

Brain Metabolism Workshop: from rare diseases to common biological pathways

Summer School – HIBRID JOURNEY



Course Director Dra. Angeles García-Cazorla

Dates

July 6-7, 2023

Imparting

SJD Barcelona Children's Hospital- Digital platform Esplugues de Llobregat. Barcelona

Education

SJD Barcelona Children's Hospital- Paseo Sant Joan de Déu, 2 08950 Esplugues de Llobregat – Barcelona - Tel+34 93 253 21 30

E-mail

Hospitalbarcelona.formacion@sjd.es Registration link

https://formacion.sjdhospitalbarcelona.org/

The workshop is supported by EJP RD NSS. Therefore we are able to offer this conference to you only for a minimal fee.*

In person: 25 € Online: 15 €

Countries in which the registration of participants will be free of charge: Countries of Latin America (including Brazil), Africa, the Middle East, Asia (not including Japan, South Korea). In this case, please send an email with a document proving your LMIC country membership to: <u>elisabet.hurtado@sid.es</u>

AIMED AT

- Health professionals dedicated to neurology (pediatric and adult) and/or inborn errors of metabolism and genetics.
- Researchers in the field of neuroscience, biochemistry and molecular biology.
- Family Associations.

FUNDED BY

This Networking event has received funding from European Union's Horizon 2020 research and innovation programme under the EJP RD COFUND-EJP N° 825575".



CO-FUNDED BY



IN PARTNERSHIP WITH:

European Reference Network

MetabERN







PROGRAM

	Thursday, 06 July 2023	
08:30-9:10	WORKSHOP OPENING Representatives of the organizing committee and of the ERNs that include neurol disorders (MetabERN, RND-ERN, EpiCare, EURO-NMD, ERN-Ithaca).	
BRAIN ENERGY	METABOLISM	Chair: Shamima Rahman, London
9:10 - 9.30	Brain energy homeostasis during neurodevelopment and aging	Felipe Barros, Chile
9:30 - 9:50	How is brain energy disturbed in inborn errors of metabolism? Main biological pathways	Isaac Marín-Valencia, New York
9:50 -10:10	Brain energy metabolism in rare genetic (non-classically metabolic) epilepsies	C. Fons, Barcelona (EpiCare)
10:10 -10:30	Mitochondria and Neurodegeneration	Alejandra Darling (RND.ERN)
10:30-11:20	Discussion	
COFFEE BREAK		
BRAIN AMINO	ACID AND PROTEIN HOMEOSTASIS	Chairs: Gumus Evren, Turkey
11:50 - 12:10	Role of amino acids and the proteostatic network in the developmetal brain	Gaia Novarino, Austria
12:10 - 12:30	Rare diseases of amino acids and neurological symptoms	Trine Tangeraas (MetabERN), Norway
12:30 - 13:10	Discussion	
LUNCH BREAK		
14:10 -15:00	ROUND TABLE: AVENUES FOR INTERACTION BETWEEN ERNs involved in neurological disorders for the development of a BRAIN METABOLISM InterERN NETWORK. Coordinators of the 3	
	ERNs	
COMPLEX LIPID	ERNS METABOLISM AND CELL TRAFFICKING IN THE BRAIN	Chair: Manuel Schiff, Paris
COMPLEX LIPID 15:00 - 15:20		Chair: Manuel Schiff, Paris Alfonso Oyarzábal, Barcelona
	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders.	Alfonso Oyarzábal,
15:00 - 15:20	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and	Alfonso Oyarzábal, Barcelona
15:00 - 15:20 15:20 - 15:40	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders. Neurological Manifestations of Cell trafficking and complex lipid	Alfonso Oyarzábal, Barcelona Pending confirmation A García-Cazorla, Barcelona
15:00 - 15:20 15:20 - 15:40 15:40 - 16:00 16:00 - 16:20 16:20 -17:00	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders. Neurological Manifestations of Cell trafficking and complex lipid disorders	Alfonso Oyarzábal, Barcelona Pending confirmation A García-Cazorla, Barcelona (MetabERN) Diego Martinelli, Rome
15:00 - 15:20 15:20 - 15:40 15:40 - 16:00 16:00 - 16:20	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders. Neurological Manifestations of Cell trafficking and complex lipid disorders Disorders of autophagy	Alfonso Oyarzábal, Barcelona Pending confirmation A García-Cazorla, Barcelona (MetabERN) Diego Martinelli, Rome (MetabERN)
15:00 - 15:20 15:20 - 15:40 15:40 - 16:00 16:00 - 16:20 16:20 -17:00 COFFEE BREAK	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders. Neurological Manifestations of Cell trafficking and complex lipid disorders Disorders of autophagy	Alfonso Oyarzábal, Barcelona Pending confirmation A García-Cazorla, Barcelona (MetabERN) Diego Martinelli, Rome
15:00 - 15:20 15:20 - 15:40 15:40 - 16:00 16:00 - 16:20 16:20 -17:00 COFFEE BREAK	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders. Neurological Manifestations of Cell trafficking and complex lipid disorders Disorders of autophagy Discussion	Alfonso Oyarzábal, Barcelona Pending confirmation A García-Cazorla, Barcelona (MetabERN) Diego Martinelli, Rome (MetabERN)
15:00 - 15:20 15:20 - 15:40 15:40 - 16:00 16:00 - 16:20 16:20 -17:00 COFFEE BREAK NEUROTRANSN	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders. Neurological Manifestations of Cell trafficking and complex lipid disorders Disorders of autophagy Discussion	Alfonso Oyarzábal, Barcelona Pending confirmation A García-Cazorla, Barcelona (MetabERN) Diego Martinelli, Rome (MetabERN) Chair: Roser Pons, Athens Matthjis Verhage,
15:00 - 15:20 15:20 - 15:40 15:40 - 16:00 16:00 - 16:20 16:20 -17:00 COFFEE BREAK NEUROTRANSN 17:30 -17:50	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders. Neurological Manifestations of Cell trafficking and complex lipid disorders Disorders of autophagy Discussion	Alfonso Oyarzábal, Barcelona Pending confirmation A García-Cazorla, Barcelona (MetabERN) Diego Martinelli, Rome (MetabERN) Chair: Roser Pons, Athens Matthjis Verhage, Amsterdam
15:00 - 15:20 15:20 - 15:40 15:40 - 16:00 16:00 - 16:20 16:20 -17:00 COFFEE BREAK NEUROTRANSN 17:30 -17:50 17:50 - 18:10	METABOLISM AND CELL TRAFFICKING IN THE BRAIN Biological Basis of Cell Trafficking in the Brain (proteins, exo, endocytosis, autophagy, transport across the cytoskeleton) and pathophysiological categories of disease Complex lipid role in membrane composition, trafficking, and Neurological disorders. Neurological Manifestations of Cell trafficking and complex lipid disorders Disorders of autophagy Discussion MISSION AND SIGNALING MOLECULES Disease mechanisms and therapeutic strategies for SNAREopathies, caused by mutations in presynaptic genes Neurotransmission in channelopathies "SynGo Clinic" a database proposal integrating biological	Alfonso Oyarzábal, Barcelona Pending confirmation A García-Cazorla, Barcelona (MetabERN) Diego Martinelli, Rome (MetabERN) Chair: Roser Pons, Athens Matthjis Verhage, Amsterdam Gabriella Horvarth, Canada







Friday, 07 July, 2023				
BRAIN METABOLISM IN MOTOR DISORDERS		Chair: Thomas Opladen, Heidelberg		
8:30 - 8:50	Metabolic pathways affected in Ataxia and Spastic Paraparesis	Roser Pons, Athens (RND-ERN)		
8:50 – 9:10	Metabolic pathways affected in Parkinsonism	Vincenzo Leuzzi, Rome		
9:10 - 9:30	Metabolic pathways affected in Dystonia and Chorea	Darío Ortigoza, Barcelona (RND-ERN)		
9:30 – 9:50	Discussion			
COFFEE BREAK				
BRAIN METABO DISEASES	LISM IN INTELLECTUAL DISABILITY and NEUROPSYCHIATRIC	<i>Chair:</i> Antonio Zorzano, Barcelona		
10:20 -10:40	Energy cost of cognition	Felipe Barros, Chile		
10:40 -11:00	Brain energy metabolism in Rett syndrome	Uliana Musokhranova, Barcelona		
11:00 - 11:20	Effects of neuropsychiatric medication on mitochondrial function.	T. Kozick, USA		
11:20 - 11:40	Discussion			
METABOLIC TRI DISEASES	EATMENTS also for RARE NEUROGENETIC "NON-METABOLIC"	Chair: Alejandra Darling		
11:40 - 12:00	Ketogenic Diet	Galina Stevanovic, Belgrade		
12:00 - 12:20	Metabolic treatments for neurodegenerative disorders	Fanny Mochel, Paris (MetabERN)		
12:20 -12:40	Strategies to cross the blood-brain-barrierBrains for Brain Foundation	Pending confirmation		
12:40 - 13:00	Discussion			
LUNCH BREAK				
14:00 - 15:00	ROUND TABLE - Patient advocacy			
LEUKODYSTROF	PHIES	Chairs: Enrico Bertini, Italy		
15:00-15:20	White matter and metabolism in the brain	Odile Boespflug-Tanguy, France		
15:20 -15:40	Factors predicting phenotypes in rare leukodystrophies amenable to treatment (X-ADL, MLD)	Nicole Wolf, The Netherlands (RND-ERN)		
15:40 - 16:00	"New treatments in genetic leukodystrophies	Caroline Sevin, Fanny Mochel, France		
16:00 -16:20	Discussion			
COFFEE BREAK				
16:50 - 17:20	Young doctors			
17:20	WORKSHOP CLOSURE			



