2.30 PM Introduction
Saba Motta - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

#### 2.40 PM Presentation

Giuseppe Lauria - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

2.50 PM Monsieur Dejerine de la Salpêtrière
Fabio Simonetti - Fondazione IRCCS Istituto Nazionale dei Tumori, Milano

3.20 PM Early onset hereditary neuropathies: natural history and trial readiness Isabella Moroni - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

3.40 PM The Next-Generation Sequencing revolution and its impact on an old disease. Franco Taroni - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

4.00 PM Inherited neuropathies: the pathway to treatment, where are we? Davide Pareyson - Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

4.20 PM Discussion



Dr Simonetti will make an historical overview of Jules Déjerine (1849-1917), successor of Fulgence Raymond to the chair of the Salpetrière. His name is linked to the sporadic form of olive-ponto-cerebellar atrophy, facio-scapulo-humeral myopathy, hypertrophic neuritis and thalamic syndrome. He definitively delivers to the memory of neurologists "Sémiologie des affections du Système Nerveux", the first text in which neurological semeiotics is treated as a discipline, with formidable iconography and case series.



Dr. Moroni will provide an overview on hereditary peripheral neuropathies, and will focalize mainly on infantile onset forms. These are rare disorders presenting with heterogeneous clinical phenotypes and causing variable degrees of impairment and disability since infancy, when they can be associated with central nervous system involvement. The inheritance pattern can be autosomal dominant, recessive and X-linked. Compared with adults, children present with relatively more frequent AR or sporadic forms, raising challenges for the differential diagnosis with acquired neuropathies.



Dr. Taroni will give an overview of the molecular bases of Charcot-Marie-Tooth disease and related neuropathies. Given the great number of genes involved and the phenotypic overlap, diagnosis of these forms is challenging. The talk will illustrate how the Next-Generation Sequencing technology contributes to the field, also generating new insights and discoveries.



Dr Pareyson will present the progresses of drug therapies for Charcot-Marie-Tooth disease (CMT) and related neuropathies. Particularly, for the most common forms of CMT, numerous promising compounds are under study in cellular and animal models, mainly targeting either the protein degradation pathway or the protein overexpression. Efforts are also devoted to develop responsive outcome measures and biomarkers for this overall slowly progressive disorder, with quantitative muscle MRI resulting the most sensitive-to-change measure.





**UPDATES IN NEUROSCIENCES** 

January- December 2018

KNOWLEDGE IS SPEAKING, WISDOM IS LISTENING

Jimi Hendrix

## **LECTURE THEMES**

- THE WHITE THAT MATTERS
  White Motter Diseases
- THE GREYISH OF BRAIN Gray Matter Diseases
- GOOD NEWS MOVING FAST Movement Disorders
- TAKING A PEEK INTO PAIN, FROM THE SKIN TO THE BRAIN Neuroalgology
- THE EXCITABLE BRAIN
  Epileptology
- LET'S FOLD AGAIN Proteinopathies
- A BRAIN SHARPER THAN
  THE KNIFE
  Neurosurgery
- IT'S HIGH TIME Innovative Therapy Approaches
- UNDISCIPLINED CELLS
  Neuro-oncology
- ONE UNIT, ONE MOTOR
  Neuromuscular Diseases





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2.30 PM - 4.00 PM

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Jimi Hendrix

# SCIENTIFIC COMMITTEE

Giuseppe Lauria Paola Caroppo Sara Prioni

## ORGANIZATION SECRETARIAT

Loredana Vincenzi Scientific Directorate Saba Motta Library

Training Office, Updating and Didatics

formazione@istituto-besta.it



## LOCATION AND TIME

Biblioteca Scientifica Fondazione IRCCS Istituto Neurologico Carlo Besta via Celoria, 11 Milano

TIME: 2.30 PM - 4.00 PM

## **RECIPIENTS**

Doctors and Specializing in Neurology, Neurosurgery, Neuropediatric, Child and Adolescent Psychiaty, Pediatrics, Psychologists, Biologists.

No admission fee.

CME CREDITS HAVE BEEN REQUESTED.