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cietà Italiana ndocrinologia













# **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	15:15
	Welcome from the Authorities	15:30
	Riccardo Zucchi, Magnifico Rettore, University of Pisa Silvia Briani, Director, University Hospital of Pisa	16:00
	Emanuele Neri, Dean, School of Medicine, University of Pisa Stefano Taddei, Director, Department of Clinical and Experimental Medicine, University of Pisa	16:30 17:30
09:20 - 09:40	<b>Chairs:</b> Ferruccio Santini and David Araujo-Vilar <b>Nuclear envelope-linked lipodystrohies</b> Giovanna Lattanzi, National Council of Research, Bologna, Italy	20:30
09:40 - 10:00	Mechanisms underpinning adipose dysfunction and metabolic disease in generalised lipodystrophy Justin Rochford, University of Aberdeen, UK	AG
10:00 - 10:20	Genome architecture and gene regulation in lipodystrophic laminopathies Philippe Collas, University of Oslo, Norway	09:00
10:20 - 10:40	Loss of phospholipase PLAAT3 causes a mixed lipodystrophic and neurological syndrome due to impaired PPARy signaling Isabelle Jéru, Assistance Publique-Hôpitaux de Paris, Paris, France	09:20
10:40 - 11:10	Morning break	
11:10 - 11:30	Chairs: Martin Wabitsch and Baris Akinci Molecular basis of acanthosis nigricans in genetic syndromes of severe insulin resistance David Araujo-Vilar, University of Santiago de Compostela, Spain	09:40
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	10:00
11:50 - 12:30	Support for patients Patients Associations: AELIP, LD UK, AILIP	11:00
12:30 - 14:00	Lunch	
14:00 - 14:15	<b>Chairs:</b> Elif Oral and Justin Rochford <b>The adipocyte response to ionizing radiations</b> Margherita Maffei, National Council of Research, Pisa, Italy	11:20
14:15 - 14:30	Assessing tissue-specific gene therapies in a pre-clinical mouse model of lipodystrophy George David Mcilroy, University of Aberdeen, UK	11:40
14.30 - 14.45	Regulation of leptin secretion in human adipocytes Daniel Tews, Ulm University Medical Center, Germany	12:00
14:45 - 15:00	Age at first clinical signs, age at diagnosis, and age at referral in a reference	12:20
	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	12:30 14:30

15:00 - 15:15	Effects of an acute metreleptin injection on hepatic lipid metabolism in patients with lipodystrophy Marianna Beghini, Medical University of Vienna, Austria	
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 - 16:00	Sponsored Oral Presentations	
16:00 - 16:30	Coffee Break and Poster Viewing	
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 29 <sup>th</sup>	
09:00 - 09:20	<b>Chair:</b> Corinne Vigouroux <b>LDLync Registry and Lead-ABC study at Michigan: updates from</b> <b>two prospective studies</b> <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 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	<b>New human leptin variants</b> Martin Wabitsch, University of Ulm, Germany	
10:30 - 11:00	Morning Break	
11:00 - 11:20	Chair: Giovanni Ceccarini Effects of early vs late metreleptin treatment in patients with generalized lipodystrophy Rebecca Brown, National Institutes of Health, Bethesda, USA	
11:20 - 11:40	<b>Clinical Factors leading to serious morbdity and mortality in CGL4</b> <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 Marie-Christine Vantyghem, Centre Hospitalier Universitaire de Lille, France	
12:00 - 12:20	<b>Current Status of the ECLip Registry</b> Julia von Schnürbein, University of Ulm, Germany	
12:20 - 12:30	Closing Remarks	
12:30 - 14:00	Lunch	
14:30 - 16:30	ECLip Registry Board Members Meeting (registry board members only)	