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ECLip Executive Board

CENTRO CONGRESSI LE BENEDETTINE - Piazza S. Paolo a Ripa D'Arno, 16

Under the patronage of:

Società Italiana Endocrinologia

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

Endo-ERN

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**
Ferruccio Santini, Director, Endocrinology Unit, University Hospital of Pisa

Welcome from the Authorities
Riccardo Zucchi, Magnifico Rettore, University of Pisa
Silvia Briani, Director, University Hospital of Pisa
Emanuele Neri, Dean, School of Medicine, University of Pisa
Stefano Taddei, Director, Department of Clinical and Experimental Medicine, University of Pisa

Chairs: *Ferruccio Santini and David Araujo-Vilar*

09:20 - 09:40 **Nuclear envelope-linked lipodystrohies**
Giovanna Lattanzi, National Council of Research, Bologna, Italy

09:40 - 10:00 **Mechanisms underpinning adipose dysfunction and metabolic disease in generalised lipodystrophy**
Justin Rochford, University of Aberdeen, UK

10:00 - 10:20 **Genome architecture and gene regulation in lipodystrophic laminopathies**
Philippe Collas, University of Oslo, Norway

10:20 - 10:40 **Loss of phospholipase PLAAT3 causes a mixed lipodystrophic and neurological syndrome due to impaired PPARγ signaling**
Isabelle Jéru, Assistance Publique-Hôpitaux de Paris, Paris, France

10:40 - 11:10 ***Morning break***

Chairs: *Martin Wabitsch and Baris Akinci*

11:10 - 11:30 **Molecular basis of acanthosis nigricans in genetic syndromes of severe insulin resistance**
David Araujo-Vilar, University of Santiago de Compostela, Spain

11:30 - 11:50 **Short stature, lipodystrophic syndrome, and cilia defects clinical and cellular findings**
Corinne Vigouroux, National Institute of Health and Medical Research, Paris, France

11:50 - 12:30 **Support for patients**
Patients Associations: President of AELIP, LD UK, AILIP

12:30 - 14:00 ***Lunch***

Chairs: *Elif Oral and Justin Rochford*

14:00 - 14:15 **The adipocyte response to ionizing radiations**
Margherita Maffei, National Council of Research, Pisa, Italy

14:15 - 14:30 **Assessing tissue-specific gene therapies in a pre-clinical mouse model of lipodystrophy**
George David Mcilroy, University of Aberdeen, UK

14.30 - 14.45 **Regulation of leptin secretion in human adipocytes**
Daniel Tews, Ulm University Medical Center, Germany

14:45 - 15:00 **Effects of an acute metreleptin injection on hepatic lipid metabolism in patients with lipodystrophy**
Marianna Beghini, Medical University of Vienna, Austria

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**
Bruno Donadille , St Antoine Hospital, PRISIS Network, Paris, France

15:15 - 15:30 **Dunnigan Lipodystrophy in Reunion Island: an exceptional prevalence**
Estelle Nobécourt, CHU de LA Réunion, S Pierre, La Réunion, France

15:30 - 15:45 **Sponsored Oral Presentation: Metreleptin Clinical Studies**
Laura Maher, Associate Director Clinical Operations Amryt DAC

15:45 - 16:00 **Sponsored Oral Presentation: Treatment with mibavademab, a novel leptin receptor agonist antibody, improves metabolic parameters in generalised lipodystrophy**
Rebecca Brown, National Institute of Health, Bethesda, USA

16:00 - 16:30 ***Coffee Break and Poster Viewing***

Chair: *David Araujo-Vilar*

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: *Corinne Vigouroux, Ferruccio Santini*

09:00 - 09:20 **LDLync Registry and Lead-ABC study at Michigan: updates from two prospective studies**
Elif Oral, University of Michigan, USA

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**
Camille Vatier, Hopital Saint Antoine APHP, Paris, France

09:40 - 10:00 **Barraquer-Simons Syndrome: our (shared) experience**
Giovanni Ceccarini, University Hospital of Pisa, Italy

10:00 - 10:20 **New human leptin variants**
Martin Wabitsch, University of Ulm, Germany

10:30 - 11:00 ***Morning Break***

Chairs: *Martin Wabitsch, Giovanni Ceccarini*

11:00 - 11:20 **Effects of early vs late metreleptin treatment in patients with generalized lipodystrophy**
Rebecca Brown, National Institute of Health, Bethesda, USA

11:20 - 11:40 **Clinical Factors leading to serious morbdity and mortality in CGL4**
Baris Akinci, Dokuz Eylul University, Turkey

11:40 - 12:00 **Immunophenotype of patients with perilipin mutations as compared to healthy or obese controls**
Marie-Christine Vantyghem, Centre Hospitalier Universitaire de Lille, France

12:00 - 12:20 **Current Status of the ECLip Registry**
Julia von Schnürbein, University of Ulm, Germany

12:20 - 12:30 **Lipodystrophy Severity Tool- now ready to use**
Baris Akinci, Dokuz Eylul University, Turkey

12:30 - 12:40 **Closing Remarks**

12:40 - 14:10 ***Lunch***

Chair: *Julia von Schnürbein*

14:30 - 16:30 **ECLip Registry Board Members Meeting (registry board members only)**