neurosciences.
- Italian Delegate to the World Federation of Neurology and to European Academy of Neurology Council.
- Past- President of the Italian Society of Neurology (President years 2009-2011)
- From 1995 he is Director of a PhD Programme on Applied Neurological Sciences at University of Siena, from 2004 of the European PhD Programme and European School of Doctorate of Applied Neurological Sciences. Since 2011 he is director of the PhD Programme on Cognitive and Neurological Sciences at University of Siena.
- He is Italian member of the Committee of European Union of Medical Specialists, in the section Neurology.
- Delegate for Research in the Dept. Medicine, Surgery and Neurosciences.
- Coordinator for the Tuscany Region of the Network on Rare Neurological Diseases.
- On 2013, he received Honoris Causa degree from the University Carol Davila, Bucharest
- Chairman of the Neuromediterraneum Forum
- Editor in Chief of Neurological Sciences, Springer-Verlag Editor.
- Co-Editor of many international journals.
- On the 2014 was nominate WHO consultant for Rare Neurological Diseases.
- From june 2014, he is Chairman of the Scientific Committee and Member of the Board of the European Academy of Neurology
- From February 2015 Co-Chairman of the Research Group Migration Neurology of the World Federation of Neurology.
- From the 1st april 2016, vice-Rector of the University of Siena.



ABSTRACTS

GENETIC LEUCODYSTROPHIES AS A MODEL OF OLIGODENDROCYTE DYSFUNCTION

ANTONIO FEDERICO
Dept Medicine, Surgery and Neurosciences, Medical School, University of Siena, Italy

Leukodystrophies are a group of orphan genetic diseases that primarily affect the white matter (WM) of the brain. Glial cells play a major role 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 describe the main well known forms (Adrenoleucodystrophy, Metachromatic Leucodystrophy, Krabbe Disease) and some rarer conditions as Vanishing White Matter disease, Vacuolating Leucodystrophy, Alexander disease, etc, describing the clinical findings for clinical suspicion and the pathogenetic mechanisms.

WHAT IT IS NEW ON NEUROMETABOLIC EXTRAPYRAMIDAL DISEASES?

ANTONIO FEDERICO
Dept Medicine, Surgery and Neurosciences, Medical School, University of Siena, Italy

We will report the more recent data on the metabolic basis of several disorders mainly involving basal ganglia, with particular regards to the different forms of Parkinsonism, dystonia, etc, with particular emphasis of the mitochondrial hypothesis of oxidative stress on the basis of the different forms of genetic Parkinson's forms, related to different mutations.
Within the metal related basal ganglia deposits, we will report our experience in Wilson diseases, in the recently described by our group Manganese Trasport Protein Deficiency, in Calcium deposition in the basal ganglia, etc.
In summary we will discuss on extrapyramidal diseases related to:

- Disorders of Heavy Metal Metabolism
- Disorders of Neurotransmitter Metabolism
- Disorders of energy metabolism
- Lysosomal Diseases
- Disorders of intermediary metabolism
- Disorders of mechanism of DNA Damage and Repair

For all of them we will describe clinical signs, diagnostic work-up and possible therapeutic strategies.

THE ROLE ON THE EUROPEAN NEUROLOGICAL SOCIETY IN THE PROMOTION OF RESEARCH AND CARE OF RARE NEUROLOGIC DISEASES, A PANDORA BOX FOR NEUROLOGY AND NEUROSCIENCES

ANTONIO FEDERICO
Dept Medicine, Surgery and Neurosciences, Medical School, University of Siena, Italy

Rare Neurological diseases are a Pandora Box for Neurology.
The list of the Rare diseases encloses more then 5000 disorders, half of them have a neurological interest, with involvement of the Central and Peripheral nervous system or Muscle or all.
They are underdiagnosticated and a global effort is necessary to improve their knowledge, the possibility to have a correct diagnosis by dissemination of information and culture on them and research, leading to possible treatments (the majority of them are without treatments and in all countries has started a cooperative effort for "orphan drugs".

In USA, since 30 years ago has been stimulated the interest on these disorders, followed 10 year later by the European Community.

Several Scientific Societies have started to have a promoting role on this field.
Since Neurology , as speciality, has the major role in the diagnosis and care of this disease, and basic and applied neurosciences in the research on their pathogenesis, EAN (European Academy of Neurology) have the main responsibility for the promotion of the knowledge of these disorders, of the informations and of the research within the neurological community in Europe.

The Scientific Committee of the EAN have organized a Task force on Rare Neurologic Diseases that will have a strict relationship with the Subspecialities Panels.

The Task Force on Rare Neurological Diseases (WG-NeuRare) will be formed by members from all the different Panels (the Chairmen (ex officio), another member and a delegate from the Patient Associations), open also to Neurologists in Training.

This could be an interesting action of the EAN Board, either from the political and ethical point of vue (orphans diseases and orphan drugs) or from a practical point of vue, giving to our members facilities to be informed on this topics and stimulating interactions for the different groups in Europe involved into research.

The aims of the Task Force will be:

- Stimulation the redaction of a list of Rare Neurological Diseases, with main symptoms and diagnostic criteria and guidelines for diagnosis
- Evaluation of the facilities for diagnosis of Rare Neurologic Diseases (RND) in Europe (a list of facilities and address), with the indication where are the main centers interested in the different disorders, where is possible to do the genetic, biochemical and other laboratory tests, etc
- Promotion of an analysis of the attitude of European Neurologist to RND and which is the state of the art of this issue in the different European Countries;
- Stimulation to promotion of registries for RND, data bank and biobanks. These are main aims of the EU, with Research projects in the Biomed Program.
- Stimulation to create European Networks for RND for diagnosis and research.
- Promotion of Teaching courses in Europe.
- Information Service for Rare Neurological Diseases, within the EAN, that will be able, with the collaboration of the different experts present in the WG, to answer to questions from patients, families and doctors (on line). Information service on new data, new findings, research founds, treatments, etc. Discussion on Rare Cases, within the Section on Web page where cases will be described and experts from SSP will answer.

With this activity, the EAN recognizes the primary role of neurologists in the care of these disorders, the necessity to improve the level of the organization of the Neurological Units in Europe and of the formation of neurologists in the care of rare neurological disorders. But also we will stimulate a better integrated relationship with Patient Associations.

UPDATE ON TREATMENT OF NEUROMETABOLIC GENETIC DISEASES

ANTONIO FEDERICO
Dept Medicine, Surgery and Neurosciences, Medical School, University of Siena, Italy

In the recent years numerous new developments in the treatment options to neurometabolic genetic diseases have been obtained. We will report on the most important data, defining symptomatic treatments and therapies able to influence the pathogenetic mechanisms of the disorders, the latter summarized in the following table.

A) Decrease of levels of toxic metabolites
- diet

B) Removal of toxic substrates
- Transfusions, plasmapheresis, peritoneal dyalisis
- Drugs

C) Substitution of deficient substance
- Leucocyte and plasma infusions
- Organs Transplantations
- Fibroblasts tranplantations
- Bone marrow transplantation

D) Direct supply of deficient metabolite

E) Enzymatic induction by coenzymes

F) Enzyme therapy

G) Gene therapy

We will report our experience in this field in several pathological conditions related to lysosomal, mitochondrial , peroxysomal or to metal disturbancies, also discussing some ethical issues related to early presymptomatic treatments.

UPDATE OF INHERITED SMALL VESSEL DISEASES

ANTONIO FEDERICO
Dept Medicine, Surgery and Neurosciences, Medical School, University of Siena, Italy

Cerebral microangiopathies are responsible of a great number of strokes. In the recent years advances in molecular genetics identified several monogenic conditions involving cerebral small vessels and predisposing to ischemic and/or hemorrhagic stroke and diffuse white matter disease leading to vascular dementia. Clinical features and diagnostic clues of these conditions, [cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL), cerebral autosomal recessive arteriopathy with subcortical infarcts and leukoencephalopathy (CARASIL), COL4A1-related cerebral small vessel diseases, autosomal dominant retinal vasculopathy with cerebral leukodystrophy (AD-RVLC), and Fabry's disease] are here reviewed. Albeit with variable phenotypes and with different defective genes, all these disorders produce arteriopathy and microvascular disintegration with changes in brain functions. Specific diagnostic tools are recommended, genetic analysis being the gold standard for the diagnosis. We will also discuss on some pathogenetic mechanism responsible for brain abnormalities evident in an early stage of the diaseses

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