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Endocrinologia



**European
Reference
Network**
for rare or low prevalence
complex diseases
Network
Hereditary Metabolic
Disorders (MetabERN)



ECLip2023

European Consortium of Lipodystrophies
IX symposium



September 28-29th 2023

Pisa, Italy

**Centro Congressi Le Benedettine
Piazza S. Paolo a Ripa D'arno, 16**

Scientific Committee:

Giovanni Ceccarini, Caterina Pelosini, Ferruccio Santini

AGENDA SEPTEMBER 28th

08:00 - 08:30	Registration
08:30 - 09:15	Introduction <i>Ferruccio Santini, Director, Endocrinology Unit, University Hospital of Pisa</i> Welcome from the Authorities <i>Riccardo Zucchi, Magnifico Rettore, University of Pisa</i> <i>Silvia Briani, Director, University Hospital of Pisa</i> <i>Emanuele Neri, Dean, School of Medicine, University of Pisa</i> <i>Stefano Taddei, Director, Department of Clinical and Experimental Medicine, University of Pisa</i> Chairs: <i>Ferruccio Santini and David Araujo-Vilar</i>
09:20 - 09:40	Nuclear envelope-linked lipodystrophies <i>Giovanna Lattanzi, National Council of Research, Bologna, Italy</i>
09:40 - 10:00	Mechanisms underpinning adipose dysfunction and metabolic disease in generalised lipodystrophy <i>Justin Rochford, University of Aberdeen, UK</i>
10:00 - 10:20	Genome architecture and gene regulation in lipodystrophic laminopathies <i>Philippe Collas, University of Oslo, Norway</i>
10:20 - 10:40	Loss of phospholipase PLAAT3 causes a mixed lipodystrophic and neurological syndrome due to impaired PPARγ signaling <i>Isabelle J\acute{e}ru, Assistance Publique-H\acute{o}pitaux de Paris, Paris, France</i>
10:40 - 11:10	Morning break Chairs: <i>Martin Wabitsch and Baris Akinci</i>
11:10 - 11:30	Molecular basis of acanthosis nigricans in genetic syndromes of severe insulin resistance <i>David Araujo-Vilar, University of Santiago de Compostela, Spain</i>
11:30 - 11:50	Short stature, lipodystrophic syndrome, and cilia defects clinical and cellular findings <i>Corinne Vigouroux, National Institute of Health and Medical Research, Paris, France</i>
11:50 - 12:30	Support for patients <i>Patients Associations: President of AELIP, LD UK, AILIP</i>
12:30 - 14:00	Lunch Chairs: <i>Elif Oral and Justin Rochford</i>
14:00 - 14:15	The adipocyte response to ionizing radiations <i>Margherita Maffei, National Council of Research, Pisa, Italy</i>
14:15 - 14:30	Assessing tissue-specific gene therapies in a pre-clinical mouse model of lipodystrophy <i>George David Mcilroy, University of Aberdeen, UK</i>
14.30 - 14.45	Regulation of leptin secretion in human adipocytes <i>Daniel Tews, Ulm University Medical Center, Germany</i>
14:45 - 15:00	Effects of an acute metreleptin injection on hepatic lipid metabolism in patients with lipodystrophy <i>Marianna Beghini, Medical University of Vienna, Austria</i>
15:00 - 15:15	Age at first clinical signs, age at diagnosis, and age at referral in a reference centre: a study of patients with lipodystrophy and/or severe insulin resistance included in the French national rare disease database <i>Bruno Donadille , St Antoine Hospital, PRISIS Network, Paris, France</i>

15:15 - 15:30	Dunnigan Lipodystrophy in Reunion Island: an exceptional prevalence <i>Estelle Nob\acute{e}court, CHU de LA R\acute{e}union, S Pierre, La R\acute{e}union, France</i>
15:30 - 15:45	Sponsored Oral Presentation: Metreleptin Clinical Studies <i>Laura Maher, Associate Director Clinical Operations Amryt DAC</i>
15:45 - 16:00	Sponsored Oral Presentation: Treatment with mibavademab, a novel leptin receptor agonist antibody, improves metabolic parameters in generalised lipodystrophy <i>Rebecca Brown, National Institute of Health, Bethesda, USA</i>
16:00 - 16:30	Coffee Break and Poster Viewing Chair: <i>David Araujo-Vilar</i>
16:30 - 17:30	ECLip Members Assembly (members only, remaining delegates to explore Pisa)
17:30 - 18:30	ECLip Registry Members Meeting (registry members only, remaining delegates to explore Pisa)
20:30	Conference dinner

AGENDA SEPTEMBER 29th

	Chairs: <i>Corinne Vigouroux, Ferruccio Santini</i>
09:00 - 09:20	LDLync Registry and Lead-ABC study at Michigan: updates from two prospective studies <i>Elif Oral, University of Michigan, USA</i>
09:20 - 09:40	National recommendations for diagnosis and care of Dunnigan syndrome and Congenital Generalized Lipodystrophy, and educational initiatives: recent achievements from the French Lipodystrophy Network <i>Camille Vatier, Hopital Saint Antoine APHP, Paris, France</i>
09:40 - 10:00	Barraquer-Simons Syndrome: our (shared) experience <i>Giovanni Ceccarini, University Hospital of Pisa, Italy</i>
10:00 - 10:20	New human leptin variants <i>Martin Wabitsch, University of Ulm, Germany</i>
10:30 - 11:00	Morning Break Chairs: <i>Martin Wabitsch, Giovanni Ceccarini</i>
11:00 - 11:20	Effects of early vs late metreleptin treatment in patients with generalized lipodystrophy <i>Rebecca Brown, National Institute of Health, Bethesda, USA</i>
11:20 - 11:40	Clinical Factors leading to serious morbidity and mortality in CGL4 <i>Baris Akinci, Dokuz Eylul University, Turkey</i>
11:40 - 12:00	Immunophenotype of patients with perilipin mutations as compared to healthy or obese controls <i>Lucie Stienne, Marie-Christine Vantyghem, Centre Hospitalier Universitaire de Lille, France</i>
12:00 - 12:20	Current Status of the ECLip Registry <i>Julia von Schn\ddot{u}rbein, University of Ulm, Germany</i>
12:20 - 12:30	Lipodystrophy Severity Tool- now ready to use <i>Baris Akinci, Dokuz Eylul University, Turkey</i>
12:30 - 12:40	Closing Remarks
12:40 - 14:10	Lunch Chair: <i>Julia von Schn\ddot{u}rbein</i>
14:30 - 16:30	ECLip Registry Board Members Meeting (registry board members only)