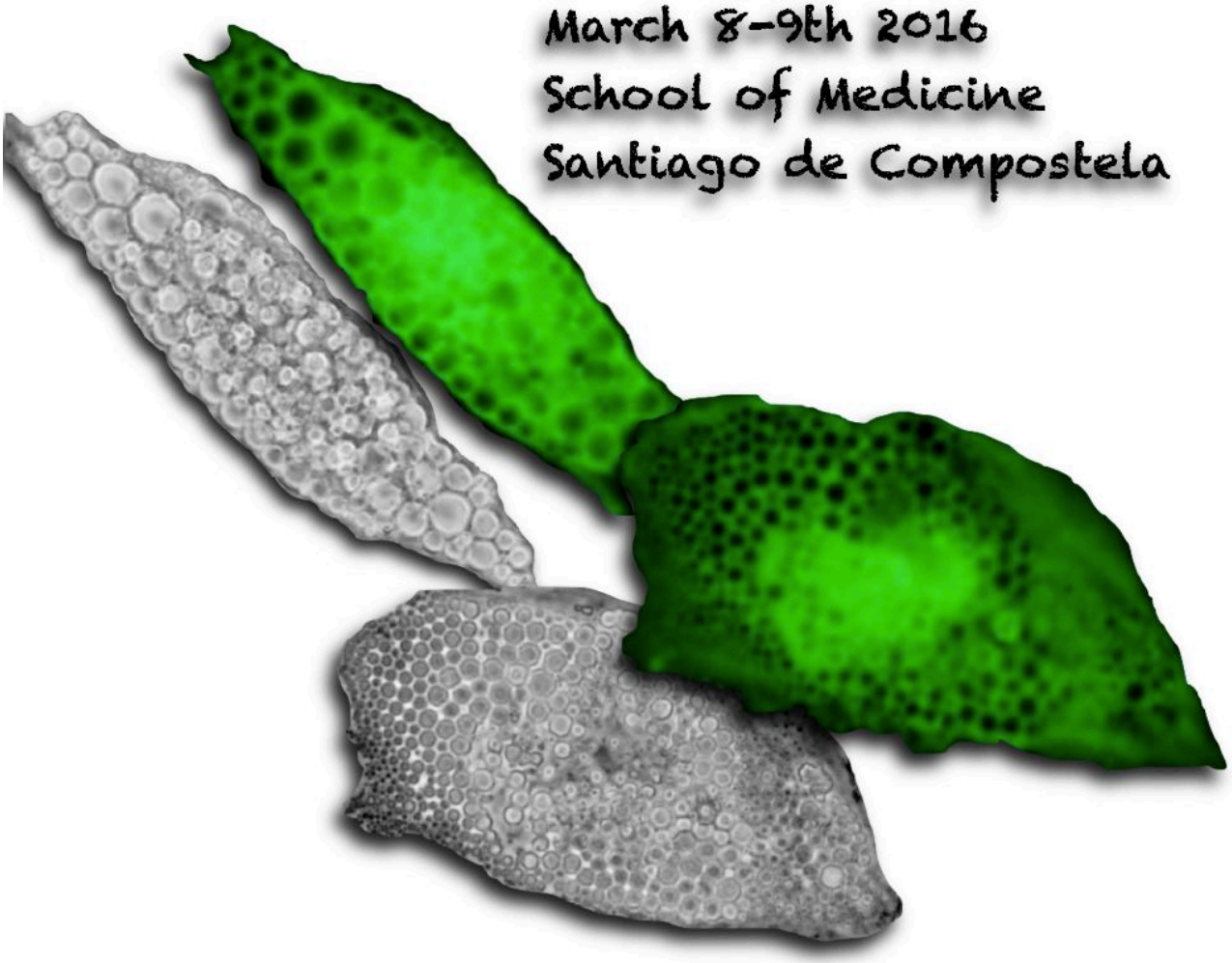
2016
ECLip Annual Meeting

March 8-9th 2016
School of Medicine
Santiago de Compostela



March 31st, World Lipodystrophy Day

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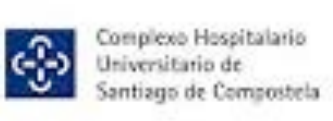


Center for Research in Molecular Medicine and Chronic Diseases

March 31st, World Lipodystrophy Day

2016 ECLip Annual Meeting
March 8-9th 2016
Santiago de Compostela

Final Program

March 8th

08:50 Welcome
Prof. David Araújo-Vilar, University of Santiago de Compostela, Spain

09:00 Opening Lecture: *Adipose tissue, foe or friend?*
Prof. Felipe F. Casanueva, University of Santiago de Compostela, Spain

09:30 Characteristic and atypical mutations in lipodystrophy patients
Prof. Katrin Hoffman, Martin Luther University Halle-Wittenberg, Germany

10:00 Central obesity, diabetes mellitus and hypertriglyceridemia: Understanding the Köbberling syndrome
Dr. Cristina Guillín-Amarelle, University of Santiago de Compostela, Spain

10:30 Fontaine-Petty syndrome, a rare cause of lipodystrophy, demonstrates a new pathogenesis for lipodystrophies
Prof. Raoul CM Hennekam, Academic Medical Center, University of Amsterdam, Netherlands

11:00 Coffee break

11:30 Progressive lipodystrophy associated with mutation in the *POLD1* gene
Prof Giuseppe Novelli, Universita Tor Vergata, Roma, Italy

12:00 Clinical reports of fat loss associated with disorders of the immune system: an opportunity for a further understanding of the pathogenesis of lipodystrophy
Prof. Ferruccio Santini/Dr Yaroslav Berger, University Hospital of Pisa, Italy

12:30 Decrease of osteocalcinemia linked with insulin resistance more than body fat mass in lipodystrophic syndromes, compared to obese and normal weight patients
Prof. Marie-Christine Vantyghem, Lille University Hospital, Lille, France

13:00 Celia's Encephalopathy (PELD): seipin, adipose tissue and beyond
Dr. Sofía Sánchez-Iglesias, University of Santiago de Compostela, Spain

13:30 Lunch

15:00 Young Researchers Session

15:00 Clinical, hormonal and molecular-genetic characteristics of inherited lipodystrophies in Russia
Dr Ekaterina Sorkina, Endocrinology Research Center, Moscow, Russia

15:20 Human induced pluripotent stem cells as a cellular tool for modelling laminopathies
Dr. Anne-Claire Guénantin, Centre de Recherche Saint-Antoine, Paris, France

15:40 Morpho-functional study of subcutaneous adipose tissue in patients affected by Familial Partial Lipodystrophy type 2-FPLD2
Dr Alessandra Gambineri, S. Orsola-Malpighi Hospital, Bologna, Italy

16:00 Coffee break

16:30 ECLip Databases (*for ECLip members only*)
Prof. Giovanna Lattanzi and Prof David Araújo-Vilar

March 9th

09:00 Lessons from *LMNA*-lipodystrophy in vitro models based on human mesenchymal stem cells that accumulate prelamin A
Dr Clara Isabel Rodríguez-López, BioCruces Health Research Institute, Spain

09:30 Modulation of brown adipose tissue differentiation in FPLD2
Prof. Giovanna Lattanzi, CNR, Unit of Bologna, Italy

10:00 Human SGBS cells as in vitro model for studying cellular phenotype of defects in lipodystrophy genes
Prof. Martin Wabitsch, University Medical Center Ulm, Germany

10:30 Maladaptative autophagy contributes to insulin resistance and altered adipocyte differentiation in Congenital Generalized Lipodystrophy due to PTRF/cavin-1 mutations.
Dr. Corinne Vigouroux, Centre de Recherche Saint-Antoine, Paris, France

11:00 Application of proteomics in lipodystrophy research
Prof. Juan Ramón Peinado-Mena, University of Castilla-La Mancha, Spain

11:30 Coffee break

12:00 Outcome of intensive dietary intervention in patients with lipodystrophy
Dr. Anne Stears, University of Cambridge, UK

12:30 Effects of metreleptin therapy on adipose tissue, liver and and systemic inflammation markers in lipodystrophic patients
Dr. Camille Vatier, Centre de Recherche Saint-Antoine, Paris, France

13:00 Antisense strategy in familial partial lipodystrophy
Prof. Harmut Schmidt, Universitätsklinikum Münster, Germany

13:30 MG132 enhances progerin clearance and reverses cellular phenotypes in Hutchinson-Gilford progeria cells.
Dr. Karim Harhouri, Faculty of Medicine La Timone, Marseille, France

14:00 Lunch

15:30 How advocacy groups can support research: the Spanish experience
Mr. Juan Carrión, President of the Spanish Federation of Rare Diseases

15:50 AELIP: an example of advocacy group supporting lipodystrophy patients and relatives
Ms. Naca Pérez de Tudela, President of AELIP

16:10 Coffee break

16:45 Closing lecture: Inflammation in progeroid laminopathies
Prof. José M. P. Freije, Universidad de Oviedo, Spain

Venue:

Aula Castelao (2nd floor)
Facultade de Medicina
Rua de San Francisco s/n
15782 Santiago de Compostela, Spain
Free registry

Contact mail: lipodistrofias@gmail.com

SPEAKERS BIOSKETCH

Dr. Yaroslav Berger

Yaroslav Berger, Ph.D., works as a postdoctoral fellow in the Department of Metabolism and Endocrinology in the University of Pisa after he DTI-IMPORT International Postdoctoral Program of the Dulbecco Telethon Institute. He obtained his PhD in 2012 at the Faculty of Medicine in the Technion - Israel institute of Technology in Haifa. His thesis was to establish the role of Endothelial Progenitor Cells in Cardiovascular Disease and Sleep Apnea. After the graduation, he continued working as a postdoc fellow at the same faculty until October 2014, when he joined the Dr. Maffei group in Pisa to find a link between the mutations in DNA polymerase delta and loss of adipose tissue in MDPL syndrome. In addition, during the last year, he joined a novel project in the lab to investigate the potential changes in regulation of the immune system in lipodystrophy.

Mr. Juan Carrión

Juan Carrión is the father of his little princess Celia. He is social worker, and coordinator of Social Work Department of Penitentiary Murcia I (Interior Ministry). He is member of the Advisory Committee of Rare Diseases in the Spanish Ministry of Health, President of the association of rare diseases D'GENES, President of the Spanish Federation of Rare Diseases (FEDER), President of the Foundation for Research on Rare Diseases (FEDER FOUNDATION), President of the Latin American Alliance for Rare Diseases (ALIBER), board member and technical coordinator of the association of families and affected of lipodystrophy (AELIP), and technical coordinator of the Multidisciplinary Center for Comprehensive Care to individuals and families with rare diseases Celia Carrión Pérez de Tudela Canovas.

Prof. Felipe F. Casanueva

Professor Felipe F Casanueva is currently Full Professor of Medicine and Head of the Endocrine Division at the Department of Medicine, University Hospital (CHUS), Santiago de Compostela University.
He is currently the President of the Spanish Society for Obesity (SEEDO) and Scientific Director of the Center for Biomedical Research on Obesity and Nutrition (CIBERobn), period 2006-2015.
Holds three Honorary Doctorates, of the University of Lodz, Poland; from the University of Caiseri, Turkey; and also from the University of Belgrade, Serbia. Is European Hormone Medal of European Society for Endocrinology's and full member of the Real Academia de Medicina y Cirugia de Galicia (RAMYCGA), and in the past was President of the Spanish Endocrine Society (SEEN); the European Federation of Endocrine Societies (Currently SEE), the International Society for Endocrinology (ISE) and The Pituitary Society.
Dr Casanueva has published more of 600 papers in peer review international journals with a cumulative Impact Factor of 1708,72. Has written 48 chapters on international books and textbooks and delivered 137 lectures by invitation at International Congresses. His current H-index is 75.
He has received several awards and acknowledgements, among them, the Xunta de Galicia Award for research (two times), The Spanish Society of Endocrinology highest award, The Rey Jaime I award, Danone, and the International Geoffrey Harris Award in Neuroendocrinology.
Member of several International Boards of learned journal and Continental Editor of Clinical Endocrinology until 2015.

Prof. José M.P. Freije

José M.P. Freije is an Associate Professor of Biochemistry and Molecular Biology at the University of Oviedo, Spain. After his predoctoral research, his work was aimed at studying the molecular mechanisms of cancer invasion and metastasis. This work led to the description of several new matrix metalloproteases, the biochemical characterization of metastasis suppressor proteins and the exploration of their utility to identify anti-cancer compounds preferentially active against the most aggressive tumor cells. The research work carried out by José M.P. Freije over the last years has approached different aspects of cancer and aging, using animal models as an essential tool to investigate the molecular mechanisms underlying these processes and to explore therapeutic strategies against the pathologic alterations that they involve.

Dr. Alessandra Gambineri

Alessandra Gambineri received her specialty trainings in Endocrinology at Bologna University in 2002 and reached her PhD in Clinical and Experimental Medical Science at the University of Verona in 2009. Since 2010 she has a position as Assistant Professor at the University of Bologna. She has conducted several clinical trials as Principal Investigator related to diabetes mellitus and lipodystrophies. She is interested in female hyperandrogenic disorders, female hypogonadism and severe insulin resistance states, steroids and metabolism, and she has published more than 75 full papers.

Dr. Anne-Claire Guénantin

Anne-Claire Guénantin, PhD, works as a post-doctoral researcher in cellular and molecular biology at Saint-Antoine Research Center (INSERM UMR_S938) and Medical University Pierre and Marie Curie (Paris 6), France, in the field of the pathophysiology of stem cells and laminopathies. After a PhD regarding cardiomyocyte differentiation of pluripotent stem cells, she joined the team of Corinne Vigouroux in 2012 to develop her expertise in endothelial and adipocyte differentiation of induced pluripotent stem cells (iPSC).

Dr. Cristina Guillín-Amarelle

Cristina Guillín-Amarelle studied Medicine at the University of Santiago de Compostela, and between the years 2009 and 2013 specialized in Endocrinology and Nutrition. At the present time, Dra Guillín works as an Endocrinologist at the Hospital of Santiago de Compostela, and is performing her predoctoral studies on the lipodystrophies topic, under the supervision of Prof. Araújo Vilar.

Dr. Karim Harhouri

Karim Harhouri is a postdoctoral fellow at Nicolas Levy's laboratory in the School of Medicine of Aix-Marseille University. His principal research interest is the molecular basis of Hutchinson-Gilford progeria, particularly he is trying to unveil the putative clearance pathways of progerin as well as potential new therapies for children with this syndrome.

Prof. Raoul Hennekam

Raoul Hennekam received his specialty trainings in Paediatrics and in Clinical Genetics at Utrecht University. He was appointed as professor of Paediatrics and Clinical Genetics in 2002 at the AMC of University of Amsterdam. During 2005-2010 he worked in London at Institute of Child Health and Great Ormond Street Hospital as professor of Clinical Genetics and Dysmorphology. He is presently working as Professor of Paediatrics and Translational Genetics in Amsterdam.

Main scientific interests include intellectual disabilities, autism, connective tissue disorders, and (molecular) dysmorphology. He is member of the Dutch Health Council, EUCERD, European Research Council, Editor of American Journal of Medical Genetics and of the European Journal of Medical Genetics, author of >450 papers in peer-reviewed journals (H-index 64) and 23 chapters in international texts, co-chair of the international Morphology Nomenclature Committee, and senior editor of 'Gorlin's Syndromes of the Head and Neck'.

Prof. Katrin Hoffmann

Martin Luther University Halle-Wittenberg, Germany

Prof. Giovanna Lattanzi

Dr. Giovanna Lattanzi is a Researcher at the National Research Council (CNR) Institute for Molecular Genetics (IGM), Bologna, Italy. She is director of the Bologna Unit of IGM. Dr. Lattanzi and her research team are elucidating the cell biology of lamins, emerin and lamin-linked proteins under normal or pathological conditions. Her first study on lipodystrophy has demonstrated the involvement of prelamin A in disease pathogenesis. Further studies have deepened prelamin A interplay with nuclear partners and chromatin and shown prelamin A involvement in genome maintenance during normal ageing. More recent papers from Dr. Lattanzi lab have reported on the efficacy of drug treatments based on the use of rapamycin alone or in combination with retinoic acid for the recovery of Mandibuloacral dysplasia and progeria cellular phenotype. Dr. Lattanzi has been partner of the Euro-laminopathies project. She is the coordinator of the Italian Network for Laminopathies aimed at connecting scientists, clinicians and families to improve quality of research and patient care.

Prof. Giuseppe Novelli

Giuseppe Novelli Is Head of the Human Genetics Research Unit at The Tor Vergata University of Rome (Italy). He served Tor Vergata University as Dean of the School of Medicine during the period 2008-2011. Professor Novelli is at present Rector of the University of Rome Tor Vergata (2013-2019). He is member of the Pharmacogenetics Working Party of the Committee for Human Medicinal Products (CHMP) at the EMA (European Medicines Agency) in London. He was Board Member of the Italian National Agency for Evaluation of Universities and Research Institutes (ANVUR) (2010-2013). He published over 400 original scientific publications including invited reviews for leader Journals in Human Genetics. The H-index is 53. Primary focus of G. Novelli was the mapping, identification and characterization of human-disease genes (e.g. Laron dwarfism, DiGeorge syndrome, Mandibuloacral dysplasia, Friedrich ataxia vitamin-E-deficiency, spinal muscular atrophy, hypoplastic glomerulocystic kidney disease, myotonic dystrophy). Recently he is actively involved in the field of complex diseases and pharmacogenetics.

Prof. Juan Ramón Peinado-Mena

Dr. Juan R. Peinado is Associate Professor of Cell Biology and Histology in the Faculty of Medicine of Ciudad Real, Spain. After his predoctoral research he made stays in several international laboratories, such as the Animal Physiology Department at the University of Nijmejen, Netherlands; the Department of Neuroendocrinology at INSERM, Rouen, France. He worked two years as Postdoc in the Department Of Biochemistry of the Louisiana Health Science Center (LSUHSC). He has also developed his research in Spanish centers of excellence such as proteolysis Laboratory located in the Scientific Park of Barcelona, belonging to CSIC; the Dept. of Biochemistry and Molecular Biology, University of Oviedo and the Department of Cell Biology, Physiology and Immunology at the University of Cordoba. His recent research focus on proteomic studies to face diseases that directly affect adipose tissue (obesity and / or lipodystrophy). He is also involved in proteomic studies on neurodegenerative disorders (Alzheimer), and brain tumor (glioblastoma).

Ms. Naca Pérez de Tudela

Naca Pérez de Tudela is the mom of Celia Carrión Pérez de Tudela Canovas. She is Thermomix commercial agent. She was founder and president of the association of rare diseases D'GENES (2008-2012). She is President of the association of families and affected of lipodystrophy (AELIP), and responsible for maintaining the Multidisciplinary Center for comprehensive care to individuals and families with rare diseases Celia Carrión Pérez de Tudela Canovas.

Dr. Clara I. Rodriguez-López

Clara I. Rodriguez obtained her Ph.D. in 1999 at the Universidad Autonoma de Madrid and the Centro de Biologia Molecular Severo Ochoa under the supervision of Prof. Manuel Fresno Escudero, working on parasite activation of the immune system. She next did a short one year postdoc in the same center in Dr. Maria Luisa Salas' Lab specializing on viral signaling. In 2000 she moved to the National Cancer Institute at Frederick (NIH; USA) for postdoctoral training in genetic manipulation in mammals. It was during those five years at the NCI where she was introduced and gained experience working on pluripotent stem cells. Dr. Rodriguez returned to Spain as researcher at the Valencia Stem Cell Bank (Centro de Investigacion Principe Felipe). In 2006 she moved to the Hospital de Cruces (Bilbao) where she was awarded an investigator contract under the Miguel Servet program to start a new research group focused on stem cell based therapy. Currently, she is Group Leader of the Stem Cells and Cell therapy Laboratory at the BioCruces Health Research Institute, interested in the potential of human

stem cells for the study of human disease and the design of new therapies. She is the Principal Investigator of a national multicenter clinical trial of cell therapy applied to pediatric patients suffering from Osteogenesis Imperfecta.

Dr. Sofía Sánchez-Iglesias

Sofía Sánchez-Iglesias is a gardener from the heart and researcher by choice. She was born in Geneva (Switzerland) where she earned her Bachelor's degree in Biochemistry. In 2010, she received her Ph.D. at the University of Santiago de Compostela (Spain). Her predoctoral research was focused on the understanding of the molecular mechanisms involved in the pathogenesis of Parkinson's disease and the development of new pharmacological strategies for treatment and prevention. The incorporation in 2011 within the group of Prof. David Araújo-Vilar was a major turnaround that allow her to gain expertise in a broad variety of experimental techniques. The new lines of research were based on the molecular basis and clinical characterisation of familial lipodystrophies, and the recently described neurodegenerative disease associated with the c.985C>T mutation in seipin. Fruits of her research were 16 publications in internationally recognised journals. During her career she received various grants and also participated in several research projects.

Prof. Ferruccio Santini

Ferruccio Santini is Associate Professor of Endocrinology at the University of Pisa and Head of the Obesity Center at the Endocrinology Unit, University Hospital of Pisa. He obtained his Medical Degree at the University of Pisa in 1985 and his PhD in Endocrinological and Metabolic Sciences in 1996. Main scientific interest include the pathogenesis and physiopathology of obesity, lipodystrophies and related diseases, thyroid physiology and thyroid diseases. His research has been published in more than 110 articles quoted in Pubmed. He is member of the Endocrine Society, European Society of Endocrinology, Italian Society of Endocrinology, Italian Society of Obesity, Italian Society of Obesity Surgery, Italian Thyroid Association.

Prof. Harmut Schmidt

Universitätsklinikum Münster, Germany

Dr Ekaterina Sorkina

Ekaterina Sorkina is an endocrinologist and research assistant at the Clamp-technologies laboratory, Institution of Diabetes, Endocrinology Research Center in Moscow, Russia
She is also PhD fellow in Endocrinology, I.M. Sechenov First Moscow State Medical University, and scientific research in inherited lipodystrophies, being her scientific advisor Anatoly Tiulpakov, MD, PhD, paediatrician-endocrinologist, Chief of the Department and Laboratory of Inherited Endocrine Diseases in Endocrinology Research Center. She is a member of Moscow State Association of Endocrinologists, Russian Association of Endocrinologists, ESE, ENEA, ENDO, DGE, ADA, and the ambassador of EYES (European Young Endocrine Scientists) in Russia. Her scientific interest covers endocrinology and genetics, anti-age medicine, different hormonal and metabolic disorders, lipodystrophies (inherited and acquired), laminopathies, inherited forms of diabetes mellitus and insulin resistance. She is an author of 20 publications, 3 of them quoted on pubmed.

Dr Anna Stears

Anna Stears is a Consultant in Diabetes and Endocrinology at Addenbrooke's Hospital, Cambridge, UK. She has responsibility for the day to day running and development of the National Severe Insulin Resistance Service, a national multidisciplinary service for adults and children with Severe Insulin Resistance and/or lipodystrophy. This highly specialised service is funded directly by NHS England. It is the only UK centre currently permitted to treat patients with lipodystrophy with leptin therapy. Anna is supported by colleagues in the Institute of Metabolic Science who have had a long standing clinical and research interest in the pathophysiology and treatment of syndromes of Severe Insulin Resistance and by a multidisciplinary team including Consultants in Paediatric Diabetes and Endocrinology, specialist nurses, dieticians and administrators

Dr. Camille Vatier

Camille Vatier is a consultant physician in Endocrinology and Nutrition at Assistance-Publique Hôpitaux de Paris, Saint-Antoine hospital, France. Her clinical activity focus comprises diabetes, and abnormal fat tissue distribution.

She joined the team of Corinne Vigouroux at Saint-Antoine Research Center (INSERM UMRS_938) in 2008 to develop her clinical expertise and experimental research in lipodystrophic syndromes and she is PhD fellow at University Pierre and Marie Curie (Paris 6) .

Since 2015, she has a teaching position in cellular biology at medical University Pierre and Marie Curie (Paris 6).

Prof. Marie Christine Vantyghem

Marie-Christine Vantyghem, MD, PhD, received his specialty trainings in Endocrinology, Diabetology and Metabolism at the University of Lille, France. She has been involved in the islet transplantation program at the University Hospital of Edmonton, Alberta, Canada on the clinical side with Prof Ryan and at the Pacific Northwest Research Institute at Seattle, USA with Prof Robertson. She was appointed professor of Endocrinology in 2011 and is head of the Endocrinology Department of Lille University Hospital, which cares for a population of more than 4 millions people. In 2009, she has obtained an interface contract with the INSERM Unit U1190, devoted to «Translational Research in Diabetes".

Her main research areas are islet transplantation and lipodystrophies with a large cohort of more than a hundred patients investigated. A biobank (plasmatheque, DNAtheque and adipose tissue bank) has been organised under the name project PHRC IL7 lipodystrophies.

She is member of the executive board of the French Endocrine Society, member of the European Society of Endocrinology, of the Endocrine Society, of the French and European Societies of Transplantation and has been or is involved in two FP7 programs and in the CITR registry. She works as an Associate editor for the Annals of Endocrinology.

Prof. Corinne Vigouroux

Corinne Vigouroux, MD, PhD, develops her medical and research activities in the field of the pathophysiology of insulin resistance and lipodystrophic syndromes.

She works at Assistance-Publique Hôpitaux de Paris, Saint-Antoine hospital, France, as an hospital endocrinologist and molecular biologist.

She has a teaching and research senior position in cellular and molecular biology at Saint-Antoine Research Center (INSERM UMR_S938) and Medical University Pierre and Marie Curie (Paris 6).

Prof. Martin Wabitsch

Martin Wabitsch MD, PhD is Professor of Pediatrics at the University of Ulm, Germany and is the Head of the Division of Pediatric Endocrinology and Diabetes at the Department of Pediatrics and Adolescents Medicine in Ulm's University Hospital (www.ped-u.de). His clinical research focus comprises childhood obesity, the physiology of body weight regulation in children, leptin deficiency in humans, and rare lipodystrophy syndromes. His experimental research focus is the biology of the human adipocyte. He and his group have established in vitro models for studying the biology of the human white and brown adipocyte including the human SGBS cell strain.
Dr. Wabitsch has had his clinical and experimental training at the Universities in Berlin (FU), Ulm, Baltimore (Johns Hopkins) and Nice (Sophia Antipolis). Within an ESPE research fellowship he performed studies on the growth hormone receptor at the Hagedorn Research Institute. He has won several research prices of scientific societies, among them the Young Investigator Award of the (ESPE). Dr. Wabitsch has been the President of

the German Society for Pediatric Endocrinology and Diabetes from 2008-2012. Dr. Wabitsch has published more than 200 peer reviewed articles.

Prof. David Araújo-Vilar

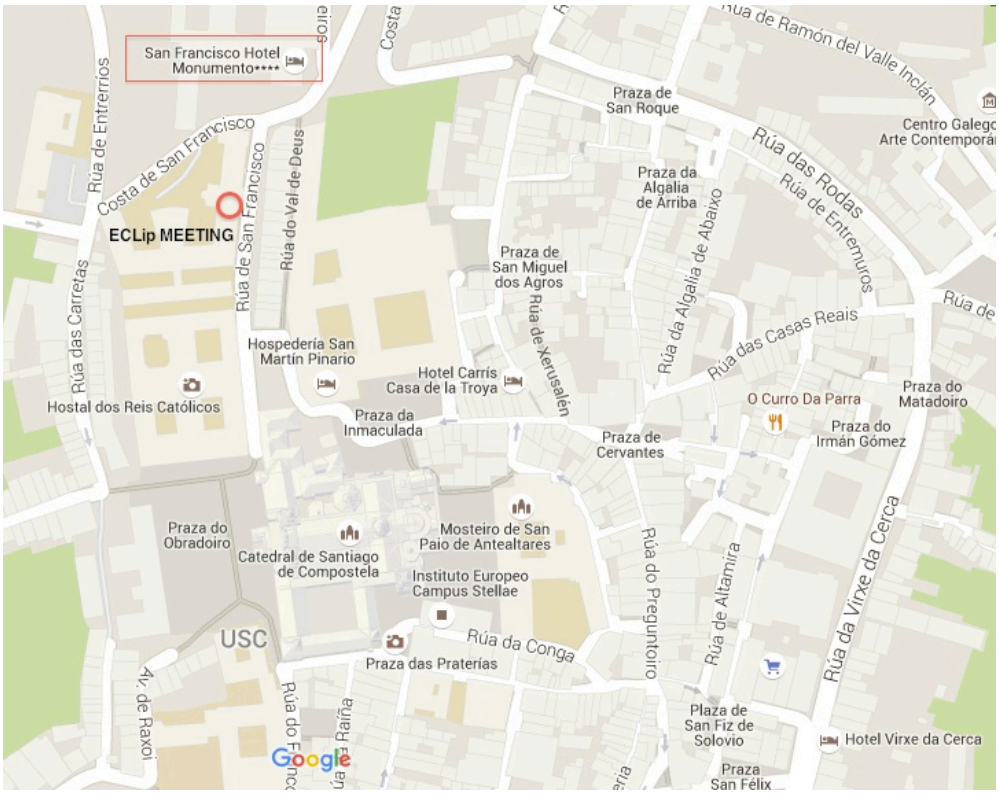
David Araújo-Vilar is a consultant physician in Endocrinology and Nutrition at the University Clinical Hospital of Santiago de Compostela and Associated Professor of Medical Genetics and Endocrinology at the School of Medicine of Santiago. Clinical and research postgraduate training was in Oxford University and in Santiago de Compostela General Hospital, including a PhD under the supervision of Prof. Cabezas-Cerrato investigating the mathematical modelling of glucose metabolism in diabetes mellitus and obesity. Over the past 10 years his clinical and research interests have centred on the genetic basis of rare lipodystrophic syndromes and severe insulin resistance syndromes. Lastly his research has particularly focused in Celia's encephalopathy and in the role of seipin in adipogenesis and neurodegeneration. He is the president of the Spanish Lipodystrophy Society and member of the Executive Board of the European Consortium of Lipodystrophies.

ACCOMMODATION

SAN FRANCISCO HOTEL MONUMENTO
www.sanfranciscohm.com
3 min walking
Price: 100-120€ (per night)

GRAN HOTEL SANTIAGO
www.hotelghsantiago.com
30 min walking
Price: 110€ (per night)

HOTEL AIRAS NUNES
HOTEL SAN CLEMENTE
5-7 min walking
Price: <90€ (per night)
www.pousadasdecompostela.com/hoteles-santiago-y-galicia



TRAVEL to city centre

Lavacolla-Santiago de Compostela Airport
By taxi: 21€ (http://www.aena.es/csee/Satellite/Aeropuerto-Santiago/es/Page/1237554869572//Taxi.html)

By bus: 3€ (http://www.aena.es/csee/Satellite/Aeropuerto-Santiago/es/Page/1237554501825//Transporte-publico.html)

WEATHER
Santiago de Compostela weather is mild (12-20°C) and rainy. Umbrella and raincoat are advisable.

FOOD
If you have any dietary restrictions please contact with lipodistrofias@gmail.com