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Workshop

Gene Therapy for Lafora Disease: State of the Art and Emerging Perspectives

Perugia, 27th February 2026

Section of Neurology – Neurophysiopatology,
Department of Medicine and Surgery,
University of Perugia

Room 9, Building B, floor -2,
Gambuli Square 1, Perugia

SCIENTIFIC RATIONALE

Lafora disease is a rare neurodegenerative disease associated with progressive myoclonus epilepsy, for which no disease-modifying therapy is currently available. Over the past few years, substantial progress has been made in understanding its molecular mechanisms, developing advanced experimental models, and exploring innovative therapeutic strategies, particularly in the field of gene therapy.
This workshop aims to bring together international experts working across basic, preclinical, and clinical research to critically review the state of the art in Lafora disease research. Special emphasis will be placed on experimental models, pathogenic mechanisms, preclinical proof-of-concept studies, translational biomarkers, and future clinical applications. By stimulating dialogue among scientists, clinicians, patient organizations, and families, the event seeks to promote knowledge exchange, collaboration, and the translation of research advances into tangible clinical benefit.

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Cinzia Costa

Scientific secretariat
Lorenzo Gaetani; Valentina Imperatore; Andrea Mancini; Miriam Sciaccaluga

FACULTY

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Luis Zafra-Puerta
Researcher, Instituto de Investigación Sanitaria-Fundación Jiménez Díaz, Universidad Autónoma de Madrid

Program

12:30 – 13:30 | Registration
13:30 – 14:00 | Welcome
14:00 – 14:30 | Institutional Remarks
Introduction: *L. Parnetti – C. Costa*

14:30 - 15:30 Session I – Clinical Context and Molecular Profiling in Lafora Disease
Chairs: C. Costa – B. Cellini

From patient to gene and back to the patient
J. M. Serratosa

Biochemical pathways in Lafora Disease
D. Chiasserini

Comparative Electrophysiological Characterization of Lafora Disease Models
L. Bellingacci

Discussion

15:30 – 16:50 Session II – Gene Therapy: Preclinical Proof-of-Concept and Key Findings
Chairs: M. Sciaccaluga – B. Bettegazzi

Intracerebroventricular EPM2A and EPM2B therapies
M. P. Sánchez

Optimizing EPM2A therapy through intraparenchymal administration
N. Iglesias-Cabeza

Intravenus delivery of EPM2A and EPM2B genes
L. Zafra Puerta

Gene therapy and neuronal function: electrophysiological insights from preclinical models
M. Sciaccaluga

Discussion

16:50 – 17:10 Coffee break

***Session III* – Translational Integration and Future Perspectives**
17:10 – 18:10
Chairs: C. Costa – P. Prontera

From Preclinical Models to Clinical Perspectives in Lafora Disease
A. Mancini

Fluid Biomarkers of Neurodegeneration in Lafora Disease
L. Gaetani

Gene Therapy for Monogenic Disease: What Are We Learning?
S. Martino

Discussion

***Community Voices* - Final Discussion with Families and Associations**
18:10 – 18:40
G. Baschirotto – M. Gatti