October 21 - 26, 2018

Viña Del Mar, Chile

VI

Latin American Course on Lysosomal Storage Diseases

Information & Program

Institutional Support: Instituto Genética Para Todos

Meeting Organization: VJS Assessoria de Eventos

Educational Grant: Shire

WELCOME ADDRESS

The organizers of the Latin American Course of Lysosomal Storage Diseases (LACLSD) welcome you to the VI Edition of this educational initiative.

After Guadalajara (2010), Mendoza (2011), Lima (2012), Gramado (2014) and Cartagena (2016), the course will now take place in Viña del Mar, a well-known conference place in Chile, where participants will find at Conference Town the appropriate atmosphere to exchange knowledge and interact scientifically and socially.

As in the previous five editions, the program of the VI LACLSD includes state-of-the-art topics addressed by top level specialists from Latin America, North America, and Europe to whom the organizers are deeply indebted.

This course aims to provide an overview and an update on the pathophysiology, clinical features, laboratory diagnosis and management of LSD to MDs from Latin America interested in this group of diseases.

The participants come from 13 Latin American countries (Argentina, Bolivia, Brazil, Chile, Colombia, Cuba, Ecuador, Honduras, Mexico, Nicaragua, Panama, Peru, and Venezuela) and will be exposed to 26 different presentations and will have the opportunity to present selected cases for group discussion.

In addition, a networking program was designed to further promote interaction and stimulate partnerships.

We would like to thank Shire for providing an unrestricted educational grant to support the course, VJS Assessoria de Eventos for the overall organization, and Instituto Genetica Para Todos for the institutional support.

Welcome to Viña del Mar and to the VI LACLSD!

Roberto Giugliani (Chair)
On behalf of the VI LACLSD Scientific Committee
(Hernan Amartino, Michael Beck, Luis Figuera)

SCIENTIFIC COMMITTEE

Roberto GIUGLIANI (Porto Alegre, Brazil - Chairperson)

Hernan AMARTINO (Buenos Aires, Argentina)

Michael BECK (Mainz, Germany)

Luis FIGUERA (Guadalajara, Mexico)

ORGANIZING COMMITTEE

Roberto Giugliani (Chair)

Vicky Simon (VJS)

Izabel de Souza (VJS/IGPT)

Fabrizio Barbosa (IGPT)

FACULTY

Hernán AMARTINO (Buenos Aires, Argentina)

Medical Doctor graduated at Universidad de Buenos Aires, Argentina, specialized in Pediatric Neurology. Head of Child Neurology Department at Hospital Universitario Austral. Buenos Aires. Argentina. Dr Amartino was trained in the field of inborn errors of metabolism at the Laboratorio de Neuroquimica by Dr Nestor Chamoles, in Buenos Aires. He has long term experience in Argentina with ERT for Fabry, Pompe and MPS. He is author of over 40 publications including books, book chapters and peer review journal articles. He is currently Principal Investigator in clinical trials for LSDs and co-director of PREIEM, an intensive training program on Mucopolysacharidosis for Latinoamerican physicians. He is also member of the following international boards related to LSDs: Pompe Registry Latam and International Boards, HOS (Hunter Outcome Survey) Latam Board, HOS CNS Task Force, and MPS IT Therapy Advisory Board.

Michael BECK (Mainz, Germany)

Professor of Pediatrics and Genetics, University of Mainz, Germany. Former head of Lysosomal Storage Disorders at the Children's Hospital, University of Mainz and Head of the Biochemical Laboratory. Consultant at the Genetic Institute of the University of Mainz. Principal Investigator in a variety of clinical studies and trials in the field of Lysosomal Storage Disorders (Natural History, Enzyme Replacement Therapy). Organizer of the 3rd (1993) and 8th (2004) International Symposium on Mucopolysaccharidoses and Related Diseases. Author of more than 300 Articles, mostly in peer-reviewed international journals.

Carmem BONFIM (Curtiba, Brazil)

Carmem Bonfim, MD, PhD, is the Head of the Pediatric Blood and Marrow Transplantation Program at the Hospital de Clinicas, Federal University of Parana, Curitiba, Brazil. Her major areas of interest are: Hematopoietic Stem Cell Transplantation (HSCT) for patients with Primary Immunodeficiencies, Inborn Errors of Metabolism and Inherited Bone Marrow Failure Syndromes. Dr Bonfim has also a special interest in newborn HSCT for selected genetic diseases detected after newborn screening and the use of alternative stem cell sources for pediatric transplantation (cord blood and haploidentical transplantation).

Maira BURIN (Porto Alegre, Brazil)

Pharmacist and Biochemist, with MsC and PhD in Biological Sciences-Biochemistry at Federal University of Rio Grande do Sul, Brazil. Senior Biochemist of the Medical Genetics Service, Hospital de Clínicas de Porto Alegre, Brazil. Training periods in London (UK), Munich (Germany), Porto (Portugal), and Bethesda (USA).

Luis FIGUERA (Guadalajara, Mexico)

MD degree obtained at Universidad de Oriente, Venezuela, and PhD degree at Universidad de Guadalajara, México. Post-Doctoral training at Baylor College of Medicine, Houston, Texas. Chairman of the Department of Genetics, Instituto Mexicano del Seguro Social, Guadalajara, México. Professor at the PhD Program of Human Genetics, Universidad de Guadalajara, México.

Roberto GIUGLIANI (Porto Alegre, Brazil)

Professor at the Department of Genetics, Federal University of Rio Grande do Sul, and Chief of the Medical Genetics Service of Hospital de Clinicas de Porto Alegre, Brazil. Is also Coordinator of the Brazilian Institute of Population Medical Genetics (INAGEMP), Director of the WHO Collaborating Centre for the Development of Medical Genetics Services in Latin America, President of the Latin American Society of Inborn Errors of Metabolism and Newborn Screening and Editor-in-Chief of the Journal of Inborn Errors of Metabolism and Screening.

Ursula MATTE (Porto Alegre, Brazil)

Biologist, has a PhD degree in Genetics and Molecular Biology at Federal University of Rio Grande do Sul, Brazil, and trainings in France, Australia and USA. Associate Professor of Genetics, Universidade Federal do Rio Grande do Sul. Head of the Experimental Research Center, and Coordinator of the Center for Gene Therapy, Hospital de Clinicas de Porto Alegre, Brazil. Afilliated Member of the Brazilian Academy of Sciences.

Joseph MUENZER (Chapel Hill, NC, USA)

Joseph Muenzer, MD, PhD, is Professor of Pediatrics and Genetics in the Division of Genetics and Metabolism, Department of Pediatrics at the University of North Carolina at Chapel Hill, USA, where he has practiced since 1993. He is actively involved in the diagnosis, management and treatment of patients with inborn errors of metabolism, especially mucopolysaccharidosis (MPS). Dr Muenzer's clinical research is directed toward the development of enzyme replacement therapy (ERT) for MPS. He was a principal investigator for the recombinant enzyme replacement clinical trials for MPS I and MPS II. Dr Muenzer is currently a principal investigator for the Phase I/II and Phase II/III clinical trials evaluating intrathecal ERT for the severe form of Hunter syndrome.

Gregory PASTORES (Dublin, Ireland)

Gregory M. Pastores, MD. Consultant, Adult Metabolic Service, Department of Medicine/National Centre for Inherited Metabolic Disorders, Mater Misericordiae University Hospital, Dublin, Ireland; Visiting Professor, Yale University, New Haven, CT, USA.

Uma RAMASWAMI (London, UK)

Uma Ramaswami is a Consultant in Inherited Metabolic Disorders based at the Lysosomal Disorders Unit (LSDU) at the Royal Free Hospital, London.Uma is the Clinical Lead for the LSDU at Royal Free Hospital and the National Clinical Lead for the UK Paediatric Familial Hypercholesterolaemia Register. She is actively involved in transition services for young patients with inherited metabolic disorders from paediatric to adult centres. Uma is interested in utilising technology such as telecommunication and apps to monitor and improve health care.Uma is a passionate faculty member involved in many inherited metabolic disorders educational and training initiatives in developing countries.Uma's research interest is mostly related to clinical research relating to understanding of the natural history and disease progression of metabolic disorders; and she is involved in a number of clinical trials in Inherited Metabolic Disorders.She has many peer reviewed publications on inherited metabolic disorders, written book chapters for inherited metabolic disorders, she is a communicating editor for the journal of inherited metabolic disorders and peer reviews manuscripts for several metabolic journals.

Paula ROZENFELD (La Plata, Argentina)

Dr Rozenfeld holds a degree in Biochemist and a PhD degree in the field of immunology, both from the National University of La Plata (UNLP), Argentina. At her postdoctoral work she began to work on lysosomal disorders. She works at Institute for Study of Immunology and pathophysiology (IIFP), UNLP-CONICET, La Plata, Argentina. She is Independent Researcher of CONICET, the National Council of Research in Argentina. She is the director of DIEL, a lab service for diagnosis of lysosomal disorders for patients from Argentina and other Latin-American countries. Her current research interests focus on the pathophysiology of immune system in lysosomal disorders. Since 2003, she has consulted, conducted research, provided diagnosis, written scientific papers and lectured in the area of lysosomal disorders.

Raphael SCHIFFMANN (Dallas, TX, USA)

Director, Institute of Metabolic Disease, Baylor Research Institute, Dallas Texas.
Clinical Professor Texas A&M University.
Neurometabolic diseases, lysosomal diseases.

Marie VANIER (Lyon, France)

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SCIENTIFIC PROGRAM

SUNDAY, October 21, 2018

Arrivals
Welcome Dinner (20:00 - 22:00, at hotel)

MONDAY, October 22, 2018

8:00 - 8:30
Welcome Address ▶ Course Objectives ▶ Introduction of Presenters and Students
Chairperson: **Roberto Giugliani**, Porto Alegre, Brazil

8:30 – 12:00
SESSION 1: INTRODUCTION TO LSDs
Chairperson: **Roberto Giugliani**, Porto Alegre, Brazil

08:30 – 09:15
GENERAL OVERVIEW OF LSDs
Presenter: **Michael Beck**, Mainz, Germany

9:15 – 10:00
CELL BIOLOGY AND BIOCHEMISTRY OF LYSOSOMES
Presenter: **Ursula Matte,** Porto Alegre, Brazil

10:00 - 10:30 - COFFEE BREAK

10:30 - 11:15
PATHOPHYSIOLOGY OF LSDs
Presenter: **Rafael Schiffmann,** Dallas, USA

11:15 - 12:00
GENETIC ASPECTS OF LSDs
Presenter: **Luis Figuera,** Guadalajara, Mexico

12:00 - 14:00 - LUNCH BREAK

SCIENTIFIC PROGRAM

14:00 - 17:30
SESSION 2: MUCOPOLYSACCHARIDOSES
Chairperson: **Hernan Amartino,** Buenos Aires, Argentina

14:00 - 14:45
CLINICAL ASPECTS OF MPS AND GLYCOPROTEINOSES
Presenter: **Joseph Muenzer**, Chapel Hill, NC, USA

14:45 - 15:30
BIOCHEMICAL AND GENETIC DIAGNOSIS
Presenter: **Maira Burin** and **Ursula Matte**, Porto Alegre, Brazil

15:30 - 16:00 - COFFEE BREAK

16:00 - 16:45
INTRAVENOUS ERT
Presenter: **Roberto Giugliani**, Porto Alegre, Brazil

16:45 - 17:30
THERAPIES FOR THE CNS
Presenter: **Joseph Muenzer**, Chapel Hill, NC, USA

18:00 - 20:00
SESSION 3: CASE PRESENTATIONS – PREPARATION
Chairperson: **Michael Beck**, Mainz, Germany

18:00 – 20:00
Group work: students will be divided in groups and will make a preliminary presentation of the case to be reported, with the supervision of a faculty member (refreshments will be available at meeting rooms)

SCIENTIFIC PROGRAM

TUESDAY, October 23, 2018

8:00 – 12:00
SESSION 4: FABRY DISEASE
Chairperson: **Luis Figuera**, Guadalajara, Mexico

08:30 – 09:15
CLINICAL CHARACTERISTICS IN MALES, FEMALES AND CHILDREN
Presenter: **Uma Ramaswami**, London, UK

9:15 – 10:00
BIOCHEMICAL AND MOLECULAR DIAGNOSIS
Presenter: **Maira Burin** and **Ursula Matte**, Porto Alegre, Brazil

10:00 - 10:30 - COFFEE BREAK

10:30 - 11:15
ENZYME REPLACEMENT AND OTHER THERAPIES
Presenter: **Michael Beck** (Mainz, Germany)

11:15 - 12:00
LONG-TERM OUTCOMES
Presenter: **Raphael Schiffmann**, Dallas, TX, USA

12:00 - 14:00 - LUNCH BREAK

14:00 - 17:30
SESSION 5: GAUCHER DISEASE
Chairperson: **Michael Beck**, Mainz, Germany

14:00 - 14:45
CLINICAL ASPECTS
Presenter: **Gregory Pastores**, Dublin, Ireland

14:45 - 15:30
BIOCHEMICAL AND GENETIC DIAGNOSIS
Presenter: **Paula Rozenfeld**, La Plata, Argentina

15:30 - 16:00 - COFFEE BREAK

16:00 - 16:45
NEUROLOGICAL MANIFESTATIONS
Presenter: **Hernan Amartino**, Buenos Aires, Argentina

16:45 - 17:30
TREATMENT AND FOLLOW-UP
Presenter: **Gregory Pastores**, Dublin, Ireland

SCIENTIFIC PROGRAM

WENESDAY, October 24, 2018

8:30 - 12:00
SESSION 6: NEURODEGENERATIVE LSDs
Chairperson: **Luis Figuera**, Guadalajara, Mexico

8:30 - 9:15
MECHANISMS OF NEURODEGENERATION
Presenter: **Hernan Amartino**, Buenos Aires, Argentina

9:15 - 10:00
NIEMANN-PICK C AND DEFECTS OF LYSOSOMAL MEMBRANE
Presenter: **Marie Vanier**, Lyon, France

10:00 - 10:30 - COFFEE BREAK

10:30 - 11:15
LEUCODYSTROPHIES
Presenter: **Hernan Amartino**, Buenos Aires, Argentina

11:15 - 12:00
STEM CELL TRANSPLANTATION
Presenter: **Carmen Bomfim**, Curitiba, Brazil

12:00 - 12:45
INFLAMMATION IN LSDs
Presenter: **Paula Rozenfeld**, La Plata, Argentina

SCIENTIFIC PROGRAM

THURSDAY, October 25, 2018

8:30 – 12:00
SESSION 7: CASE REPORTS
Chairperson: **Luis Figuera**, Guadalajara, Mexico
Moderators:
Juan Francisco Cabello, Santiago, Chile and **Uma Ramaswami**, London, UK

8:30 - 12:00
Presentation of cases by the students and discussion with faculty members

12:00 - 14:00 - LUNCH BREAK

14:00 - 17:30
SESSION 8: WRAP-UP
Chairperson: **Roberto Giugliani**, Porto Alegre, Brazil

14:00 - 14:45
NEUROLIPIDOSES, LIPOFUSCINOSES AND POMPE DISEASE
Presenter: **Juan Francisco Cabello**, Santiago, Chile

14:45 - 15:30
GUIDELINES FOR THE LABORATORY DIAGNOSIS OF LSDs
Presenter: **Marie Vanier**, Lyon, France

15:30 - 16:00 - COFFEE BREAK

16:00 - 16:45
SCREENING FOR LSDs
Presenter: **Roberto Giugliani**, Porto Alegre, Brazil

16:45 - 17:30
FUTURE PROSPECTS
Presenter: **Michael Beck**, Mainz, Germany

17:30 -
CLOSURE
Presenter: **Roberto Giugliani**, Porto Alegre, Brazil

FRIDAY, October 26, 2018

CHECK-OUT and DEPARTURES