E A N

TASK FORCE FOR RARE NEUROLOGIC DISEASES

2ND TEACHING COURSE

5 - 7 SEPTEMBER 2018

GRAND HOTEL ITALIA | CLUJ -NAPOCA | ROMANIA

PROGRAM COORDINATORS

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

Chairmen of EAN Task Force for Rare Neurologic Diseases

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President European Federation of NeuroRehabilitation Societies (EFNR)

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FACULTY

IN ALPHABETICAL ORDER

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Ovidiu Băjenaru / Romania

Peter Van den Bergh / Belgium

Jean-Marc Burgunder / Switzerland

Antonio Federico / Italy

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Holm Graessner / Germany

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Ramona Moldovan / Romania

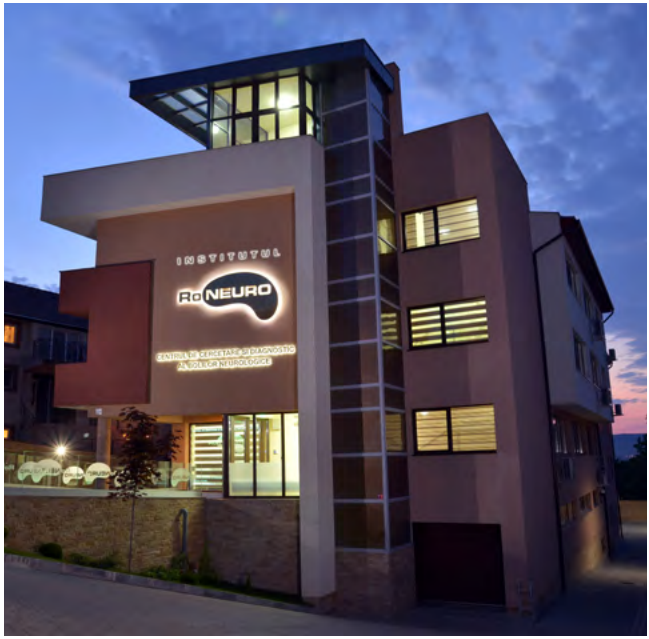
Maria Judit Molnár / Hungary

Dafin F. Mureșanu / Romania

Davide Pareyson / Italy

Antonio Toscano / Italy

GENERAL INFORMATION



GENERAL INFORMATION

REGISTRATION DESK

All materials and documentation will be available at the registration desk located at SSNN booth.
The staff will be pleased to help you with all enquiries regarding registration, materials and program. Please do not hesitate to contact the staff members if there is something they can do to make your stay more enjoyable.

LOGISTIC PARTNER:

SYNAPSE
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LANGUAGE

The official language is English. Simultaneous translation will not be provided.

CHANGES IN PROGRAM

The organizers cannot assume liability for any changes in the program due to external or unforeseen circumstances.

NAME BADGES

Participants are kindly requested to wear their name badge at all times. The badge enables admission to the scientific sessions and dinners.

FINAL PROGRAM & ABSTRACT BOOK

The participants documents include the program and abstract book which will be handed out at the registration counter.

COFFEE BREAKS

Coffee, tea and water are served during morning coffee breaks and are free of charge to all registered participants.

MOBILE PHONES

Participants are kindly requested to keep their mobile phones turned off while attending the scientific sessions in the meeting rooms.

CURRENCY

The official currency in Romania is RON.

ELECTRICITY

Electrical power is 220 volts, 50 Hz. Two-prong plugs are standard.

TIME

The time in Romania is Eastern European Time (GMT+2).

ACADEMIC PARTNERS

European Academy
of Neurology
www.ean.org

Foundation for the
Study of Nanoneurosciences and
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European
Reference
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(ERN-RND)

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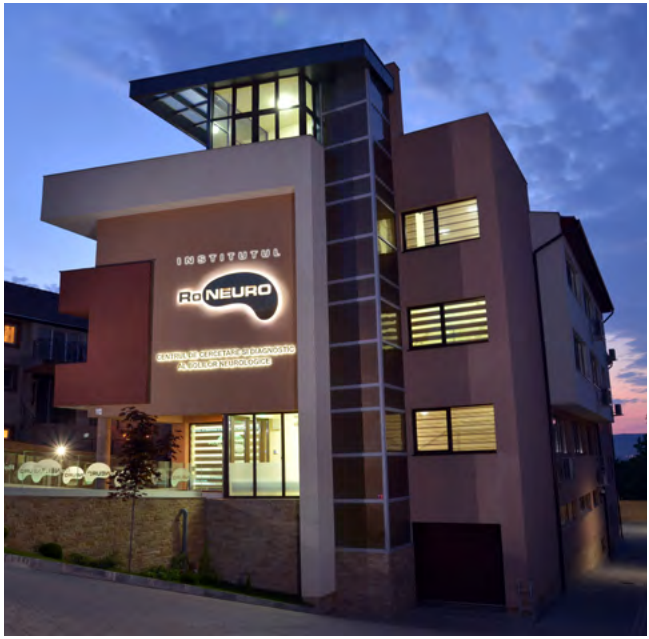
SCIENTIFIC PROGRAM



WEDNESDAY 5 SEPTEMBER, 2018

09:00	Opening Registration
10:00 – 10:30	***WELCOME BY HOSTING AUTHORITIES AND CHAIR***

SESSION 1

Chairpersons:	Dafin F. Mureșanu (Romania), Peter van den Bergh (Belgium), Maria Judit Molnár (Hungary)
10:30 - 10:45	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
10:45 - 11:15	Holm Graessner (Germany) RND-ERN
11:15 - 11:45	Reetta K. Kälviäinen (Finland) Epicare-ERN
11:45 - 12:30	Maria Judit Molnár (Hungary) Rare neurologic diseases and the web: how we can improve the diagnosis
12:30 – 13:00	Jean Marc Burgunder (Switzerland) Perspectives on gene targeting therapies in rare inherited neurologic diseases
13:00 - 14:30	***LUNCH***

WEDNESDAY 5 SEPTEMBER, 2018

SESSION 2

Chairpersons: Davide Pareyson (Italy),
Ovidiu Băjenaru (Romania)

14:30 - 15:30	Davide Pareyson (Italy) Inherited peripheral neuropathies
15:30 – 16:15	Peter Van den Bergh (Belgium) Acquired peripheral neuropathies
16:15 - 16:30	***COFFEE BREAK***
16:30 – 17:00	Ovidiu Băjenaru (Romania) Gaucher disease and Parkinson's disease
17:00 - 19:00	Case discussions on peripheral neuropathies, and epilepsy
20:00	***DINNER***

THURSDAY 6 SEPTEMBER, 2018

SESSION 3

Chairpersons: Holm Graessner (Germany),
Jean Marc Burgunder (Switzerland)

09:00 – 09:45	Antonio Toscano (Italy) HyperCKemia
09:45 - 10:30	Antonio Toscano (Italy) What we can learn by muscle biopsy
10:30 - 11:15	Dafin F. Mureșanu (Romania) Rare neurologic disorders in the context of rare causes of stroke
11:15 - 11:45	**COFFEE BREAK**
11:45 - 12:30	Alessandro Filla (Italy) Dominant ataxias
12:30 - 13:15	Alessandro Filla (Italy) Recessive ataxias
13:15 - 15:00	**LUNCH**
15:00 - 15:45	Jean Marc Burgunder (Switzerland) Hereditary spastic paraparesis
15:45 - 16:30	Jean Marc Burgunder (Switzerland) Huntington's disease and other rare forms of chorea
16:30 - 17:30	Cases discussion on muscle, vascular diseases, ataxia.
20:00	**DINNER**

FRIDAY 7 SEPTEMBER, 2018

SESSION 4

Chairpersons: Alessandro Filla (Italy), Alberto Albanese (Italy)

09:00 – 09:45	Alberto Albanese (Italy) Distonias
09:45 – 10:30	Antonio Federico (Italy) Syndromes of mineral accumulation into the brain: clinical and pathogenetic aspects
10:30 - 11:00	***COFFEE BREAK***
11:00 – 11:30	Antonio Federico (Italy) Genetic leucodystrophies as a model of oligodendrocyte dysfunction
11:30 - 12:00	Ramona Moldovan (Romania) Journey from theory to practice: setting up a Huntington disease service in Romania
12:00 – 13:30	***LUNCH***

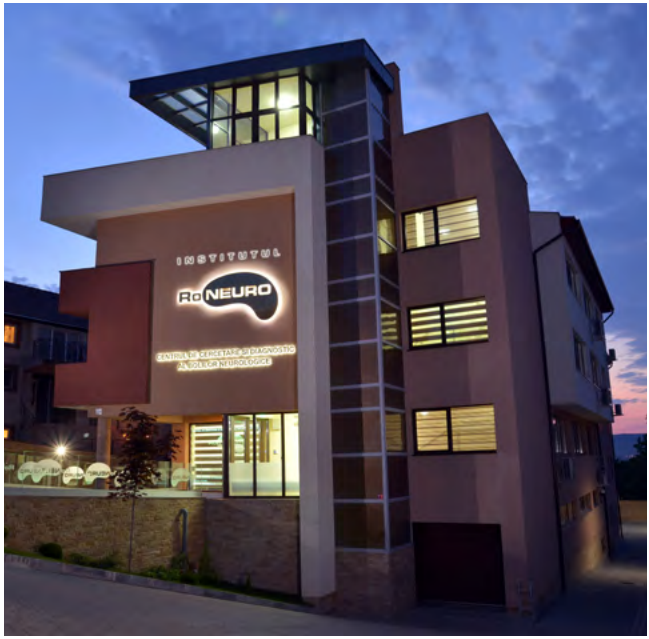
FRIDAY 7 SEPTEMBER, 2018

SESSION 5

Chairpersons: Antonio Federico (Italy), Antonio Toscano (Italy)

13:30 – 14:00	Holm Graessner (Germany) Perspectives for under-diagnosed patients
14:00 – 14:30	Antonio Toscano (Italy) Treatments for muscle glycogenoses
14:30 - 15:00	Antonio Federico (Italy) Update on treatment of neurometabolic genetic diseases
15:00 – 15:30	Antonio Federico (Italy) Treatments of mitochondrial diseases
15:30 – 16:00	***COFFEE BREAK***
16:00 – 17:30	Case discussions on leucoencephalopathies, dystonias, Fabry, etc
17:30	***CONCLUDING REMARKS***
19:00	***SPECIAL EVENT AT THE ROMANIA NATIONAL OPERA CLUJ NAPOCA***

ABSTRACTS



ACQUIRED PERIPHERAL NEUROPATHIES

PETER VAN DEN BERGH
Full Professor of Neurology at the Université catholique de Louvain (UCL)
Director of the Neuromuscular Reference Centre of the University Hospital Saint-Luc, Brussels, Belgium

Electrophysiology plays a crucial role in the characterization and diagnosis of peripheral neuropathies. It provides insight in the type and mechanism of peripheral neuropathy by giving information on the spatial pattern (generalized, multifocal, focal), the fibre type involved (motor, sensory), pathology (axonal, demyelinating), and the severity and time course (acute, ongoing, chronic). Electrophysiological studies are key in the early detection and characterization of inflammatory demyelinating neuropathies and in differentiating these from primary axonal neuropathies.

Inflammatory demyelinating neuropathies constitute a significant proportion of the acquired peripheral neurophaties. They include Guillain-Barré syndrome (GBS) chronic inflammatory demyelinating polyradiculoneuropathy (CIDP), multifocal motor neuropathy (MMN), multifocal demyelinating neuropathy with persistent conduction block (Lewis-Sumner syndrome), and paraproteinemic neuropathies. A proper diagnosis as early as possible is very important because timely immune treatment can largely reduce morbidity and disability. The diagnosis is based on a constellation of clinical and laboratory features, including electrophysiological studies, spinal fluid examination, and in selected cases serological studies and peripheral nerve biopsy.

In CIDP, electrophysiological criteria for demyelination are designed to exclude abnormalities that can be explained by axonal degeneration (Eur J Neurol 2010). Therefore, lesser degrees of demyelination cannot be defined with certainty. Optimised electrophysiological criteria are capable, however, to support the diagnosis with different levels of probability (possible, probable, definite) in the very large majority of cases. In GBS, much effort has gone into developing criteria which can distinguish axonal and demyelinating subtypes. The discovery of reversible conduction failure (RCF) has led to the concept of nodopathy/paranodopathy, where conduction slowing and conduction block are due to the immune attack mainly at the nodal axolemma level. There is no actual demyelinisation as defined pathologically and if the immune attack continues, conduction failure may not reverse and axonal degeneration will ensue. Recent electrophysiological studies support this pathophysiological mechanism and show that the dichotomous distinction between axonal and demyelinating in GBS is not tenable (Muscle nerve, 2018).

PERSPECTIVES ON GENE TARGETING THERAPIES IN RARE INHERITED NEUROLOGIC DISEASES

JEAN-MARC BURGUNDER
Professor of Experimental Neurology at the Faculty of Medicine of the University in Bern, Switzerland

Rare inherited neurologic disorders typically have a variable phenotype including movement disorders, cognitive decline, behavioural changes and other symptoms from central nervous system involvement in various intensities, often together with peripheral, muscular and systemic impairment. This is due to complex functions of the mutated proteins, which leads to numerous biological changes, and these are addressed by multiple symptomatic therapies. The mutations, as a single cause of the diseases, would suggest a simple target to develop therapeutic strategies. However, the proteins are typically involved in numerous interactions. For example the translation of the elongated CAG repeat leads to a protein with multiple disturbed functions but also to its accumulation. Preclinical studies and clinical trials have addressed these downstream pathways leading to a high level of knowledge about the molecular disease processes. Unfortunately this has not lead to protective effect demonstration with disease course modification in most of the cases. Direct gene repair is being studied in animal models but the strategy is not ready for human use. However targeting gene transcript might also address this complexity, as a single molecule should be modulated. This can be done by synthesized nucleic acids, which bind to mRNA in order to inactivate it. Application of this method in cell and animal models has paved the way for clinical studies in several disorders. Safety of intrathecally administered antisense oligonucleotides targeting mRNA has now been demonstrated. Such an approach has now lead to clinical use in spinal muscular atrophy and found to be safe in Huntington's disease. Major emerging issues including availability and costs of such treatments have to be addressed now. These treatments have the potential to modify the care programs of patients with inherited neurologic disorders, but not to remove their need. Work presently done in management improvement will have to proceed beyond the application of such treatments.

HEREDITARY SPASTIC PARAPARESIS

JEAN-MARC BURGUNDER
Professor of Experimental Neurology at the Faculty of Medicine of the University in Bern, Switzerland

Spasticity, from the Greek word meaning drawing or pulling, is a symptom felt as tightness or stiffness of muscle during movement. It is often accompanied by weakness. At examination increased tone is found at the start of a rapid passive movement. Increased tendon reflexes and positive pyramidal signs usually accompany spastic tone. Spastic paraplegia has a very large differential diagnosis with a long list of acquired and hereditary disorders. Among the latter, the group of hereditary spastic paraparesis includes forms with spasticity as the major feature and forms in which the latter is part of a more complex syndrome. Inheritance may be dominant, recessive, X-chromosomal or maternal. There is a profound overlap between this group of rare disorders and other hereditary neurogenetic disorders including the spinocerebellar ataxia and mitochondrial disorders. Furthermore, spasticity may be present in some rare metabolic disorders, some of which may be treated at the source of the molecular disorder. The diagnosis is finally confirmed by genetic testing in familial disorders, either by direct genetic testing of genes suggested by examination, or by panel or exome sequencing. However, a thorough assessment of the phenotype may readily disclose the actual form, for example cerebrotendinous xanthomatosis, in which a treatment to modify the disturbed metabolic pathway is established. Precise diagnosis is important in order to recognise also other forms, albeit rare, with options for treatment, which would modify the course of the disease. Phenotype assessment will have to be performed on a regular basis in the course of the disease, the frequency may higher in some disorders more rapidly progressing disorders or in order to assess effect of treatment, or very low in those disorders with very slow progression, like hereditary spastic paraplegia due to some spastin mutation. Symptomatic treatment will have to address the present complains and signs present at a particular point of time in the course, and this will change over time. They may include antispastic drugs, but physical therapy plays a very important role to retain function as long as possible. In the future, therapies aimed at an upstream molecular targeting will hopefully be available and the increasing knowledge of the molecular mechanisms of the hereditary spastic paraplegias provides hope in this regard.

HUNTINGTON'S DISEASE AND OTHER RARE FORMS OF CHOREA

JEAN-MARC BURGUNDER
Professor of Experimental Neurology at the Faculty of Medicine of the University in Bern, Switzerland

Huntington's disease is a monogenetic disorder with variable age at onset of motor, cognitive and behavioral symptoms. This is the most frequent hereditary cause of chorea, and is due to a CAG triplet elongation in the huntingtin, one example of a dynamic mutation. The mechanism of dynamic mutations explains the change of onset from one to the next generation, and the age at onset is negatively correlated with the number of CAG repeats. Mutations in other genes may also lead to a syndrome with prominent chorea, including benign hereditary chorea, chorea-acantocytosis, and HD-like syndromes. Furthermore, chorea may be an accompanying feature in other neurogenetic disorders classified in other groups according the most prominent presentation, like in spinocerebellar ataxia or in leucoencephalopathy with axonal inclusions. The diagnosis is finally confirmed by gene testing, however a thorough evaluation of the phenotype is important in order to understand the causes of suffering and impairment in a particular patient. This information, which will have to be reassessed regularly during the course of the disease, which may last for decades, is important for the design of appropriate symptomatic treatment. The discovery of genetic causes of chorea has allowed exploring molecular aspects of the disease mechanisms, and, in some instances, allowing molecular-based treatments. They have also shed light in normal brain functioning. The management of these disease with long duration and complex phenotypes should involve a multidisciplinary team specialized in such disorders. Typically such a team will include neurologists, geneticists, psychiatrists, physiotherapists, ocupational therapists, nurses, who work in a comprehensive way to take care of patients from the time of presymptomatic genetic testing to end-of-life care.

GENETIC LEUCODYSTROPHIES AS A MODEL OF OLIGODENDROCYTE DYSFUNCTION

ANTONIO FEDERICO
Department of Medicine, Surgery and Neurosciences, Medical School, University of Siena, Siena, Italy
Past Chairman of the Scientific Committee and Past Member of the Board of the European Academy of Neurology
Chairmen of EAN Task Force for 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.

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
Department of Medicine, Surgery and Neurosciences, Medical School, University of Siena, Siena, Italy
Past Chairman of the Scientific Committee and Past Member of the Board of the European Academy of Neurology
Chairmen of EAN Task Force for 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.

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

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Past Chairman of the Scientific Committee and Past Member of the Board of the European Academy of Neurology
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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 ON TREATMENT OF NEUROMETABOLIC GENETIC DISEASES

ANTONIO FEDERICO

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

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

Chairmen of EAN Task Force for 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.

TREATMENT OF MITOCHONDRIAL DISEASES

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Mitochondrial diseases are disorders of energy metabolism, interesting all the organs, mainly muscle, eye and brain and characterized by an heterogenous symptomathology.

Treatments are mainly symptomatic, using drugs able to interact with energy metabolism of the cells (mainly vitamins, anti-oxidants, etc).

We will report the main drugs used and other treatments that are able to have a positive effect on mitochondrial metabolism and others drugs currently used to improve symptoms that, having negative effects on energy metabolism of mitochondrial functions, are to be avoided.

We will discuss the current use of dietary supplement and exercise therapies as well as emerging therapies that may be broadly applicable across multiple mitochondrial diseases or for specific disorders.

Examples of non-tailored therapeutic targets include: activation of mitochondrial biogenesis, regulation of mitophagy and mitochondrial dynamics, bypass of biochemical defects, mitochondrial replacement therapy, and hypoxia.

Other tailored-therapies are: scavenging of toxic compounds, deoxynucleoside and deoxynucleotide treatments, cell replacement therapies, gene therapy, shifting mitochondrial DNA mutation heteroplasmy, and stabilization of mutant mitochondrial transfer RNAs.

DOMINANT ATAXIAS

ALESSANDRO FILLA
Full Professor of Neurology and Chairman of the Department of Neurological Sciences at Federico II, Naples, Italy

The dominant ataxias or spinocerebellar ataxias (SCAs) are a large and diverse group. Dominant ataxias were first described by Menzel (1891), Sanger Brown (1892) and Marie (1893). A CAG repeat expansion was found responsible for SCA1 in 1993 followed by other CAG ataxias. The next generation sequencing widened the number of SCAs that amount to 47 with 35 identified genes. The prevalence varies on geography and ethnicity with an average of 2.7 x 10-5 (1.5-4.0 x 10-5). The most common SCAs are caused by expanded CAG repeats that encode polyglutamine (poyQ) in the relevant disease proteins. The polyQ ataxias include SCA1-3, SCA6-7, SCA17 that are caused by expanded polyQ sequences in ataxin-1 (ATX1), ATX2-3, in the subunit α of voltage-gated calcium channel (CACNA1A), and TATA box-binding protein (TBP). Anticipation and intranuclear inclusions characterize most polyQ SCAs. Other SCAs are caused by non-coding repeat expansions (SCA8, SCA10, SCA31, SCA36). Still other SCAs result from point mutations (SCA5, SCA11, SCA13, SCA14, SCA15/16, SCA23, SC26, SCA27, SCA28, SCA34, SCA35, SCA38, SCA40, SCA41, SCA42, SCA43). Some peculiar features may point to the diagnosis (slow saccade to SCA2, pyramidal signs to SCA1 and SCA3 and SCA7, pure cerebellar SCA6, parkinsonism SCA2-3, SCA6, SCA17) dyskinesia (SCA3 and SCA17) and retinal degeneration (SCA17). Disease severity tends to be greater and survival shorter for patients with polyQ SCAs than with a non-polyQ SCA.

Lowering the levels of the toxic protein is currently the most compelling strategy towards developing a disease-modifying therapy for polyQ ataxias. New therapeutic approaches as antisense oligonucleotides or miRNA are on the reach.

RECESSIVE ATAXIAS

ALESSANDRO FILLA
Full Professor of Neurology and Chairman of the Department of Neurological Sciences at Federico II, Naples, Italy

The recessive cerebellar ataxias represent a clinical and genetic heterogeneous group. Next generation sequencing allowed to identify new genes, widened the phenotypes of previously identified disorders, and modified the laboratory testing approach. At the present, a conservative estimate counts about 45 recessive ataxias plus 30 hereditary disorders where ataxia is a prominent feature. The average prevalence is 3.3x10-5 (1.8-4.9x10-5). The presentation concerns a clinical approach that can point to choice of the genetic test and to diagnosis. Recessive ataxias are divided into main groups. Recessive spastic ataxias that include SPG7, ARCA2, Friedreich variant with retained reflexes, ARCA1, ARSACS, cerebrotendinous xanthomatosis (CTX). Recessive ataxia with peripheral neuropathy that include Friedreich ataxia, MIRAS/POLG, AOA1-2, Ataxia telangiectasia (AT), ARSACS. Recessive ataxias without peripheral neuropathy that include ARCA1/SYNE1, ARCA2, ARCA3, SPG7. Recessive ataxias with other additional features that include hypogonadism (Boucher-Neuhauser, Gorgon Holmes, Marineso-Sjogren, Wolfram, Congenital Disorders of Glycosylation (CDG)); parkinsonism (CTX, FXTAS); dyskinesia (Niemann-Pick type C (NPC), AOA, AT); myoclonus (neuronal ceroid lipofuscinosis (NCL), MERRF, sialidoses); dementia/intellectual disability (Kufs disease, GM2 gangliosidosis, CTX, CDG); epilepsy (GLUT-1, MIRAS, ARCA2, NCL, MERRF, CTX, NPC). The diagnostic flowchart suggests accurate definition of the phenotype including MRI and peripheral nerve study; screen for acquired causes of ataxia; 1st step genetic testing for GAA and single gene by Sanger; 2nd step genetic testing by clinical target/exome sequencing.

PERSPECTIVES FOR UNDER-DIAGNOSED PATIENTS & RND-ERN

HOLM GRAESSNER
Managing Director, Rare Disease Centre, University Hospital Tübingen, Germany

The two talks I will be giving will both focus on diagnosis. Using rare movement disorders patients as examples I will explain how the European Reference Network for Rare Neurological Diseases (ERN-RND) and the H2020 project Solve-RD. Solving the unsolved Rare Diseases. contribute finding a diagnosis for a patient with a rare neurological disease.

ERN-RND (ern-rnd.eu) is a network of 32 Healthcare Providers from 13 EU member states. ERN-RDN builds on existing expert centres and mature networks dedicated to rare neurological diseases (RND) as well as established rare disease infrastructures such as Orphanet, EURORDIS and RD-Connect. Through coordination and knowledge transfer, ERN-RND establishes a patient-centred network to address the needs of patients with RND of all age groups, with or without a definite diagnosis, by implementing an infrastructure for diagnosis, evidence-based management, treatment and collection of patient data.

The network is active in two main areas: (i) knowledge generation, transfer and dissemination in order to harmonise quality of diagnosis and treatment provided in the network and beyond; (ii) introduction, piloting and role-out of the Clinical Management System (CPMS), an e-health platform provided to the ERNs by the European Commission, to consult on complex cases applying multidisciplinary expert panels.

Solve-RD (solve-rd.eu) echoes the ambitious goals set out by the International Rare Diseases Research Consortium (IRDiRC) to deliver diagnostic tests for most rare diseases by 2020. Our main ambitions are thus i) to solve large numbers of rare disease, for which a molecular cause is not known yet by sophisticated combined omics approaches, and ii) to improve diagnostics of rare disease patients through contribution to, participation in and implementation of a "genetic knowledge web" which is based on shared knowledge about genes, genomic variants and phenotypes.

Solve-RD fully integrates with the newly formed European Reference Networks (ERNs) for rare diseases which have begun to operate in 2017. Four ERNs (ERN-RND, -EURO-NMD, -ITHACA, and -GENTURIS) build the core of Solve-RD. Solve-RD will deliver seven implementation steps to address these:

i) Collect large amount of RD data , ii) Discover new phenotype patterns, iii) Re-analyse exomes/genomes, iv) Apply novel molecular strategies, v) Facilitate functional analysis, iv) Work towards clinical utility and vii) Towards therapy.

JOURNEY FROM THEORY TO PRACTICE: SETTING UP A HUNTINGTON DISEASE SERVICE IN ROMANIA

RAMONA MOLDOVAN
Department of Psychology, Babeș-Bolyai University, Cluj-Napoca, Romania

In Romania, the diagnosis and care of patients with Huntington's Disease (HD) and support of their families is problematic given the absence of integrated services and multidisciplinary teams adequately trained to approach HD. Thus, there is a fragmentation of all actions that are necessary for HD families in terms of diagnosis, treatment and support. At the moment, there is no HD center in Romania and there is no standard practice for HD patients. Setting up a regional HD center in Cluj that would start by serving Transylvania has been our objective for a couple of years and our efforts so far have started to show results. The aim of the current paper is to discuss our progress in developing the first HD multidisciplinary clinic in Romania.

RARE NEUROLOGIC DISEASES AND THE WEB: HOW WE CAN IMPROVE THE DIAGNOSIS

MARIA JUDIT MOLNAR
Institute of Genomic Medicine and Rare Disorders
Semmelweis University, Budapest, Hungary

The importance of the Internet as a medium sharing health and medical information has increased considerably during the last decade. The distribution of information about rare diseases is an important factor to improve the overall situation of people affected by a rare disease, so the Internet as a worldwide open-access medium has become more important during the last decade. The Internet can improve the dissemination of information about rare diseases to the general public and, in particular, to medical professionals, patients, and relatives of patients. For the latter group it is one of the most frequently used information resources and often the primary source to search for information after getting a diagnosis. Patients reported that they are often overstrained by information, it is not possible to assess the quality of the information, to find the right information, such as social-legal advice. For medical professionals, it is important to have access to the latest innovative research results and evidence-based therapeutic options and to use web based tools for diagnostic purposes. The presentation will give an overview how the Internet is empowering the rare disease community, how internet-driven patient-finding is working, how web based applications can support you in the

identification of your patient's diagnosis and to find the optimal treatment options and how the improved web-based patient engagement can support your diagnostic work up.

RARE NEUROLOGIC DISORDERS IN THE CONTEXT OF RARE CAUSES OF STROKE

DAFIN F. MUREȘANU
Chairman Department of Clinical Neurosciences
'Iuliu Hatieganu' University of Medicine and Pharmacy, Cluj-Napoca, Romania

According to the World Health Organization, 15 million people suffer stroke worldwide each year. Of these, 5 million die and another 5 million are permanently disabled. Europe averages approximately 650,000 stroke deaths each year.

Stroke is the number one cause of permanent disability globally and the second most common cause of dementia. Although stroke among young adults is generally considered a rare event, with a previous study reporting that about 5% of all strokes in the United States occurred in a young adult population aged between 18 and 44 years, there is growing evidence of an increasing trend of stroke in young adults. It has been documented that stroke incidence in young adults aged between 20 and 54 years has significantly increased between 1999 and 2005.

Many risk factors for cerebrovascular diseases have been established including no modifiable factors such as age, gender, and race, as well as acquired risk factors such as hypertension, smoking, diabetes, and obesity. These factors, however, only account for a portion of the stroke risk suggesting that other variables, including genetics, must be involved in the etiology of stroke. The exact contribution of genetics to the incidence of stroke still remains largely unknown; however, it is clear that stroke can result from both monogenic and polygenic diseases. Common monogenic causes of stroke include cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) and its autosomal recessive form, CARASIL, as well as sickle cell disease, and Fabry disease.

Among rarer monogenic and polygenic causes of stroke we have: mitochondrial encephalomyopathy, lactic acidosis, and stroke like episodes (MELAS), hereditary endotheliopathy with retinopathy, nephropathy, and stroke (HERNS), homocystinuria, moyamoya disease, and inherited connective tissue disorders, including type IV collagen α1- chain gene (COL4A1) mutation, Marfan syndrome, and vascular Ehlers–Danlos syndrome (VEDS).

Despite all recent advances in neuro-technologies applied for stroke diagnostic, up to a third of strokes are rendered cryptogenic or of undetermined etiology. This number is specifically higher in younger patients. At times, inadequate diagnostic workups, multiple causes, or an under-recognized etiology contributes to this statistic.

The current presentation will give a brief overview related to most studied rare causes of stroke: aortic arch atheroma, cervical dissection, PFO & ASA, hereditary conditions, thrombophilia, acquired hypercoagulable status and vasculitis.

INHERITED PERIPHERAL NEUROPATHIES

DAVIDE PAREYSON
Head of Functional Department on Rare Neurological Diseases
Head of Rare Neurodegenerative and Neurometabolic Diseases Unit
Department of Clinical Neurosciences
IRCCS Foundation, C. Besta Neurological Institute Milan, Italy

All inherited peripheral neuropathies (IPN) are rare diseases. The review will deal mainly with Charcot-Marie-Tooth disease (CMT) and hereditary transthyretin amyloidosis (hATTR) and briefly with other rare IPN.

CMT and related disorders represent a heterogeneous group of hereditary neuropathies. CMT affects both sensory and motor nerves, distal Hereditary Motor Neuropathies (dHMN) are phenotypically similar disorders involving only motor nerves, while Hereditary Sensory and Autonomic Neuropathies (HSAN) are rare distinct disorders affecting sensory and sometimes autonomic nerves and Hereditary Neuropathy with liability to pressure palsies (HNPP) is a recurrent focal neuropathy. Next generation sequencing led to several new gene uncovering in the field and is modifying the diagnostic approach. More than 90 genes have been identified as responsible for these disorders, encoding proteins with very diverse function. Numerous promising compounds are under study in cellular and animal models, including therapies targeting protein degradation pathways, protein overexpression, or directly the specific genetic mutations. Therefore, clinical trials are underway or close to start, with current efforts devoted to developing responsive outcome measures and finding biomarkers for this overall slowly progressive disorder.

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. Early diagnosis is fundamental in this otherwise lethal disorder and is easier in familial cases in endemic regions, where Val30Met is by far the predominant mutation. Diagnosis is often delayed in non-endemic region where other mutations are also found, onset occurs later in life, presentation is often atypical with a sensory-motor polyneuropathy involving all fibre types and progression is definitely faster. Orthotopic liver transplantation was until few years ago the only available therapy. The TTR tetramer stabilizer tafamidis proved able to slow down disease progression particularly in the early disease phases and is currently approved in several countries for treatment of symptomatic patients with stage I polyneuropathy; another tetramer stabilizer, diflunisal, also produced significant slowing in disease progression in treated patients and is currently used in some countries as an off-label treatment. TTR gene silencing approach with Antisense Oligonucleotides (Inotersen) or interfering RNA lipid nanoparticles (Patisiran) proved very effective in both two recently completed phase III trials, and other therapies are under investigation. The development of such novel therapies is changing the natural history of ATTR-neuropathy from a relentlessly progressive disorder inexorably leading to death into an effectively treatable disorder.

HYPERCKEMIA

ANTONIO TOSCANO
Department of Clinical and Experimental Medicine of the University of Messina, Italy

Creatine kinase is a key enzyme that catalyzes the release of high-energy phosphates from creatine phosphate, mainly present in skeletal muscle. In muscle disorders, there is often a gross elevation of CK (hyperCKemia) because of its leak into serum in large amounts. HyperCKemia is present in various types of muscle diseases such as muscular dystrophies, limb-girdle myopathies, congenital myopathies and metabolic myopathies, although in the latter case, serum CK may be within the normal range (50-200 U/l) in the interictal periods.

To establish what are the causes provoking hyperCKemia in patients, it is necessary to follow an algorithm including:

a) a very careful personal and family history which is very relevant to the diagnosis because of possible different ways of inheritance; then physical and neurologic examinations are necessary, especially to establish the specific muscle involvement.
b) routine blood tests (including other specific muscle enzymes as transaminases, LDH, electrolites, etc;
c) exercise tests: to monitor mainly lactate and pyruvate levels either basic or after forearm test (for suspected glycogenoses) and after cycloergometric test (for suspected lipid storage myopathies or mitochondrial myopathies);

a) neurophysiological examinations: electromyography and electroneurography to properly address the differential diagnosis among muscle, nerve, neuromuscular junction or motor neuron disorders;
b) neuroimaging: CT or MRI scans may help in detecting occult muscle or central nervous system abnormalities;
c) muscle biopsy: this is one of the most relevant investigations to study the morphological alterations of skeletal muscle;
d) biochemical studies: to determine a specific alteration of the enzyme activities cascade in the different metabolic pathways;
e) molecular-genetic studies: to analyse patient's DNA (and sometimes his/her relatives) to confirm clinical and biochemical diagnosis or to make a diagnosis based on specific DNA markers.

Having variably taken in consideration those diagnostic factors, we will be able to reach, in the majority of cases, the correct diagnosis, also in order to start an appropriate and timeline therapy.

WHAT WE CAN LEARN BY MUSCLE BIOPSY

ANTONIO TOSCANO
Department of Clinical and Experimental Medicine of the University of Messina, Italy

Muscle biopsy is often requested in order to identify and diagnose various kind of muscular disorders. This biopsy is a minor surgical procedure which needs a small incision to remove a very limited portion of a specific tissue for examination.

It is important to perform the muscle biopsy after a series of other clinical examinations as neurophysiological and laboratory evaluations. Morphological aspects could have a critical role, addressing and providing diagnostic evidence that either establishes a disease etiology or focuses the differential diagnosis.

Muscle biopsy is an important tool for the evaluation and diagnosis of patients presenting with acute or progressive weakness who are suspected of having an underlying neuromuscular disorder.

For example, in case of a rapidly progressive muscle weakness, a muscle biopsy is the most valuable diagnostic tool which may allow clinicians to distinguish between a necrotizing, metabolic or inflammatory myopathy and facilitate rapid and appropriate therapeutic management.

As regard as the appropriate use of muscle biopsy, sometimes this procedure needs to be preceded by serum molecular genetic testing to rule-out specific muscle pathologies as a dystrophinopathy or a myotonic muscular dystrophy type I, where the clinical examination is fairly predictive of the diagnosis.

When choosing the site for biopsy, the most important step is to locate a muscle that is affected by the disease. While this sounds simple, it is not always straightforward and can be challenging. If the disease process is chronic, progressive, and appears diffuse and symmetric, choosing the site is typically easy and can be done using Medical Research Council (MRC) strength grading, electrodiagnostic testing or muscle MRI. However, in acute onset weakness, when there is little concern that end-stage pathology is present, a muscle that is severely to moderately affected should be chosen.

TREATMENTS FOR MUSCLE GLYCOGENOSES

ANTONIO TOSCANO
Department of Clinical and Experimental Medicine of the University of Messina, Italy

Glycogen storage diseases are either dinamic or chronic and progressive muscle disorders with a wide range of phenotypic presentations . They are usually caused by an inherited deficiency of different glycogenolytic or glycolytic enzymes. In this field, the most relevant treatment has been applied to Pompe disease. In fact, since 2006, Pompe disease (as acid alpha-glucosidase deficiency) has been treated with an enzyme replacement therapy (ERT) with alglucosidase alfa that has been licensed for this specific purpose. Several studies have evaluated the clinical efficacy and safety of alglucosidase alfa treatment in juvenile and adult patients with late-onset Pompe disease (LOPD).

Overall, at least two-thirds of patients were stabilized or exhibited improvements in creatine kinase levels and muscular and/or respiratory function following treatment with alglucosidase alfa. ERT was well tolerated; the majority of adverse events were mild or moderate infusion-related reactions. In conclusion, alglucosidase alfa treatment offers an effective and well tolerated treatment that attenuates the progression of LOPD in the majority of patients. Nowadays, several other clinical trials are ongoing with different perspectives in Pompe disease.
McArdle disease (Glycogen Storage Disease type V) is a dynamic disorder caused by the absence of the glycolytic enzyme, muscle phosphorylase. People present with exercise-induced pain, cramps, fatigue, and myoglobinuria, which sometimes may result in an acute renal failure if severe.

About McArdle disease, at the moment, there is no evidence of significant benefit from any specific nutritional or pharmacological treatment in McArdle disease. However, small trials have suggested that low dose creatine produces slight benefit as well as ingestion of oral sucrose, immediately before physical exertion, improves exercise tolerance. Being McArdle disease a rare disorder, it is necessary to develop international multicentres collaboration for future trials.

CURRICULUM VITAE

ALBERTO ALBANESE
ITALY

Alberto Albanese is Professor of Neurology and Head of the Department of Neurology at the Humanitas Research Hospital in Milan. Is certified in Neurology and in Psychiatry and has published over 250 publications, including more than 200 scientific papers on indexed journals and several chapters on multi-authored books. Is Editor in Chief of Frontiers in Movement Disorders and Associate Editor of the European Journal of Neurology. He is a member of the Faculty of 1000, and has peer reviewed for a number of neurological journals.

Prof. Albanese has been President of the International Neurotoxin association and is a honorary member of the French Neurological Society and of the Swiss Neurological Society. He is regular member of several scientific societies.

OVIDIU BĂJENARU
ROMANIA

Corresponding Member of the Romanian Academy
Member of the Romanian Academy of Medical Sciences of Romania
Professor of Neurology and Director of the Clinical Neuroscience Department at the University of Medicine and Pharmacy "Carol Davila" Bucharest, Chairman of the Department of Neurology – University Emergency Hospital Bucharest

- Graduate of the Faculty of Medicine – University of Medicine and Pharmacy (UMF) „Carol Davila" Bucharest (1983)
- Specialist in Neurology (1989), Senior Neurologist (1994); competence in MRI

diagnostic in neurologic disorders (1991)

- PhD (1993) - UMF „Carol Davila" Bucharest
- 2006: Doctor Honoris Causa –University „Ovidius" – Constanta
- Postdoctoral specialization at the University „René Descartes" (Paris) during 1993-1994, in clinical Neurology (CHU „Saint-Anne" and „Kremlin-Bicetre") and research grants in Clinical and Experimental Neurophysiology (CHU „Cochin-Port Royale" and Faculté de Medecine Paris V)
- 2001-2013: President of the Romanian Society of Neurology
- Since 2013: Honorary President ad vitam of the Romanian Society of Neurology
- Since 2001: Coordinator and Chairman of all annual National Congresses of the Romanian Society of Neurology and many other scientific events and teaching courses organized for neurologists in Romania
- Visiting Professor in Vietnam (2013) and Kazakhstan (2015), on behalf of WFN
- Member of the Executive Committee of ENS (European Society of Neurology) between 2005-2009, of the Scientific Committee of ECTRIMS (2004-2009)
- Member of European Academy of Neurology (since 2014), American Academy of Neurology, International Parkinson's Disease and Movement Disorders Society, European Stroke Organisation, Danube Neurological Association (member of the Scientific Board and Deputy Secretary General), and others
- Since 2008: official representative of Romania for UEMS - European Board of Neurology (secretary of the Executive Committee between 2010-2015) and member of the examination board for the title of European Neurologist
- Author of more than 1000 scientific papers reported and published in scientific journals, among 147 cited in ISI Web of Science (Hirsch index 16) and Pubmed. Author of chapters in 2 international books of neurology and author and co-author in more than 15 medical books published in Romania.
- Coordinator of the National Diagnostic and Treatment Guidelines in Neurological Disorders
- National Principal Investigator and Investigator in more than 50 international, multicentric, controlled clinical trials in: stroke, Parkinson's disease and movement disorders, multiple sclerosis, dementia, epilepsy, and others.
- Director of more national research grants
- 9 awards of excellency in medicine from different socio-professional national and international organizations, the Romanian Ministery of Health and the Romanian Orthodox Patriarchate
- Initiator and coordinator of the National Medical Programs of the Ministery of Health and National Health Insurance System for the treatment of: acute stroke, multiple sclerosis, rare neurological diseases, advanced Parkinson's disease (1999 – 2015)
- President of Consultative Commision of Neurology of the Ministery of Health and National Health Insurance System (2008 – 2015)

PETER VAN DEN BERGH

BELGIUM

POSITION:
Full Professor of Neurology at the Université catholique de Louvain (UCL) and Director of the Neuromuscular Reference Centre of the University Hospital Saint-Luc, Brussels, Belgium.

ACADEMIC AND CLINICAL RESPONSABILITIES :
Head of the neuromuscular diseases and neuromuscular rehabilitation clinic, the electromyography lab, and the neuromuscular pathology lab; Teaching of clinical neurophysiology and neuromuscular pathology.

Creation of the NEUROMUSCULAR REFERENCE CENTRE at the University Hospital Saint-Luc, of which he is director and coordinator (since 1999).

Current RESEARCH INTERESTS :
- pathogenesis and treatment of inflammatory neuropathies (founding member of the Inflammatory Neuropathy Consortium – INC, a standing committee of the Peripheral Nerve Society - PNS)
- electrodiagnostic criteria of inflammatory neuropathies
- development of functional scales for neuromuscular disorders

MEMBERSHIPS AND INVOLVEMENT IN SCIENTIFIC SOCIETIES
- Organizer of an International Symposium on Neuromuscular Disorders – December 1999, Brussels, Belgium
- Organizer of an International Symposium of Neuromuscular Diseases – October 2005, Brussels, Belgium
- Organizer of Progress in Neuromuscular Disorders, the 10th Anniversary Symposium of the Neuromuscular Centre UCL St-Luc – November 2010, Brussels, Belgium
- Member of the Treat-NMD Alliance (coordinator for the 6 Belgian Neuromuscular Centres) (since 2008)
- Chairman of the EFNS and since 2015 the European Academy of Neurology (EAN) Scientific Neuropathy panel (since 2009)
- Member of the Biomedical Alliance Europe (BMA) for the European Affairs Subcommittee of the EAN (2016)
- Chairman of the Scientific Council of the Institute of Myology, Paris, France (since 2010)
- Member of the AFM Pathophysiological Basis of the Muscular Dystrophies commission

(2012-2017)

- President of the Belgian-Dutch Neuromuscular Study Group (since 1998)
- Member of WMS since 1997, participated at all WMS congresses except two
- Organizer of the 11th International Congress of the WMS (WMS11) – October 2006, Bruges, Belgium
- Editorial Board member of Neuromuscular Disorders since 2001
- Executive Associate Editor of Neuromuscular Disorders since 2016
- Member of the WMS Executive Board since 2007
- Member of the WMS Program Committee since 2007
- Member of the PNS since 2007)
- Founding member of the Board of INC (PNS)
- Vice-Chairman of INC (since 2018)
- Fellow of the EAN since 2018
- Board member of EURO-NMD responsible for the Neurophysiology Group) since 2017

JEAN-MARC BURGUNDER
SWITZERLAND

Jean-Marc Burgunder has graduated in Medicine at the Faculty of Medicine in Bern, Switzerland, and trained in internal medicine, neurology and neuroscience in Switzerland and at the Institutes of Mental Health in Bethesda, USA. He is a Professor of Experimental Neurology at the faculty of medicine of the University in Bern. He has spent some years as a Professor of Medicine at the National University of Singapore. He is a visiting Professor of Neurogenetics at the Central South University in Changsha and at the Sichuan University in Chengdu (China). He also holds a position as an adjunct professor at the Sun Yat Sen University in Guangzhou in China. He is Director of the Neurocenter, including the Swiss HD Center at Siloah in Gümligen (Bern), devoted to the care of patients with rare neurological disorders, along with the provision of general neurology services for the area. Chair of the EHDN Executive Committee, Founding Steering committee member of the Chinese Huntington's Disease Network, Fellow of the European Academy of Neurology, Chair of the European Reference Network on Rare Neurological disorders Advisory board.

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 to 30 october 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
- Chairman of the European Academy of Neurology Task Force on Rare Neurologic Diseases

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)
- Chairman of the EAN Task Force on Rare Neurologic Diseases
- 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 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 Assembly.
- 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, Bucarest

- 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.
- Chairman of the European Academy of Neurology Task Force on Rare Neurologic Diseases

The main scientific interest is Rare Neurologic Diseases (genetic, neurodegenerative and neurometabolic diseases): organization of platforms and methodologies for improving diagnosis. Teaching clinical and genetic strategies for diagnosis of rare neurologic diseases.

ALESSANDRO FILLA
ITALY

Full Professor of Neurology and Chairman of the Department of Neurological Sciences at Federico II, Naples, Italy.

EDUCATION:

1972 Medical Degree at the University of Naples
1973 Educational Council for Foreign Medical Graduates Certification
1975 Board Certification in Neurology

CLINICAL TRAINING/RESEARCH EXPERIENCE:

1976-78 Fellow of Research at the Department of Neurobiology of the Institute of Clinical Research of Montreal (Dr. A. Barbeau)
1992 Stage at the National Hospital, Queen Square, London (Dr. A. Harding)
1994 Stage at Columbia University, New York (Dr. S. Fahn)
1996 Stage at Hopital de la Salpetriere (Dr. Y. Agid)

ACADEMIC AND PROFESSIONAL DUTIES

1973 Assistant Professor of Anatomy
1974 Lecturer of Neurology at the Federico II University, Naples

1982 Senior Lecturer of Neurology
1982 Visiting Professor of Pharmacology and Neuropediatrics at University of Arizona
1992 Associate Professor of Neurology
2001 Full Professor of Neurology
2003-09 Chairman of the Clinical Department of Neurological Sciences
2007-12 Chairman of University Department of Neurological Sciences
2007 to date Director of the School of Neurology Residents
2018 co-chairman Neurogenetic Panel of the European Academy of Neurology

RESEARCH FIELDS
Molecular genetics and molecular pathogenesis of the recessive and dominant ataxias;
Molecular genetics and molecular pathogenesis of the hereditary spastic paraplegias;
Molecular genetics and molecular pathogenesis in Parkinson disease and parkinsonisms;
Epidemiology of hereditary ataxias and spastic paraplegias;
Clinical studies in hereditary ataxias;
Trials in hereditary ataxias.

280 pubblications, H-Index=46

HOLM GRAESSNER
GERMANY

POSITION TITLE:
Managing Director, Rare Disease Centre, University Hospital Tübingen

EDUCATION/TRAINING

Institution and location	Degree	Completion date	Field of study
Technical University Ilmenau, Germany	Dipl.Ing.	1993	Biomedical Engineering
University of Rostock, Germany	M.A.	1999	German language and literature / philosophy
University of Rostock, Germany	PhD	2004	History of Science
University of Reutlingen, Germany	MBA	2008	International Marketing

BIOGRAPHY

Holm Graessner has been Managing Director of the Rare Disease Centre, since 2010, at the University and University Hospital Tübingen, Germany. www.zse-tuebingen.de

He is Coordinator of the European Reference Network for Rare Neurological Diseases (ERN-RND). www.ern-rnd.eu

Together with Olaf Riess, he coordinates the H2020 Solve-RD project on "Solving the unsolved rare diseases". www.solve-rd.eu

He received his PhD "Summa cum laude" in 2004 and, then, he obtained his MBA degree in 2008.

From 2003 until now, he has been coordinating and managing more than 10 EU funded collaborative projects. The main focus of these projects are rare and neurological diseases, among them EUROSCA, MEFOPA, SENSE-PARK, MULTISYN, NEUROMICS and PROOF.

He has been co-leading one of the four working groups of the German Action Plan for Rare Diseases.

Since 2017, in his function as the coordinator of ERN-RND, he is a member of the Rare Disease Task Force of the European Academy of Neurology. In the Coordinator's Group of the European Reference Networks, he leads the cross-border healthcare working group.

PUBLICATIONS

1. RD-Connect, NeurOmics and EURenOmics: collaborative European initiative for rare diseases. Lochmüller H, Badowska DM, Thompson R, Knoers NV, Aartsma-Rus A, Gut I, Wood L, Harmuth T, Durudas A, Graessner H, Schaefer F, Riess O; RD-Connect consortium; NeurOmics consortium; EURenOmics consortium. Eur J Hum Genet. 2018 Feb 27. doi: 10.1038/s41431-018-0115-5. [Epub ahead of print] PMID: 29487416
2. European Reference Networks : Consequences for healthcare in Germany. Graessner H, Schäfer F, Scarpa M, Wagner TOF. Bundesgesundheitsblatt Gesundheitsforschung Gesundheitsschutz. 2017 May;60(5):537-541. doi: 10.1007/s00103-017-2533-x. German.
3. Participatory design in Parkinson's research with focus on the symptomatic domains to be measured. Serrano JA, Larsen F, Isaacs T, Matthews H, Duffen J, Riggare S, Capitanio F, Ferreira JJ, Domingos J, Maetzler W, Graessner H; SENSE-PARK Consortium. J Parkinsons Dis. 2015;5(1):187-96. doi: 10.3233/JPD-140472. PMID: 25588357

Since 2017, in his function as the coordinator of ERN-RND, he is a member of the Rare Disease Task Force of the European Academy of Neurology. In the Coordinator's Group of the European Reference Networks, he leads the cross-border healthcare working group.

REETTA KÄLVIÄINEN
FINLAND

Reetta Kälviäinen is the professor of Clinical Epileptology/Neurology in the University of Eastern Finland and the Director of the Kuopio Epilepsy Center in the Kuopio University Hospital. Her special research interest is clinical epileptology including identifying biomarkers of seizure activity, epileptogenesis, progression, and drug-resistancy in cohorts of newly diagnosed and drug-resistant chronic patients. These aspects of scientific projects are combined with therapeutic neuropharmacological interventions. She serves in the steering group of the European Reference Network for rare and complex epilepsies EpiCare (of which Kuopio Epilepsy Center is a member) and co-chairs the Epilepsy Scientific Panel of the European Academy of Neurology.

RAMONA MOLDOVAN
ROMANIA

PROFESSIONAL INTERESTS
I am currently lecturer at the Department of Psychology, Babeș-Bolyai University (RO) and visiting researcher at the University of Manchester and Bournemouth University (UK). I am a clinical psychologist trained in cognitive and behavioral psychotherapies with the Albert Ellis Insitute (USA) and in genetic counselling at the University of Manchester (UK). My clinical work focuses on serious mental illnesses as well as neurodevelopmental and neurodegenerative disorders. My research interests are related to the applications of clinical psychology and cognitive and behavioural therapies in genetic counselling. I am particularly interested in translating "big data" in evidence-based psychosocial interventions and training.

SCIENTIFIC EXPERTISE

Over the last 10 years I participated in or coordinated a number of experimental and clinical trials investigating the mechanisms of change and efficacy of cognitive and behavioral psychotherapies (e.g. for major depression, generalized anxiety disorder, ADHD, autistic spectrum disorders) as well as psychological factors associated with several psychiatric and genetic disorders (e.g. Huntington's Disease). Since 2013 I am head of a research group investigating psychiatric genetic counselling. My current work includes a randomized controlled trial exploring the efficacy of genetic counselling for psychiatric disorders (e.g. schizophrenia, schizoaffective and bipolar disorder) and a pilot trial investigating the role of psychological interventions for post predictive testing for Huntington Disease.

MARIA JUDIT MOLNÁR

HUNGARY

Maria Judit Molnár MD, PhD, Professor of Neurology, Psychiatry, Clinical Genetics, and Clinico-pharmacology, Doctor of the Hungarian Academy of Sciences is the director of Semmelweis University's Institute of Genomic Medicine and Rare Disorders, among others president of the Hungarian Medical College of Clinical Genetics, past president of the Hungarian Society of Clinical Neurogenetics, secretary of the Hungarian Society of Personalized Medicine, Co-Chair of the Neuromuscular Scientific Panel and management board member of the Neurogenetic Scientific Panel of the European Academy of Neurology. She was the vice-rector for Scientific Affairs at Semmelweis University (Budapest, Hungary) between 2012 and 2015, where she was also responsible for International Affairs. She has been adjunct professor at the Montreal Neurological Institute, McGill University, between 1999 -2012. Dr. Molnár is the Facilitator of a Challenge Group of the International Consortia of Personalized Medicine initiated by the European Commission. She is the member of the steering committee of the Association of Academic Health Centers Internationals.

Dr. Molnár is recognized as a leading experts on the diagnosis and treatment of rare neurological disorders. The Institute of Genomic Medicine and Rare Disorders lead by her offers a comprehensive state of the art, patient-centered multidisciplinary care for patients with rare neurological disorders including genetic testing, neuropathological investigations and genetic counselling as well. Dr. Molnar's research covers a broad range of basic and clinical studies on rare neurological disorders, utilizing a broad spectrum of technologies including clinical science, molecular genetics including next generation sequencing and bioinformatics as well. The Institute of Genomic Medicine and Rare Disorders is the

part of the European Reference Network of Rare Neurological Disorders (ERN-RND) and Neuromuscular Disorders (ERN-NMD). Dr. Molnár is the member of the management board of the ERN-RND as the work package leader for Education and capacity building.

She plays important role in the organization of rare disease management in Hungary and acts as an ambassador promoting the personalized healthcare. She is the President of the Advisory Board of Rare Disorders, the official advisory board of the Hungarian Insurance Fund. She was the principal investigator of several clinical trials, published 1 book, 20 book chapters, 137 papers with more than 1500 citations. She owns 2 patents. She is active in postgraduate education, 7 PhD students defended their thesis and 5 are active in their education. Several neurologists and clinical geneticist has been trained by her.

DAFIN F. MUREȘANU

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 (EFNRS), 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 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. Mureșanu 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. Mureșanu 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 (176 papers indexed on Web of Science-ISI, H-index: 18) as well as contributions in monographs and books published by prestigious international publishing houses. Prof. Dr. Dafin F. Mureșanu 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.

DAVIDE PAREYSON
ITALY

Born in Turin, Italy, 21st June, 1959
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 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).

He is the Coordinator of the Italian National Registries of Charcot-Marie-Tooth disease and of Spino-Bulbar Muscular Atrophy (www.registronmd.it).

He is among the organizers of three ENMC workshops on CMT and participated into two other workshops.

He organized the 6th International meeting of the Charcot-Marie-Tooth and Related neuropathies consortium (CMTR) in Mestre-Venice, 8-10th Sept. 2016.
He has co-authored 229 papers on peer-reviewed Journals (Pubmed) mainly on hereditary disorders and neuromuscular diseases. H-index = 40 (Scopus)

POSITIONS AND HONORS
Positions:

1981-1982	- Internship, Clinic of Medical Pathology, Milan University.
1982-1984	- Internship, IRCCS Foundation, C.Besta Neurological Institute (INCB), Milan.
1985-1989	- Postdoc. Fellowship, Besta Institute, Milan.
1989-1996	– Assistant Neurologist in the Department of Neurology, INCB, Milan, Italy.
1996-2003	– Associate Neurologist in the Department of Neurology, INCB, Milan.
2003-2007	– Associate Neurologist in the Department of Biochemistry and Genetics, INCB, Milan.
2007-2010	– Head of the Simple Unit (SOS) “Clinic of Central and Peripheral Degenerative Neuropathies” - Department of Clinical Neurosciences, INCB, Milan.
2010-2017	– Head of the Simple Department Unit (SOSD) “Clinic of Central and Peripheral Degenerative Neuropathies” (named Rare Neurological Diseases since 2016) - Department of Clinical Neurosciences, INCB, Milan.
2012-....	– Head of Functional Department. on Rare Neurological Diseases, INCB,

Milan.

2017-.... - Head of the Complex Unit (UOC) "Rare Neurodegenerative and Neurometabolic Diseases", INCB, Milan.

OTHER EXPERIENCE, PROFESSIONAL MEMBERSHIPS, HONORS:

2016-2019 - Chair of the CMTR, Charcot-Marie-Tooth neuropathy & Related diseases consortium

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

2012-2014 - Member of the Executive Committee of the European Neurological Society (ENS)

2006-2013 - Coordinator of the Clinical Neurogenetics Subcommittee of the ENS

2016- - Deputy Chair of the Neuropathy Group of the EURO-NMD ERN (European Reference Network for Neuromuscular Disorders).

2017- - 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).

RESEARCH SUPPORT
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ANTONIO TOSCANO
ITALY

Antonio Toscano is Professor of Neurology, since 2009, at the Department of Clinical and Experimental Medicine of the University of Messina, Italy.

He received his MD "cum laude" in 1981 and, then, he specialized in Neurology in 1985 in the University of Messina.

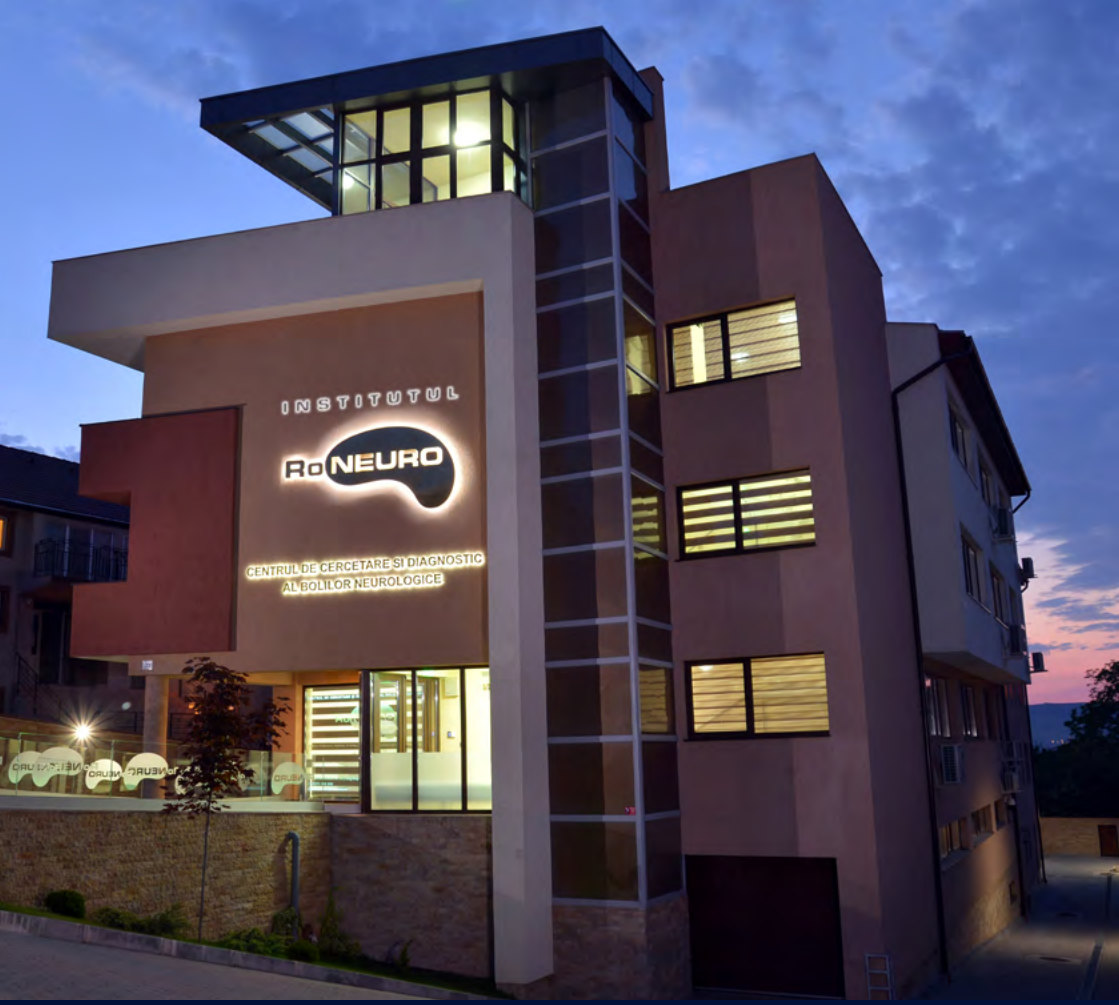
From 1986 to 1987, he attended as a fellow "The National Hospital for Nervous Diseases, London, UK, under the guide of dr. John Morgan-Hughes, studying Mitochondrial Disorders. Since 2016, he is responsible of a ERN Reference Center for Rare Neuromuscular disorders at the University Hospital of Messina, Italy.

He has been President and past President of the Italian Association of Myology (AIM) (2009-2015) Since 2016, Chairman of the EAN Panel for Muscle and Neuromuscular Junction Disorders,
Since 2017, Treasurer of the Italian Society of neurology (SIN)i

Since 2018, Dean of the Faculty of Medicine of the University of Messina

He is also member of: a) National Board of the Italian Neurological Society (SIN), b) Board of the European Consortium for Pompe Disease (EPOC), c) International board of the Pompe registry, d) several other National and International Scientific Societies and Groups

His main research interests are focused on Neuromuscular and Neurodegenerative Disorders with particular attention to Metabolic Myopathies and, more specifically, to pathogenic, clinical and therapeutic aspects of muscle glycogenoses (i.e. Pompe disease), lipid storage myopathies and mitochondrial encephalomyopathies or other rare neurodegenerative disorders. In these fields, he has published over 190 papers.



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