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UNIVERSITY OF
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"IULIU HATIEGANU" UNIVERSITY
OF MEDICINE AND PHARMACY

DOCTORAL SCHOOL NEUROSCIENCE

PROGRAM

2017-2018 | SECTION 6

22 FEBRUARY, 2018
"MULTIMEDIA" AUDITORIUM, "IULIU HATIEGANU" UMF CLUJ-NAPOCA
8 VICTOR BABES STREET | CLUJ-NAPOCA | ROMANIA

PhD NEUROSCIENCE PROGRAM COORDINATOR

Dafin F. Mureşanu

Co-Chair EAN Scientific Panel Neurorehabilitation

President of the European Federation of NeuroRehabilitation Societies (EFNR)

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

INTERNATIONAL GUEST LECTURERS

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Chair of the EAN Scientific Committee

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

Chair of the EAN Rare Neurological Diseases Task Force

Francesca Federico

Aggregate Professor and Researcher M-psi/04 Developmental Psychology

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COURSE PROGRAM

COURSE PROGRAM

FEBRUARY 22ND, 2018

"MULTIMEDIA" AUDITORIUM, "IULIU HATIEGANU" UMF CLUJ-NAPOCA

8 VICTOR BABES STREET | CLUJ-NAPOCA | ROMANIA

09:50 – 10:00	Welcome Address
10:00 – 10:40	Antonio Federico / Italy Genetic leucodystrophies as a model of oligodendrocyte dysfunction
10:40 – 11:20	Antonio Federico / Italy What it is new on neurometabolic extrapyramidal diseases?
11:20 – 11:50	Coffee Break
11:50 – 12:30	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
12:30 – 13:10	Antonio Federico / Italy Update on treatment of neurometabolic genetic diseases
13:10 – 14:40	Lunch Break
14:40 – 15:20	Antonio Federico / Italy Update of Inherited small vessel diseases
15:20 – 16:00	Francesca Federico / Italy Learning disabilities: psychological and physiological basis
16:00 – 16:30	Coffee Break
16:30 – 17:10	Francesca Federico / Italy Learning disabilities: which treatment is helpful?
17:10 – 17:50	Francesca Federico / Italy Dyslexia: how to improve reading abilities without reading

INTERNATIONAL GUEST LECTURERS

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.

FRANCESCA FEDERICO
ITALY

WORKING EXPERIENCE

from december 30th 2008 to date - Confirmed researcher on developmental psychology	Department of Social and Developmental Psychology, Sapienza, University of Rome
2014-to date	Collaboration with clinical center for Learning disabilities and intellectual disabilities, Department of Social and Developmental Psychology, Sapienza, University of Rome
2014- to date	Lecturer of the course of "Cognitive Developmental Psychology" (6 credits) at the under graduate degree of Educational Science, Sapienza University of Rome.
2014 to date	Lecturer of the course "Developmental Psychology" at the undergraduate degree in Healthcare Assistance Science, Centro studi SanGiovanni di Dio (1 credit), Sapienza, University of Rome
2014 to date	Lecturer of the course "Developmental Psychology" at the undergraduate degree in Healthcare Assistance Science, ASL RM B (1 credit), Sapienza, University of Rome
2009 -2013	Lecturer of the course "Developmental Psychology" at the undergraduate degree in Social Work (Class) (4 credits), Sapienza University of Rome
2008-2010	Trainer for teachers of communal nests in the town of Rome
2008-2011	Lecturer of Laboratory of communication at the undergraduate degree of Psychological Intervention during Development and at socio-educational Institution, Sapienza, University of Rome
2008-2011	lecturer of Media and Communication Psychology at the Science and Technique of Developmental Psychology, Sapienza, University of Rome
2007-2008	Lecturer of psychology of Communication at the under graduate degree of Psychological Models during Development and aging, Sapienza, University of Rome
AA 2005-2006	Advisor of Developmetal Psychology at Sat2000 channel on the issues: "brain development during adolescence", Theory of mind development", memory development.
AA 2002-2004	Lecturer of Phisiological Psychology, Sapienza, University of Rome.
	Lecturer of general Psychology, Sapienza, University of Rome

ISTRUZIONE E FORMAZIONE

2009, Octobre 16	PhD thesis "Role of cholinergic system on Spatial Memory: an annimal model"
AA. 2004-2008	European PhD on Applied neurological Science, University of Siena
2003-2006	Clinical Training on Bioenergetic Analysis, SIAB
January 2005- March 2005	Visiting student al FMRIB John Ratchliff Hospital di Oxford
2004 December 10	PhD Thesis "Spatial procedural memory: analysis in different experimental models.
AA. 2001-2004	PhD in Psychobiology and Psychopharmacology, Sapienza, University of Rome
Agosto 2001	Visiting student at Comlumbia University, Presbiterian Medical Centre, New York
A.A. 1995-2001	BA and MA in General and Experimental Psychology , (Mark: 110/100; summa cum laude) Faculty of Psychology, Sapienza, University of Rome.

INTERNATIONAL PUBBLICATIONS

•Federico F, Marotta A, Martella D, Casagrande M (2016). Development in Cognitive Control of Social Processing: evidence from the Attention Network Test. British Journal of Developmental Psychology (in press) Br J Dev Psychol. 2017 Jun;35(2):169-185. doi: 10.1111/bjdp.12154. Epub 2016 Aug 4.

•Laghi F, Federico F, Lonigro A, Levanto S, Ferraro M, Baumgartner E, Baiocco R. (2015). Peer and Teacher-Selected Peer Buddies for Adolescents With Autism Spectrum Disorders: The Role of Social, Emotional, and Mentalizing Abilities. J Psychol. 2016;150(4):469-84. doi: 10.1080/00223980.2015.1087375. Epub 2015 Sep 23

•Foti F, Menghini D, Mandolesi L, Federico F, Vicari S, Petrosini L. Learning by observation: insights from Williams syndrome. PLoS One. 2013;8(1)

•Federico F, Delogu F, Raffone A.Maintenance and manipulation of object sequences in working memory: a lifespan study. Neurol Sci. 2014 Dec;35(12):1883-7. doi: 10.1007/s10072-014-1851-0. Epub 2014 Jun 14.

•Foti F, Mazzone L, Menghini D, De Peppo L, Federico F, Postorino V, Baumgartner E, Valeri G, Petrosini L, Vicari S. Learning by observation in children with autism spectrum disorder. Psychol Med. 2014 Aug;44(11):2437-47. doi: 10.1017/S003329171300322X. Epub 2014 Jan 17..

•Federico F, Marotta A, Adriani T, Maccari L, Casagrande M. Attention network test--the impact of social information on executive control, alerting and orienting. Acta Psychol (Amst). 2013 May;143(1):65-70.

•Foti F, Menghini D, Mandolesi L, Federico F, Vicari S, Petrosini L. Learning by observation: insights from Williams syndrome. PLoS One. 2013;8(1):e53782. doi: 10.1371/journal.pone.0053782. Epub 2013 Jan 10

•Menghini D, Di Paola M, Federico F, Vicari S, Petrosini L, Caltagirone C, Bozzali M. Relationship between gray matter brain anormalities. Behav Genet. 2011 May;41(3):394-402. doi: 10.1007/s10519-010-9419-0. Epub 2010 Dec 10

• Cutuli D, Foti F, Mandolesi L, De Bartolo P, Gelfo F, Federico F, Petrosini L.Cognitive performances of cholinergically depleted rats following chronic donepezil administration.J Alzheimers Dis. 2009 May;17(1):161-76.

• De Bartolo P, Mandolesi L, Federico F, Foti F, Cutuli D, Gelfo F, Petrosini L.Cerebellar involvement in cognitive flexibility. Neurobiol Learn Mem. 2009 Oct;92(3):310-7.

• Mandolesi L., De Bartolo P., Foti F., Gelfo F., Federico F.,Leggio M. G. and Petrosini L. (2008), Environmental enrichment provides a cognitive reserve to be spent in the case of brain lesion. J Alzheimers Dis. 2008 Sep;15(1):11-28.

•Cutuli D, Foti F, Mandolesi L, Bartolo PD, Gelfo F, Federico F, Petrosini L.(2008),Cognitive performance of healthy young rats following chronic donepezil administration. Psychopharmacology (Berl). 2008 May;197(4):661-73. doi: 10.1007/s00213-008-1084-0. Epub 2008 Feb 29

• Mandolesi L, Leggio MG, Spirito F, Federico F, Petrosini L. (2007) Is the cerebellum involved in the visuo-locomotor associative learning?Behav Brain Res. Nov 22;184(1):47-56.

•D. Menghini, F. Federico, M. di Paola, M. Bozzali,L. Petrosini C. Caltagirone & S.Vicari (2007). Relationship between gray matter brain anormalities. Journal of Intellectual Disability Research 51 (9) , 656

• Matteis M., Caltagirone C., Troisi E., Federico F., Silvestrini M., Petrosini L. (2006) , Changes of cerebral blood flow velocity during gestures with or without meaning. A functional transcranial doppler study. Eur J Neurol. Jan;13(1):24-9.

• Federico F, Leggio MG, Mandolesi L, Petrosini L. (2006) The NMDA receptor antagonist CGS 19755 disrupts recovery following cerebellar lesions. Restor Neurol Neurosci.;24(1):1-7.

• Leggio M. G., Federico F., Neri P., Graziano A., Mandolesi L. and Petrosini L. (2006) NMDA receptor activity in learning spatial procedural strategies: I. The influence of hippocampal lesions. Brain Res Bull. Oct 16;70(4-6):347-55.

• Federico F., Leggio M. G., Neri P., Mandolesi L. and Petrosini L. (2006) NMDA receptor activity in learning spatial procedural strategies. II. the influence of cerebellar lesions .Brain Res Bull. Oct 16;70(4-6):356-67.

NATIONAL PUBBLICATIONS

• Leggio MG, Mandolesi L, Federico F, Gelfo F, Petrosini L (2005) Environmental enrichment promotes improved spatial abilities and enhanced dendritic growth in the rat. Behav Brain Res. May 20.

• E.Cannoni, A. Di Norcia, F. Federico (2014)The bicycle drawing test: mechanical reasoning and/or visual-spatial abilities? RASSEGNA DI PSICOLOGIA, n. 3, vol. XXXI,

• F. Laghi, R. Baiocco, A. Perduto, F. Federico, M. D'Alessio (2011). Modelli identificativi proposti in tv e biange eating disorder in adolescenza. PSICHIATRIA DELL'INFANZIA E DELL'ADOLESCENZA, vol. 78, p. 433-448,

• F. FEDERICO, RAFFONE A, DALESSIO M (2010). Sviluppo delle funzioni di "mantenimento" e di "manipolazione" nella memoria di lavoro.. PSICHIATRIA DELL'INFANZIA E DELL'ADOLESCENZA, vol. 77, p. 48-77,

• Federico F. (2008). Effetti del consumo di alcolici sul sistema nervoso degli adolescenti. Giornale di Neuropsicofarmacologia, anno XXX, N1, pg 9-18.

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DAFIN F. MUREȘANU
ROMANIA

Professor of Neurology, Senior Neurologist, Chairman of the Neurosciences Department, Faculty of Medicine, University of Medicine and Pharmacy "Iuliu Hatieganu" Cluj-Napoca, Past President of the Romanian Society of Neurology, President of the Society for the Study of Neuroprotection and Neuroplasticity (SSNN), member of the Academy of Medical Sciences, Romania, secretary of its Cluj Branch. He is member of 16 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 a specialist in Leadership and Management of Research and Health Care Systems (specialization in Management and Leadership, Arthur Anderson Institute, Illinois, USA, 1998 and several international courses and training stages in Neurology, research, management and leadership). 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 400 scientific participations as "invited speaker" in national and international scientific events, a significant portfolio of scientific articles (157 papers indexed on Web of Science-ISI, H-index: 17) as well as contributions in monographs and books published by prestigious international publishing houses. Prof. Dr. Dafin F. Muresanu has been honoured with: the University of Medicine and Pharmacy "Iuliu Hatieganu" Cluj-Napoca, Faculty of Medicine, "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, University of Medicine and Pharmacy "Iuliu Hatieganu" 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.

ABSTRACTS

GENETIC LEUCODYSTROPHIES AS A MODEL OF OLIGODENDROCYTE DYSFUNCTION

ANTONIO FEDERICO
Dept Medicine, Surgery and Neurosciences, Medical School, University of Siena, Italy.
Chair Scientific Committee EAN and Chair of EAN TF Rare Neurologic Diseases

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.
Chair Scientific Committee EAN and Chair of EAN TF Rare Neurologic Diseases

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.
Chair Scientific Committee EAN and Chair of EAN TF Rare Neurologic Diseases

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.
Chair Scientific Committee EAN and Chair of EAN TF Rare Neurologic Diseases

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
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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

LEARNING DISABILITIES: PSYCHOLOGICAL AND PHYSIOLOGICAL BASIS / LEARNING DISABILITIES: WHICH TREATMENT IS HELPFUL? / DYSLEXIA: HOW TO IMPROVE READING ABILITIES WITHOUT READING

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A learning disability is any physical or mental condition that interferes with learning. Learning disabilities can affect spoken or written language, coordination, self-control and attention. Although learning disabilities occur in very young children, disorders are usually not recognized until a child reaches school age. Learning disabilities affect the ability to interpret what one sees and hears or to link information from different parts of the brain. Such difficulties extend to schoolwork and can impede learning to read or write or to do math. Learning disabilities do not reflect IQ (intelligence quotient), or how smart a person is. Consistent difficulties in the achievement of learning have as a consequence different emotional correlates. The most important one is that learning disability affects self-esteem. Emotional intelligence (EI) is the ability to assess, regulate, and utilize emotions and has been found to be associated with academic self-efficacy and a variety of better outcomes, including academic performance. Students with learning disabilities (LD) are well acquainted with academic difficulty and maladaptive academic behavior. In comparison to students without LD, they exhibit high levels of learned helplessness, including diminished persistence, lower academic expectations, and negative affect (Hen & Goroshit, 2014). Students with LD showed lower levels of achievement, effort investment, academic self-efficacy, sense of coherence, positive mood, and hope, and higher levels of loneliness and negative mood (Lackaye & Margalit 2006). In order to trait learning disability is important to rehabilitate cognitive skills impaired by the disability as working memory (Maehler & Schuchardt, 2016), attention , executive functions (Titz & Karbach,2014) and also very important is to work on emotional side and to potentiate self-esteem and self-efficacy.

Between learning disabilities Dyslexia is the most studied and known disorder. Children affected by dyslexia have trouble in reading accurately and fluently. They may also have trouble with reading comprehension, spelling and writing. It is widely recognized, although not universally (see Shaywitz et al., 1992), that dyslexia is more frequent in males (from 2:3 to 4:5, depending on the study), with significant familial occurrence. Children with this condition often have associated deficits in related domains such as oral language acquisition (dysphasia), writing abilities (dysgraphia and misspelling), mathematical abilities (dyscalculia), motor coordination (dyspraxia), postural stability and dexterity, temporal orientation (`dyschronia'), visuospatial abilities (developmental right-hemisphere syndrome), and attentional abilities (hyperactivity and attention deficit disorder) (Weintraub and Mesulam, 1983; Rapin and Allen, 1988; Dewey, 1995; Gross-Tsur et al., 1995, 1996; Fawcett et al., 1996). Evidence for a genetic origin of dyslexia has been increasingly accumulating during the last few years and will not be reviewed here. The reader is referred to more specific writings by Pennington (Pennington, 1991, 1997, 1999), Schulte-Körne and colleagues (Schulte-Körne et al., 1996), Smith and colleagues (Smith et al., 1998), and Flint (Flint, 1999), and to recent discoveries regarding the involvement of specific chromosomes (Fagerheim et al., 1999; Fisher et al., 1999; Gayan et al., 1999). However, genetic transmission is probably complex and non-exclusive. Reading involves multiple cognitive processes, two of which have been of particular interest to researchers: 1) grapheme-phoneme mapping in which combinations of letters (graphemes) are mapped onto their corresponding sounds (phonemes) and the words are thus "decoded," and 2) visual word form recognition for mapping of familiar words onto their mental representations. Together, these processes allow us to pronounce words and gain access to meaning. In accordance with these cognitive processes, studies in adults and children have demonstrated that reading is supported by a network of regions in the left hemisphere (Price, 2012), including the occipito-temporal, temporo-parietal, and inferior frontal cortices. The occipito-temporal cortex holds the "visual word form area." Both the temporo-parietal and inferior frontal cortices play a role in phonological and semantic processing of words, with inferior frontal cortex also involved in the formation of speech sounds. These areas have been shown to change with age (Turkeltaub, et al., 2003) and are altered in people with dyslexia (Richlan et al., 2011). Evidence of a connection between dyslexia and the structure of the brain was first discovered by examining the anatomy of brains of deceased adults who had dyslexia during their lifetimes. The left-greater-than-right asymmetry typically seen in the left hemisphere temporal lobe (planum temporale) was not found in these brains (Galaburda & Kemper, 1979), and ectopias (a displacement of brain tissue to the surface of the brain) were noted (Galaburda, et al., 1985). Then investigators began to use MRI to search for structural images in the brains of research volunteers with and without dyslexia. Current imaging techniques have revealed less gray and white matter volume and altered white matter integrity in left hemisphere occipito-temporal and temporo-parietal areas (Bailey et al., 2016).

Recently the treatment of dyslexia is focused on rehabilitation of different skills that don't imply directly reading skills; Franceschini et al. (2013) demonstrate that only 12 hr of playing action video games-not involving any direct phonological or orthographic training-drastically improve the reading abilities of children with dyslexia. Habib et al., (2016) developed a series of musical exercises involving jointly and simultaneously sensory (visual, auditory, somatosensory) and motor systems, with special emphasis on rhythmic perception and production in addition to intensive training of various features of the musical auditory signal. Dyslexic children showed significant improvement in categorical perception and auditory perception of temporal components of speech and also they showed additional improvements in auditory attention, phonological awareness (syllable fusion), reading abilities, and repetition of pseudo-words.

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