

Provider EDU.co ID 1357

PROGETTO

di Formazione a Distanza (FAD)

TITOLO EVENTO

**Clinical Approach to LHON:
From Diagnosis to Treatment Through Real Case Studies**

OBIETTIVO FORMATIVO: Application of evidence-based practice (EBM - EBN - EBP) principles and procedures in daily clinical work

Destinatari dell'attività formativa:

Professione | Discipline

- BIOLOGO | BIOLOGO
- ORTOTTISTA/ASSISTENTE DI OFTALMOLOGIA | ISCRITTO NELL'ELENCO SPECIALE AD ESAURIMENTO
- ORTOTTISTA/ASSISTENTE DI OFTALMOLOGIA | –
- MEDICO CHIRURGO | GENETICA MEDICA; LABORATORIO DI GENETICA MEDICA; NEUROLOGIA; OFTALMOLOGIA
- INFERMIERE | INFERMIERE

N. crediti ECM: 5

Numero partecipanti: 100

ORE ATTIVITA' FORMATIVA: 5

Responsabile Scientifico: Piero Barboni

RAZIONALE

SCIENTIFIC RATIONALE:

Neuro-ophthalmology is a highly specialized and often underexplored field that focuses on the intersection between neurology and vision. Early recognition of neuro-ophthalmologic conditions is critical to improve outcomes, yet remains challenging due to their rarity and complexity.

Leber's Hereditary Optic Neuropathy (LHON) is a rare mitochondrial disease typically presenting with acute or subacute vision loss in young adults. Its low prevalence contributes to frequent diagnostic delays or misdiagnosis, which significantly impacts patient care and genetic counseling.

This paper-based FAD course provides both theoretical and practical insights into LHON, aiming to improve diagnostic accuracy, raise awareness of its genetic underpinnings, and promote interdisciplinary management strategies. Learning is built around real-life clinical cases, enabling participants to enhance clinical reasoning and differential diagnosis skills through case-based reflection.

LEARNING OBJECTIVES:

Upon completion of the course, participants will be able to:

- Recognize the key clinical features of LHON;
- Differentiate LHON from other optic neuropathies;
- Understand the appropriate diagnostic pathway when LHON is suspected;
- Assess the clinical relevance of mitochondrial genetics in patient management;
- Value interdisciplinary collaboration in the management of LHON.

Course Content and Structure

This paper-based distance learning course is organized into a sequence of **modular learning units**, aimed at improving diagnostic reasoning, clinical decision-making, and awareness of Leber's Hereditary Optic Neuropathy (LHON) through immersive and interactive case-based learning.

Course Structure

The course is composed of **five thematic modules**, each divided into two distinct phases:

- **Module A** (Case Presentation & Clinical Decision-Making):
 - Presentation of a real clinical case related to LHON or its differential diagnosis.
 - After reading the case, participants will be prompted to answer a series of **multiple-choice questions** designed to assess their diagnostic orientation, interpretation of clinical findings, and therapeutic reasoning.
 - Responses will be collected regardless of their correctness to promote reflection and engagement.

- **Module B** (Debriefing & Educational Deep Dive):
 - In the second module, a **structured debriefing** will be provided.
 - The correct answers to the questions from Module A will be explained in detail, supported by **scientific references, guidelines, and visual or textual materials** to consolidate the participant's learning.
 - This part will also highlight critical learning points and interdisciplinary management considerations.

At the end of the five dual-module sequences, the course concludes with two final modules:

- **Module 11 – Satisfaction Questionnaire** (standard evaluation form)
- **Module 12 – Final CME Assessment Test**, with multiple-choice questions **different from those presented during the clinical cases**, covering the full scope of the course.

Detailed Case Modules

◆ **MODULE 1 – Clinical Case 1: Unexplained visual loss in glaucoma**

Patient Profile: A 70-year-old woman with a known diagnosis of glaucoma experiences sudden vision deterioration despite well-controlled intraocular pressure. Her family history reveals a distant relative affected by LHON.

Diagnostic Focus:

- The underestimated value of **family history** in optic neuropathies.
- Possible **coexistence of LHON and glaucoma**: coincidence or causation?
- What diagnostic re-evaluation should be considered?

Learning Points:

- Importance of mitochondrial DNA screening in atypical glaucomatous vision loss.
 - Risk of misdiagnosis and therapeutic inertia.
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◆ **MODULE 2 – Debriefing on Case 1**

Explanation of correct answers with literature insights on genetic testing indications and overlapping pathologies.

◆ **MODULE 3 – Clinical Case 2: Acute monocular visual loss and amblyopia in the fellow eye: is it a correct diagnosis?**

Patient Profile: A 26-year-old man was referred for a neuro-ophthalmological evaluation following the finding of painless acute visual loss and optic disc edema in his right eye.

Diagnostic Focus:

- Acute painless visual loss in a young adult
- Differential diagnosis: optic neuritis vs LHON
- Atypical and unilateral presentation of LHON
- OCT and fundoscopic clues supporting hereditary optic neuropathy

Learning Points:

- LHON can mimic optic neuritis, especially at onset
- Optic disc edema with microangiopathy suggests LHON
- Presumed amblyopia may represent early-onset LHON
- OCT and genetic testing are key for accurate diagnosis

◆ **MODULE 4 – Debriefing on Case 2**

Insights into neurosurgical implications and rare recovery pathways in LHON.

◆ **MODULE 5 – Clinical Case 3: Childhood LHON with Strabismus**

Patient Profile: A 3-year-old girl presents with sub-acute visual loss and **strabismus**, and is positive for the 3460ND1 LHON mutation. She receives idebenone and occlusion therapy.

Diagnostic Focus:

- Atypical pediatric presentation of LHON.
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- Is strabismus a **negative prognostic factor**?
- Evidence on idebenone use in **early childhood**.

Learning Points:

- Pediatric management strategies for hereditary optic neuropathies.
- Role of neuro-visual rehabilitation.

◆ **MODULE 6 – Debriefing on Case 3**

Visual pathway testing interpretation and LHON diagnostic flowcharts.

◆ **MODULE 7 – Clinical Case 4: Recovery of visual acuity and visual field over four years after LHON onset.** A 16-year-old boy, belonging to an Italian family affected by LHON, complained of subacute, painless visual loss in his right eye (OD).

Diagnostic Focus:

- Subacute painless bilateral visual loss in a young patient with familial LHON
- Genetically confirmed LHON (m.11778/ND4)
- Long-term functional recovery after severe visual impairment
- Structure–function correlation on OCT and visual fields

Learning Points:

- LHON may show delayed visual recovery, even years after onset
- Visual acuity and visual field recovery may follow different timelines
- Idebenone may contribute to stabilization and late functional improvement
- Residual viable fibers and neuroplasticity may support late recovery
- Long-term follow-up is essential to capture meaningful outcomes

◆ **MODULE 8 – Debriefing on Case 4**

Analysis of clinical evidence for visual recovery timelines and therapy impact.

◆ **MODULE 9 – Clinical Case 5: Visual Recovery After Brain Surgery**

Patient Profile: Two brothers, now aged 60 and 62, both experienced bilateral vision loss in their early 20s. Remarkably, both had partial vision recovery following neurosurgical intervention.

Diagnostic Focus:

- Differential diagnosis between **optic neuropathy and structural CNS pathologies**.
- Role and incidence of **opto-chiasmatic arachnoiditis**.
- Association between **meningeal inflammation/surgery and recovery** in mitochondrial diseases.

Learning Points:

- How rare structural lesions may overlap with mitochondrial presentations.
- The concept of reversible dysfunction in neuro-ophthalmology.

◆ **MODULE 10 – Debriefing on Case 5**

Therapeutic considerations in early-onset LHON and integrated vision care.

📄 **MODULE 11 – Satisfaction Questionnaire**

Standard course evaluation to assess content relevance, clarity, and applicability.

📄 **MODULE 12 – Final CME Assessment Test**

A multiple-choice test composed of questions **not previously encountered**, aimed at verifying overall learning and ensuring independent knowledge acquisition.

Compilazione Questionario ECM (erogato in modalità digitale)

ECM TABELLA FACULTY

I CV DELLA FACULTY SONO ARCHIVIATI E CONSULTABILI PRESSO L'ENTE ACCREDITANTE

NOME E COGNOME	LAUREA/PROFESSIONE	SPECIALIZZAZIONE /DISCIPLINA	AFFILIAZIONE/ ENTE DI APPARTENENZA/LIBERA PROFESSIONE	DESCRIZIONE ATTIVITA' PROFESSIONALE/FORMATIVA
Piero Barboni	Medicina e Chirurgia	Oftalmologia	Ospedale San Raffaele, Milano	Medico Specialista

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Il provider, ai sensi dall' art. 47 del DPR n.445/2000, consapevole delle conseguenze previste dall'art. 76, dichiara:

- di aver fornito all'interessato l'informativa sul trattamento dei dati personali (art. 13 del Regolamento europeo 2016/679; artt. 68, 70, 76, 96 Accordo Stato-Regioni 2017 "La formazione continua nel settore salute" - Rep. Atti 14/CSR del 2.2.2017 - Par. 4.6, lett. j) Manuale Nazionale di Accreditemento per l'Erogazione di Eventi ECM);*
- di aver informato l'interessato che il programma dell'evento ECM, di cui le suddette informazioni contribuiscono a formarne il contenuto minimo, verrà inserito nel catalogo degli eventi E.C.M. tenuto dall'ente accreditante;*