

# ECLip 2019 Meeting

## VII Symposium of Lipodystrophies

Venue:  
CREER centre  
Burgos (Spain)

4-5th October

European Consortium of Lipodystrophies



RARE  
DISEASES  
INTERNATIONAL  
A EURORDIS INITIATIVE

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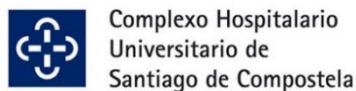


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# ECLip 2019 Meeting – Final Programme

## OCTOBER 4th, SCIENTIFIC SESSIONS

### 09:00 Welcome

Prof. David Araújo Vilar, University of Santiago de Compostela, Spain  
Naca Pérez de Tudela, President of AELIP, Spain  
Juan Carrión, President of Spanish Federation of Rare Diseases  
Aitor Aparicio García, Director of CREER

### 09:20 Epigenetic mechanisms in FPLD2

Prof. Philippe Collas, University of Oslo, Norway

### 09:40 Human SGBS cells as in vitro model for studying cellular phenotype of defects in lipodystrophy genes

Prof. Martin Wabitsch, University of Ulm, Germany

### 10:00 Differential expression of brown and white adipose tissue markers in FPLD2 adipocytes

Prof. Giovanna Lattanzi, CNR-Unit of Bologna, Italy

### 10:20 Novel understandings in AGL unlock mysteries in the interaction between immune system and metabolic regulation

Prof. Elif A. Oral, University of Michigan, USA

### 10:40 Detection and characterization of autoantibodies against perilipin 1 in patients with acquired generalized lipodystrophy

Dr. Fernando Corvillo, La Paz University Hospital, Madrid, Spain

### 11:00 Coffee break

### 11:30 Molecular underpinnings of generalised lipodystrophies: studies of seipin and AGPAT2 function

Prof. Justin Rochford, University of Aberdeen, UK

### 11:50 The mouse *BSCL2*<sup>Celia/Celia</sup>: brain, liver and fat phenotype

Dr. Sofía Sánchez Iglesias, University of Santiago de Compostela, Spain

### 12:10 Leptin restores brown adipose tissue thermogenesis in lipodystrophy potentially through improving bile acid homeostasis

Dr. Susan Kralisch-Jäcklein, University of Leipzig, Leipzig, Germany

### 12:30 How is lipodystrophy linked to cardiometabolic disease: insights from SHORT syndrome

Prof. Robert Semple, University of Edinburgh, UK

### 12:50 Lipodystrophy epidemiology in Spain: the experience of a nationwide reference centre

Dr. Antía Fernández Pombo, University of Santiago de Compostela, Spain

### 13:10 The importance of European networking on lipodystrophy: the Hungarian experience

Dr. Éva Csajbók, University of Szeged, Hungary

### 13:30 Metreleptin in Familial Partial Lipodystrophy: the Greek experience

Prof. Vaia Lambadiari, Athens University Medical School, Greece

### 14:00 Lunch

## OCTOBER 4th, SCIENTIFIC SESSIONS

**15:30 Diagnostic challenge in FPLD4**

Prof. Marie Christine Vantyghem, University of Lille, France / Dr. Isabelle Jéru, Sorbonne University, Paris, France

**15:50 Variable phenotype of *LMNA*-associated lipodystrophy**

Prof. Hartmut H-J. Schmidt, University of Münster, Germany

**16:10 Cardiovascular phenotypes in *LMNA*-linked lipodystrophy: implications for clinical practice**

Prof. Corinne Vigouroux, Sorbonne University, Paris, France

**16:30 Generalized lipodystrophy associated with delayed neuro-somatic development and multiple dysmorphism in a neonate with a compound heterozygous missense mutation in the *SYNE2* gene**

Prof. Ferruccio Santini, University of Pisa, Italy

**17:00 Coffee break**

**17:20 Unsolved case with suspected lipohypotrophic syndrome**

Dr. Deimante Brazdziunaite, Vilnius University, Lithuania

**17:40 Deep phenotyping reveals unusual presentations and emerging clinical phenotypes of lipodystrophy: lessons from observational registries and the LDLync study**

Dr. Baris Akinci, Dokuz Eylul University, Izmir, Turkey / University of Michigan, USA

**18:00 ECLip Lipodystrophy Registry**

*For Registry board members only*

**21:00 Speakers' dinner**

## OCTOBER 5th, SCIENTIFIC SESSIONS

**09:00** **ECLip Lipodystrophy Registry: ongoing and future projects** *(for Registry members only)*

Prof. Martin Wabitsch / Dr. Julia von Schnürbein, University of Ulm, Germany

**09:40** **ECLip Lipodystrophy Registry: Presentation and first results**

Prof. Martin Wabitsch / Dr. Julia von Schnürbein, University of Ulm, Germany

**10:00** **LipoDDX®: a mobile application for identification of rare lipodystrophy syndromes**

Prof. David Araújo Vilar, University of Santiago de Compostela, Spain

**10:20** **Diet therapy in patients with lipodystrophy syndromes: perspectives and challenges**

Dr. Yulia Tikhonovich, Endocrinology Research Centre, Moscow, Russia

**10:40** **Leptin improves insulin sensitivity independent of food intake in patients with lipodystrophy**

Dr. Rebecca J. Brown, National Institutes of Health, Bethesda, USA

**11:00** **Coffee break**

**11:20** **Recent trials targeting hepatokines: what have we learned?**

Prof. Elif A. Oral, University of Michigan, USA

**11:40** **How patient organizations can partner with lipodystrophy stakeholders in all phases of research and development**

Andra Stratton, co-Founder & President, Lipodystrophy United, USA

**12:00** **The German Lipodystrophy advocacy group**

Sabine Tilp, German Lipodystrophy Advocacy Group

**12:20** **Final conclusions**

Prof. David Araújo Vilar, University of Santiago de Compostela, Spain

**12:30** **ECLip assembly** *(for ECLip members only)*

Legal framework, Governing board election, ongoing and future projects

**14:30** **Lunch**

# VII Symposium of Lipodystrophies (Patients & Relatives) - Final Programme

## OCTOBER 4th

**17:30** Working meeting among representatives of international patient associations and delegations

## OCTOBER 5th

**13:00** Coordination Meeting for 2020 Lipodystrophy Day

Representatives of patient associations and delegations

**14:30** Lunch

**15:30** Welcome to patients, relatives and entities working in the field of Lipodystrophies

Naca Eulalia Pérez de Tudela Cánovas, President of AELIP, Spain

**15:45** Mutual help groups, support networks to face the day to day

Manuel Illera Hernández, Psychologist of AELIP, Spain

**16:15** International cooperation project in Peru, International Network on Lipodystrophies

Juan Carrión, President of the Spanish Federation of Rare Diseases, Spain

**16:45** Immunological aspects of lipodystrophies

Prof. Margarita López Trascasa, Autonomous University of Madrid, IdiPAZ, Madrid, Spain

**17:15** Basic knowledges for laity people to understand genetic diseases

Prof. David Araújo Vilar, University of Santiago de Compostela, Spain

**17:45** Coffee break

**18:00** Nutritional workshop: Diet and Lipodystrophies

Dr. María González Rodríguez, University Hospital Complex of Santiago de Compostela, Spain

**18:45** Self-care and personal image workshop

Maite Embid, Unidad de Maquillaje Terapéutico del Hospital Ramón y Cajal, Madrid, Spain

**19:30** Closure of the VII International Symposium of Lipodystrophies and group photo

Naca Eulalia Pérez de Tudela Cánovas, President of AELIP, Spain

Save the date!

# *ECLip*2020

VIII Symposium of Lipodystrophies

*Clare College, Cambridge, UK*

*15<sup>th</sup>/16<sup>th</sup> October 2020*

