GENOMIT: a Global Registry for Mitochondrial Disorders

Thomas Klopstock

Thomas Klopstock (1), Carole L. Linster (2), Martin Jakubec (2), Boriana Büchner (1), Holger Prokisch (3)
1) Department of Neurology, Friedrich-Baur-Institute, LMU University Hospital, Ludwig-Maximilians-Universität (LMU) München, Munich, Germany
2) Luxembourg Centre for Systems Biomedicine, University of Luxembourg
3) Institute of Human Genetics, TUM-University Hospital, Munich, Germany

The GENOMIT registry is an international clinician-driven platform designed to overcome the fragmentation of rare disease data by harmonizing international mitochondrial cohorts into a unified network for clinical trial readiness. A prime example of the registry's capability is the natural history study for Leber Hereditary Optic Neuropathy (LHON), which leverages aggregated, high-quality phenotype-genotype data to map precise disease progression and establish definitive endpoints for upcoming therapeutic trials.

Moving beyond standard genomics, an ongoing collaboration with research groups in Luxembourg is currently utilizing patient biobank samples to perform advanced metabolomics and exposomics analyses. By overlaying systemic metabolic profiles and environmental exposure factors onto established clinical phenotypes, this partnership aims to uncover novel disease modifiers and actionable therapeutic targets, demonstrating how active clinician participation in GENOMIT directly drives the next generation of translational mitochondrial medicine.

References

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Biosketch – Thomas Klopstock

Thomas KLOPSTOCK, Prof, MD, FEAN

ORCID ID: 0000-0003-2805-4652

CURRENT POSITIONS

Professor of Neurology, LMU University Hospital, LMU Medizin, Ludwig-Maximilians-Universität (LMU) München, Munich, Germany.

Project Leader, German Center for Neurodegenerative Diseases (DZNE), Munich, Germany.

OTHER RELEVANT PROFESSIONAL ACTIVITIES AND ACCOMPLISHMENTS

Professor of Neurology at LMU Munich, Germany. Long-standing clinical and scientific expertise in mitochondrial and other neurogenetic disorders. Speaker of German network for mitochondrial disorders (mitoNET) and of international collaborative project TIRCON (Treat Iron-Related Childhood-Onset Neurodegeneration). Member of German Center for Neurodegenerative Diseases (DZNE), and board of directors of Munich Center for Rare Diseases (MZSE). Ample experience in clinical trials in mitochondrial and other neurogenetic diseases (e.g., LHON, PKAN, Friedreich ataxia). Author of >400 peer-reviewed publications. Main goal is contributing to disease-modifying treatments of mitochondrial and other neurogenetic disorders.

Biosketch – Carole Linster

Carole LINSTER, Prof, PhD

ORCID ID: 0000-0001-7143-0719

CURRENT AND PAST POSITIONS

Current: Full Professor of Biochemistry, University of Luxembourg (UL); Group Leader, Enzymology & Metabolism, Luxembourg Centre for Systems Biomedicine (LCSB).

EDUCATION

PhD Thesis in Biomedical Sciences, UCLouvain, Sept. 2006.

Master's Degree in Health Sciences, UCLouvain, Sept. 2004.

Master's Degree in Biomedical Sciences, UCLouvain, June 2001.

Bachelor's Degree in Biomedical Sciences, UCLouvain, June 1999.

AWARDS AND HONORS

Grants: >€9 million raised from competitive and private sources; current projects: FNR CORE, FNR TREK, ELA International, EJP RD, Pélican, private fundraising; Partner PI in rare disease EU consortia (CHARLIE, GENOMIT); former PI of FNR INITIATE project (Losch Centre).

Since 2024: Effective member, Institut Grand-Ducal de Luxembourg, Section des Sciences.

2020: FNR Award for Outstanding Scientific Publication.

2009: Postdoctoral Recognition Award from the UCLA Molecular Biology Institute.

2000/2001: Paul Mahieu Prize for the Best Undergraduate Thesis in Biomedical Sciences (UCLouvain).

OTHER RELEVANT PROFESSIONAL ACTIVITIES AND ACCOMPLISHMENTS

Carole Linster is a Full Professor at the University of Luxembourg and leads the Enzymology & Metabolism Group at the Luxembourg Centre for Systems Biomedicine (LCSB). Her research combines classical biochemistry with metabolomics to uncover new enzyme functions in cellular metabolism, with a particular focus on metabolite damage-and-repair pathways and inherited metabolic disorders. Together with her colleagues, she discovered multiple novel metabolite repair enzymes and identified a rare pediatric neurodegenerative disease caused by NAXD deficiency. Her group integrates mechanistic enzymology with disease modeling in yeast, human cells, and zebrafish to dissect disease mechanisms and explore therapeutic strategies, highlighting how metabolomics can bridge molecular understanding with translational opportunities in human health.