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Leonardo Caporali

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Michele Carbonelli

IRCCS Institute of Neurological Sciences, Bologna UOC
Neurological Clinic Bellaria Hospital

Valerio Carelli

Medical Director of UOC Neurological Clinic AUSL Bologna
and Head of the Neurogenetics Laboratory

Maria Lucia Cascavilla

Head of Neuro-ophthalmology service and ocular electrophysiology
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Thomas Klopstock

Friedrich-Baur-Institute, Dept. of Neurology, LMU Klinikum Munich, Germany

Chiara La Morgia

Researcher at the Biomedical and Neuromotor Sciences Department, Bologna
Medical Director UOC Neurological Clinic IRCCS Neurological Sciences Institute, Bologna

Alessandra Maresca

Healthcare researcher - IRCCS Neurological Sciences Institute, Bologna

Martina Romagnoli

Biotechnologist fellow IRCCS
Neurological Sciences Institute, Bologna

Patrick Yu-Wai-Man

Professor of Ophthalmology and Honorary Consultant Ophthalmologist,
Moorfields Eye Hospital, London (UK), Addenbrooke's Hospital,
Cambridge (UK), University of Cambridge, Cambridge (UK)

Course Venues:

MILAN

Ophthalmology Clinic Vita-Salute University IRCCS San Raffaele Hospital
Via Olgettina, 60 - 20132 Milan

BOLOGNA

IRCCS Neurological Sciences Institute of Bologna
UOC Neurological Clinic, Bellaria Hospital
Department of Biomedical and Neuromotor Sciences, Bologna University
Via Altura, 3 - 40139 Bologna

Destinatari dell'attività formativa ECM - Italia

Medico Chirurgo: Oftalmologia, Neurologia

Event Addressed to Ophthalmologists and Neurologists

PROVIDER ECM 362
ORGANIZING SECRETARIAT:



First Class - Meetings and Conferences
Via L. Mascheroni 17, 20145 Milano
Tel. +39 02 30066329 - Fax 0586.349920
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Viale Italia, 173 - 57128 Livorno
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5 - 6 October 2022

MASTER CLASS
on LHON

Leber's Hereditary Optic Neuropathy

MILANO
BOLOGNA



Course Directors:

Dott. Piero Barboni (Milano)
Prof. Valerio Carelli (Bologna)

in collaboration with



ALMA MATER STUDIORUM
UNIVERSITÀ DI BOLOGNA
DIPARTIMENTO DI
SCIENZE BIOMEDICHE E NEUROMOTORIE

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BOLOGNA



This is an international project addressed to a selected target of medical specialists in Ophthalmology and Neurology, specialized in the diagnosis and therapy of Leber's disease and it is projected and supported by an international Faculty

The program will start from a general framework of the pathology and goes on to diagnosis, treatment and scientific research.

The course will be held in two centers of Excellence, one specialized in research and the other in the treatment of LHON. The part dedicated to clinical treatment will be held at the Department of Ophthalmology of the San Raffaele Hospital in Milan, while the in-depth study of LHON research will be held at the Institute of Neurological Sciences in Bologna.

5 OCTOBER 2022

MILAN, *Ophthalmology Clinic Vita-Salute University IRCCS San Raffaele Hospital Via Olgettina, 60 Milan*

08.30 - 09.00 **Introduction and Presentation of the Course** | *F. Bandello*

Reference Centres and Management of NO diseases and Rare Eye Disease
F. Bandello, P. Barboni, M.L. Cascavilla, M. Battista

09.00 - 10.00 **LHON - Disease description:**

- **Introduction and background**
- **Epidemiology**
- **Disease demographics**
- **Clinical manifestations**
- **Visual prognosis and recovery**
- **Anatomical and metabolic consideration**

Discussion - Q&A

10.00 - 11.00 **Ophthalmoscopic findings:**

Asymptomatic stage, Pre-symptomatic stage, Symptomatic stage

Discussion - Q&A

11.00 - 11.30 **Coffee Break**

11.30 - 12.30 **OCT findings**

Retinal nerve fiber layer analysis

Ganglion cell analysis

Optic nerve head analysis

Discussion - Q&A

12.30 - 14.00 **Lunch**

14.00 - 17.00 **Flow charts and focus on Differential diagnosis**

Atypical features in practice: Childhood onset, Late onset, LHON plus

Interactive presentation and discussion of clinical cases

17.00 **Closing remarks - first day**

6 OCTOBER 2022

BOLOGNA, *RCCS Neurological Sciences Institute of Bologna UOC Neurological Clinic, Bellaria Hospital Department of Biomedical and Neuromotor Sciences, Bologna University Via Altura, 3 Bologna*

08.30 - 09.00 **Introduction and Presentation of the second part of the Course** | *V. Carelli*

Reference Centres and Management of LHON and Rare Eye Diseases | *V. Carelli*

09.00 - 10.00 **Basis of Mitochondrial medicine:**

- **Mitochondrial genetics** | *V. Carelli*
- **Leber's disease: genetics framing** | *V. Carelli*
- **Cellular pathogenic mechanism and ipSCs** | *A. Maresca*

Discussion Q&A | *V. Carelli*

10.00 - 11.00 **Genetics, databases and new perspectives in LHON:**

- **Genetic testing, analysis of patient and family genetic tree and counselling** | *L. Caporali*
- **LHON & DOA database and ongoing observational studies** | *M. Romagnoli*
- **Artificial intelligence approaches to LHON** | *M. Romagnoli*

11.00 - 11.30 **Coffee Break**

11.30 - 12.30 **Pharmacological approaches in LHON** | *T. Klopstock*

Regulatory rules for medical prescription in UE

12.30 - 13.30 **Lunch**

13.30 - 14.30 **Genetical approach to LHON** | *P. Yu-Wai-Man*

Clinical trials on gene therapy

14.30 - 16.00 · **Neuro-ophthalmological follow-up** | *M. Carbonelli*

· **Genotype-Phenotype correlations** | *C. La Morgia*

· **Triggering and hormonal factors** | *G. Amore*

· **Extra-ocular features** | *C. La Morgia*

· **Interactive presentation and discussion of clinical cases**

16.00 - 16.30 **Visit to the laboratories** | *L. Caporali, A. Maresca, V. Carelli*

16.30 **Closing remarks**