Défi Science
Maladies Rares du Neurodéveloppement
FILIÈRE NATIONALE DE SANTÉ

Xtraordinaire
Association de familles

Maladies rares Héréditaires du Métabolisme
Filière nationale de santé

INTERNATIONAL SYMPOSIUM ON CEREBRAL CREATINE DEFICIENCY SYNDROMES

September 29-30, 2023
Novotel Bercy, Paris - France

PROGRAM

ultragenyx pharmaceutical

CERES BRAIN THERAPEUTICS

Table of Contents

Welcome to the 2023 CCDS International Symposium in Paris !

On behalf of Xtraordinaire and the Scientific Committee, we would like to thank you all for attending our symposium.

We are truly honored to have attendees from 14 countries, 44 families, 19 CTD children, more than 40 speakers and professionals.

This year our symposium will focus on epilepsy, girls with CTD, clinical trials & biomarkers, newborn screening, gene therapy and update on research. We will also hear from the patients' associations, ACD and Xtraordinaire.

Moreover, we will emphasize the importance of collaborating between countries and how researchers. doctors, specialists, and laboratory experts can share their experience and expertise.

All together, we are stronger.

Carole Chehowah
Franck Defranco
Xtraordinaire

.

Symposium Scientific Committee

Olivier Braissant
Service of Clinical Chemistry, University Hospital of Lausanne

Vincent Desportes
Head of the Department of Child Neurology, University Hospital HFME, Lyon, France

Aloïse Mabondzo
PhD, HDR, Head of Neurovascular Unit Research & Therapeutic Innivation Laboratory at CEA Saclay in France

Fanny Mochel
Professor of Genetics at Sorbonne University

Association Xtraordinaire
Carole Chehowah
Vice President, co-manager of the CTD commission

Franck Defranco
Co-manager of the CTD commission

AGENDA

Friday, September 29

7:45-8:45 Registration / Welcome Coffee
9:00 Opening session
9:30-12:30 Morning sessions
12:30-13:45 Lunch
14:00-18:00 Afternoon sessions
18:00 Families gathering and discussion time
19:30-22:30 Dinner (boat departure @ 20h00)

Saturday, September 30

7:30-8:45 Breakfast
9:00-12:30 Morning sessions
12:30-13:45 Lunch
14:00-16:15 Afternoon sessions
16:15 Families Debriefing
17:00 Farewell Drinks

CCDS INTERNATIONAL SYMPOSIUM - PARIS 2023

PROGRAM - Friday, September 29 - morning sessions

7:30-8:45 - Registration / Welcome Coffee

9:00-9:30 - Opening session
Carole Chehowah / Franck Defranco
Gajja Salomons "Opening remarks"

Nicola Longo "Creatine and cyclocreatine transport in human fibroblasts"

9:30-10:30 - Session 1 - Epilepsy chaired by Juliette Bouchereau
Aurore Curie “CREAT_criteria : what have we learned regarding CTD patient epilepsy ?”
Laurent Bailly “Epilepsy in adult patients with creatine transporter deficiency“
Rima Nabbout "Precision medicine and epilepsy: present and future"

Roundtable / Q&As : Diagnosis, epilepsy & therapeutics approaches
Laurent Bailly, Aurore Curie, Rima Nabbout

Coffee break

11:00-12:30 - Session 2 - “GIRLS with CTD” chaired by Fanny Mochel
Taylor Kane "Remember The Girls: Not Just Carriers"
Jiddeke van de Kamp "It is different for girls: X-linked creatine transporter deficiency in females"
Audrey Mittelman & Carole Chehowah "Key findings of the Girls' questionnaire"
Olivier Braissant "Female characterization in the *Slc6a8*^{Y389C} rat model of CTD"

Roundtable / Q&As : Recognition, specificities, diagnosis, mums & sisters
Olivier Braissant, Carole Chehowah, Taylor Kane, Audrey Mittelman, Marzia Pasquali, Jiddeke van de Kamp

12:30-13:45 ***Lunch buffet in the main dining room***

CCDS INTERNATIONAL SYMPOSIUM - PARIS 2023

PROGRAM - Friday, September 29 - afternoon sessions

14:00-17:00 - Session 3 "Into clinical trials" chaired by Olivier Braissant

Imaging

Pierre Gilles Henry "Can magnetic resonance spectroscopy contribute to clinical trials targeting creatine metabolism?"
Nicolas Tournier "Imaging biomarkers for CNS drug development"
Laura Baroncelli "Neuroimaging tools to develop a biomarker of brain function for creatine deficiency".
Maurizio Balestrino "Selective alteration of the left arcuate fasciculus in creatine transporter deficiency"

Coffee break

Clinical Endpoints

Fanny Mochel "Lessons from the Study of the Nervous System Metabolism: Outcome Measures in Clinical Trials for Rare Diseases"
Andreas Schultze "Designing clinical trials for CDS"
Aurore Curie "CREAT_criteria, a prospective study in Creatine Transporter Deficiency (SLC6A8) patients to determine the most relevant outcome measures : preliminary results"

Roundtable / Q&As :
Laura Baroncelli, Aurore Curie, David Germanaud, Pierre-Gilles Henry, Fanny Mochel, Andreas Schultze, Nicolas Tournier

17:30-18.00 - Session 4 - Importance and role of patients' groups
"Expectations & Engagement - Patients empowerment, A driving force for rare disease research"
Heidi Wallis, ACD / Carole Chehowah, Xtraordinaire

18:00 – Families gathering and discussion time

19:15 - Meet in front of the hotel
19:30-22:00 - ***Dinner Cruise on "le Mistinguett"***

PROGRAM - Saturday, September 30 - morning sessions

9:00-10:15 - Session 5 - "Importance of early diagnosis and newborn screening" chaired by Vincent des Portes
Marzia Pasquali "GAMT deficiency: the road to newborn screening"
Andreas Schultze "GAMT deficiency – the perfect screening condition"
Gajja Salomons "Neonatal screening in the Netherlands"
David Cheillan "Neonatal screening in France : Overview of new organization, recent developments and future challenges"

Roundtable / Q&As :
David Cheillan, Marzia Pasquali, Gajja Salomons, Andreas Schultze

10:15-10:30 Ad Scientam
Alizé Vivès "Transforming neurological and mental health patient journey through digital biomarkers"

10:30-11:00 Panel discussion "Growing up and living CTD"
Jiddeke van de Kamp, Dan Coller, Karine Defranco, Ludovica Iovino, Kim Soesbergen

Coffee break

11:15-12:30 - Session 6 - "From the Future – part 1 - Advanced Therapies" chaired by Aloïse Mabondzo
Léa Broca-Brisson "Deciphering Neuronal Deficit and Protein Profile Changes in Human Brain Organoids from Patients with Creatine Transporter Deficiency"
Melanie Brandabur - Ultragenyx "CTD Program update from Ultragenyx"
Thomas Joudinaud - Ceres Brain Therapeutics "Update on CBT101 development: A steeple race toward clinical trials."

12:30-13:45 ***Lunch buffet in the main dining room***

PROGRAM - Saturday, September 30 - afternoon sessions

14:00-15:30 - Session 6 - "From the Future – part 2 - Gene therapy" chaired by Aloïse Mabondzo

"Gene therapy ABC" **by Olivier Braissant**

Ludovica Iovino "Creatine Transporter Deficiency: the long journey towards successful gene therapy"
Olivier Braissant "Rescue of myocytes and locomotion through AAV9 intracisternal gene therapy in a rat model of creatine transporter deficiency"
Gerry Lipshutz "Development and Insights into Gene Therapy for Guanidinoacetate Methyltransferase Deficiency"

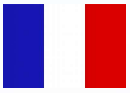
Roundtable / Q&As : "pros and cons, potential for CTD patients"
Olivier Braissant, Laura Baroncelli, Ludovica Iovino, Gerry Lipshutz

15:45-16:15 - Symposium Conclusion

Farewell Drinks

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Association Xtraordinaire



Xtraordinaire is a French based non-profit organization, founded by parents in 2006, for families and patients with rare X-linked neurodevelopmental Syndromes, such as Creatine Transporter Deficiency, a community dedicated to improve lives of the patients and their families, and to encourage research programs.

Key missions :

- Help, support and inform families about their rights, access to education, caregiving and therapeutic solutions
- Work to build awareness, educate the global community
- Promote, encourage and fund research programs
- Create a strong network to strengthen partnership between researchers, physicians and patients
- Represent and fight for patients and families interests
- Advocate to increase and shorten the path to diagnosis

The Commission for Creatine Transporter Deficiency (SLC6A8) represents more than 80% of the French CTD families, and has been crucial in initiating, funding, promoting research programs and upcoming therapeutic trials. Xtraordinaire CTD commission is also in contact with European CTD Families. Xtraordinaire has also been partnering with the US Association for Creatine Deficiencies since 2016.

Representing Xtraordinaire SLC6A8 commission and organizers of the symposium , Carole Chehowah and Franck Defranco, respectively parents of a 33 year old CTD girl, and a 18 year old CTD boy.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Laurent Bailly

Neurologist (MD, MSc)
La Pitié-Salpêtrière Hospital Paris

Laurent Bailly is an adult neurologist (MD, MSc) at the epilepsy unit in Pitié-Salpêtrière hospital (Paris, France). He is a member of the reference center for rare and complex epilepsies at Pitié-Salpêtrière hospital, headed by Pr Navarro and Dr An. He is involved in the clinical care of adult patients with genetic or metabolic epilepsy and in scientific projects regarding these topics. Through his intervention, he will share a literature review and the experience of the reference center concerning epilepsy in adult patients with creatine transporter deficiency.

Prof. Maurizio Balestrino

Department of neuroscience
University of Genoa

Maurizio Balestrino received the Degree in Medicine and Surgery, with honors, in 1980 and the Diploma of Specialist in Neurology in 1984. From 1983 through 1986 he was a Research Associate in the Department of Physiology at Duke University, where he learnt experimental methods in electrophysiology and started a keen interest in the anoxic brain damage and in neuroprotection. Still at that time he became interested in creatine as a neuroprotectant. Back in Italy he continued to have clinical responsibilities as a neurologist and to carry out experimental research in brain anoxia or ischemia. At present he is Associate Professor of Neurology at the University of Genova. He works as a clinician in the Stroke Unit. He is also Director of the Laboratory of Experimental Neurophysiology in the same Department. He has been Principal Investigator or Partner in national and international research projects. His research interests include creatine as a medicinal compound, creatine-derived compounds with therapeutical effects, and the therapy of acute ischemic stroke. He aims at bridging the gap between experimental research and clinical therapy.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Laura Baroncelli

Neuroscience Institute CNR
IRCCS Stella Maris Foundation

Dr. Baroncelli graduated in Biology at the University of Pisa in 2005 and trained in the PhD program in Neurobiology at the Scuola Normale Superiore from 2006 to 2009. Following a fellowship at Scuola Normale Superiore, she was awarded in 2010 a two-year post-doctoral fellowship at the Accademia Nazionale dei Lincei, in Italy. Since 2011, she is Tenured Researcher at the Neuroscience Institute (IN) of CNR in Pisa. In 2017, she was awarded a six-month travel grant within the program “Post-Doctoral Fellowship- 2017” of Fondazione Umberto Veronesi for a training period with two-photon microscopy at the University Medical Center of Göttingen. Moreover, she is responsible of the group for the research in rare neurodevelopmental disorders and co-responsbile of the fNIRS lab at the Stella Maris Institute. Her scientific production was highly fruitful leading to the publication of 58 original research papers in international peer-reviewed journals (H index Scopus: 25). She was awarded personal fundings by Fondazione Roma, LUMOS Pharma, Italian Ministry of Health, Lejeune Foundation, Telethon Foundation and the European Joint Programme for Rare Diseases for the study of creatine-related disorders and other neurodevelopmental. She is also Academic Editor of Neural Plasticity and Scientific Report, and reviewer for various international journals and national agencies.

Juliette Bouchereau

PH - Hôpital Necker enfants malades

Juliette Bouchereau is a pediatrician specializing in metabolic disorders. After working in the neuropediatric department at Robert Debré hospital, she now practices at the Necker-Enfants malades hospital in Paris, in the reference center for hereditary metabolic diseases. She is in charge of children and adults with all types of metabolic diseases, including neurometabolics, in close collaboration with neuropediatricians.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Prof. Olivier Braissant, PhD

Service of Clinical Chemistry
University Hospital of Lausanne

After studies of biology at the University of Lausanne, I did a PhD and a postdoc focused on transcription factors of the nuclear receptors family, also specializing on cell biology and physiology of central nervous system, being particularly interested in its development.

I joined the Service of Clinical Chemistry of the University Hospital of Lausanne (CHUV) in 1997, in which I lead the research section, working on metabolic diseases (including IEM) affecting brain development. Since more than 25 years, I developed my research line on the way Creatine Deficiency Syndromes (CDS), Urea Cycle Diseases (including ammonium intoxication of CNS) and Organic Acidurias affect brain development and function, through the use of both original in vitro and in vivo experimental models, including for the development of new treatment strategies.

I am also specialized for the IEM diagnostic laboratory of the CHUV, and teach biochemistry, IEM and brain development to medical and biology students of the University of Lausanne (UNIL) (bachelor, master and PhD/MD-PhD levels).

Melanie Brandabur

Senior Medical Director
Ultragenyx

Melanie Brandabur, MD received her BA degree from the University of Illinois in Urbana and her MD degree from Rush Medical College in Chicago. She completed her neurology residency and Movement Disorders and Neuropharmacology fellowship at Rush-Presbyterian-St. Luke's Medical Center in Chicago. This was followed by a post-doctoral basic sciences fellowship in Neurodegenerative Diseases. Dr. Brandabur is currently a Senior Medical Director in Global Clinical Development at Ultragenyx Pharmaceutical Inc., where she works on the development of therapeutic agents for neurodevelopmental rare diseases. During her clinical career as a specialist in Parkinson's disease and Movement Disorders, Dr. Brandabur served as the Medical Director for three National Parkinson Foundation Centers of Excellence; at the University of Illinois, at Alexian Neurosciences Institute and at the Parkinson's Institute in Sunnyvale, California

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Léa Broca-Brisson

University Paris-Saclay, CEA

Department of Medicines and Technologies for Health (MTS), Neurovascular unit and Therapeutic Innovation Laboratory (LENIT), Gif-sur-Yvette cedex 91191, France.

I joined the CEA in February 2020 as a PhD student, under the supervision of Dr Aloïse Mabondzo, in the Neurovascular unit and Therapeutic Innovation Laboratory. My research focuses on the development of human brain organoids from induced pluripotent stem cells (iPSCs) derived from CTD patients, in order to determine the efficacy of therapeutic agents as a treatment for CTD.
As a cell biologist, I am interested in understanding the functions of cells, their interactions with the environment, and their potential dysfunctions. I aim to develop more predictive models to better study pathophysiology and determine the efficacy of drug candidates.

David Cheillan

PharmD / PhD
Hospices Civils de Lyon

Medical biologist (PharmD) and PhD in Neuroscience, I am Biochemist in the department “inborn errors of metabolism and neonatal screening” in the university Hospital of Lyon, France. In the field of newborn screening (NBS), I am in charge of the “neonatal screening” unit for the Lyon area with the responsibility to screen in this region (~45 000 newborns/years) for the diseases of the French screening program (6 diseases until 2022 and we have implemented 6 new IEM since January 2023). At the national level, I am the coordinator of the French NBS laboratories for Ministry of Health and vice-president of the French society for neonatal screening (SFDN). I am also in charge of the biological and molecular diagnosis of different inborn errors of metabolism (peroxisomal disorders, creatine deficiency, galactosemia, CDG syndromes and bile synthesis defects).

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Aurore Curie

MCU-PH/Neuro-pediatrician/Dr
Hospices Civils de Lyon

French National Reference Center for Rare Diseases with Intellectual Disability, Department of Child Neurology, Woman Mother and Child Hospital, Lyon University Hospital, Bron, France; Lyon Neuroscience Research Center, CNRS UMR5292, Inserm U1028, Lyon, France; Claude Bernard Lyon 1 University, Lyon, France Bio Aurore Curie is a child neurologist (MD, PhD) at the Child Neurology Department of Lyon Hospital (Assistant Professor) and the Reference Center for Intellectual Disability (ID) from rare causes (Co-Head). She is affiliated to the Lyon Neuroscience Research Center (CNRS UMR5292, Inserm U1028, Lyon, France) and also part of the DéfiScience national network for rare diseases of brain development and ID. She coordinates a French Inter University Diploma (DIU) on Neurodevelopmental Disorders. She has a strong expertise in genetics (especially in X-linked ID) and in neuroscience. She developed new outcome measure adapted to ID patients (HCL/CNRS patent). She contributed to the development of the research platform “Cognitoscope”. Her clinical and research expertise is dedicated to X-Linked ID and other ID from rare causes. She described cognitive profiles of neurodevelopmental disorders (including ARX, PQBP1, Rab-GDI, SLC6A8 mutated patients) using eye-tracking and neuroimaging analysis, and contributed to several multisite clinical trials for Fragile X syndrome. She also furthered our knowledge on placebo effect in ID patients, and the different trial plans that can be used in ID patients to test for an effect (Randomized controlled double blind Clinical Trials (RCT) but also n-of-1 trials, also called Single-Case Experimental Designs or SCEDs).

Vincent des Portes, MD, PhD

Head of the department of child neurology,
University Hospital HFME, Lyon

Professor of Pediatrics, Medical school Lyon Sud, University Lyon 1, France; Head of DéfiScience, the French national network for rare diseases of Neurodevelopment.

Vincent des Portes brings 20 years of experience in genetic syndromes with intellectual disability. His work spans therapeutic trials in genetic syndromes with intellectual disability. In the frame of an international consortium his team published a set of recommendations on the reliability of preclinical data, the choice of endpoints, and the design of future trials. He also focuses on clinical characterization of genetic syndromes with intellectual disability, contributing to the clinical phenotyping of various syndromes. Lastly, his team has been monitoring patients born with agenesis of the corpus callosum (ACC) for the last 15 years.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

David Germanaud

MD, PhD

Child neurologist specializing in developmental cognitive neurology and neurodevelopmental conditions, former head and actual team member of the Robert-Debré Reference Centre for Rare Diseases with intellectual disability and other early-onset severe neurodevelopmental disorders (DéfiScience network), Genetics Department of Robert-Debré University Hospital, AP-HP (Paris). Developmental neuroscientist specializing in developmental neuroimaging, head of the Applied, Clinical and Translational Neuroimaging Lab (UNIACT), NeuroSpin CEA Paris-Saclay (Gif/Yvette), and member of the InDEV team "Imaging neurodevelopmental phenotypes", Inserm UMR1141 NeuroDiderot, dedicated to the multimodal study (clinical, psychometric and imaging) of anatomical-functional variability, vulnerability and plasticity during brain development. My research group "Functional neuroanatomy, brain size & scaling, fetal alcohol" is dedicated to the imaging study of neurodevelopmental disorders associated with abnormal brain growth and scaling, mainly growth failure in the context of fetal alcohol, but also in rare monogenic models (PQBP1, NR2F1, PTEN, FGFR2...) in collaboration with reference clinical teams. One of its main objectives has recently been to identify new neuroanatomical markers with diagnostic or prognostic values in Fetal Alcohol Spectrum Disorders (FASD), and to contribute to a better understanding of the anatomical-functional developmental phenotype of these patient.

Pierre-Gilles Henry

MS, PhD

University of Minnesota

Dr. Henry is a tenured Associate Professor of Radiology at the Center Magnetic Resonance Research, University of Minnesota, USA. He graduated with a degree in Electrical Engineering from Supélec (now CentraleSupélec) near Paris, France. He holds a MS and PhD in Neuroscience from University Paris 6 (now Sorbonne University) in Paris. Dr Henry is an expert in Magnetic Resonance Spectroscopy (MRS) to study brain metabolism and neurochemistry. Currently, his primary focus is on studying brain and spinal cord alterations in Friedreich Ataxia using not only MRS, but also MRI, diffusion MRI, and QSM. He is also working on developing new techniques for real-time correction of motion during MRI scans.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Ludovica Iovino

PhD, Neuroscience Institute, CNR
IRCCS Stella Maris Foundation

Dr. Ludovica Iovino graduated in Biology at the University of Florence in 2015 and trained in the PhD program in Biomedical Sciences, curriculum Neurophysiology, at the University of Florence from 2015 to 2018. Her PhD research activity was devoted to understand the basic neural mechanisms underlying the respiratory rhythm generation. In 2019, following a short period as visiting researcher at the University of Florence, she was awarded a three-year post-doctoral fellowship at the Department of Biology of the University of Padova, where she studied how aberrant glutamate handling by glial cells contributes to different brain pathophysiological conditions. Since November 2022, she joined Dr. Laura Baroncelli's Lab at the Neuroscience Institute of CNR in Pisa for a second postdoctoral project granted by the Telethon Foundation. Her research activity is currently dedicated to the study of preclinical efficacy of gene therapy in the treatment of Creatine Transporter Deficiency. Her scientific led to the publication of 12 original research papers and 2 Scientific Reviews in international peer-reviewed journals.

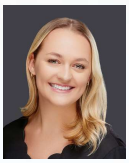
Thomas Joudinaud

MD, PhD - CEO & co-founder of Ceres brain Therapeutics

Thomas Joudinaud MD, PhD is a medical doctor by training, former cardiac surgeon in Paris with a PhD from the University of Montana in the USA.
Seeking new professional challenges, Thomas decided to switch career and move to strategy consulting for pharma companies. He joined the Boston Consulting Group in Paris where he worked for 7 years, mainly on business strategy projects all over the world for pharma companies. Then, he joined AEC Partners, a Strategy Consulting Firm specialized in HealthCare, advising biotechs and startups.
He is a board member of Osivax, a vaccine-focused BioTech company.

Ceres Brain Therapeutics, spin-off from the leading academic French center, the CEA, was created in 2019 by the CEA, Dr Aloise Mabondzo, PhD, Dr Henri Bénech, PharmD, PhD, Dr Thomas Joudinaud, MD, PhD and other researchers from the CEA. Ceres Brain Therapeutics aims at tackling the challenges of neurological diseases by developing new therapeutic approaches such as the creatine-to-neurones(TM) CBT101 solution. Privately owned and founded, Ceres is presently conducting the IMPD/IND studies for CBT101.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS



Taylor Kane

Founder of Remember the Girls

Taylor Kane's passion for rare disease advocacy began in grade school, shortly after her father died from the rare X-linked condition adrenoleukodystrophy (ALD) and she learned that she was a genetic carrier of the disease. Not only did Taylor help raise substantial money for ALD research, she successfully lobbied the New Jersey legislature and governor to enact a law requiring the screening of newborns for ALD in New Jersey. Over the years, Taylor has become deeply involved in the rare disease community, speaking at numerous rare disease-related events and contributing to a variety of campaigns as a thought leader and voice for the rare disease community. Taylor is the founder and consulting executive director of Remember The Girls, an international nonprofit organization which aims to break the stigma facing females with X-linked disorders. Taylor is a summa cum laude graduate of The George Washington University who primarily resides in New Jersey, but spends a great deal of time in the United Kingdom and the Czech Republic. She is an award-winning activist, an accomplished speaker, and a respected author, having recently published a memoir, Rare Like Us: From Losing My Dad to Finding Myself in a Family Plagued by Genetic Disease.

Gerry Lipshutz

Physician - Scientist

University of California

Gerald S. Lipshutz, MD, MS, received his medical degree from the University of California Los Angeles (UCLA) School of Medicine and completed his postgraduate training at the University of California San Francisco School of Medicine. Dr. Lipshutz is a Professor-in-Residence within the Departments of Surgery and the Department of Molecular and Medical Pharmacology. He is also a member of the Intellectual and Developmental Disabilities Institute at UCLA along with the Broad Center; he presently holds the Goldwyn Chair. Clinically, within the David Geffen School of Medicine his clinical and interests include liver and pancreas transplantation and gene and cell therapies for single gene metabolic disorders of the liver. Dr. Lipshutz has been an invited participant in National Institutes of Health (NIH) conferences and has served as a grant reviewer for both Wellcome Trust and the US National Institutes of Health where he is presently a standing member of the GDD Study Section.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Nicola Longo

Professor of Pediatrics, Chief, Medical Genetics
University of Utah

Dr. Nicola Longo received his M.D. and Ph.D. in molecular biology and pathology from the University of Parma, School of Medicine in Italy. He then trained in Pediatrics, Medical and Biochemical Genetics at Emory University in Atlanta, Georgia, USA He is a Professor of Pediatrics and Adjunct Professor of Pathology, Nutrition and Integrative Physiology at the University of Utah in Salt Lake City, UT. He is also the Chief of the Division of Medical Genetics, Director of the Training Program in Medical Biochemical Genetics and Medical Director of the Biochemical Genetics Lab at ARUP Laboratories in Salt Lake City. His clinical research concerns the molecular bases of metabolic disorders, their identification through newborn screening, their natural history, and the development of novel therapies. He is the principal investigator on more than 20 active clinical trials, has published >180 peer-reviewed articles, in addition to numerous review articles and 30 book chapters on metabolic disorders.
He has a long-standing interest in membrane transporters for which he has worked on amino acid, glucose, carnitine and creatine transporters. He follows several patients with brain creatine deficiency and has an active interest in developing new methods to facilitate their detection by newborn screening, improving existing therapies and developing new ones for these conditions.

Aloïse Mabondzo

PhD, HDR
CEA Paris Saclay

Dr Aloïse Mabondzo, research director is a head of Neurovascular Unit Research & Therapeutic Innivation Laboratory at CEA Saclay in France. He joined the CEA, the Life Science Division, in May of 1998 as the leader of a neurovascular pharmacology Lab with a strong focus on in vitro blood-brain barrier (BBB) modeling and pathophysiology of the brain. His Lab has developed fully characterised in vitro screening tools allowing the optimisation of the molecules under development for brain penetration. His innovative research has made possible the development of research programs in the neuroscience field : Alzheimer's disease, nanotoxicology, ischemic hypoxia encephalopathy, X-linked creatine transporter deficiency disease. Dr Aloïse Mabondzo is author or co-author of 64 articles in peer reviewed journals, seven patents, gave lectures as lecturer and as well as guest speaker, poster presentation in the scientific congress, and he often reviews articles for scientific journals. He has directed twenty PhD students, and seven postdoctoral positions have been part of his team. As a Neuroscientist, Dr Mabondzo aims to bridge the gap between experimental research and clinical therapy for cerebral diseases. He is a cofounder of CERES BRAIN THERAPEUTICS, a spin-off from the French alternative energies and Atomic Energy Commission (CEA), committed to focus its resources to the preclinical development of advance drug over coming years in order to provide CTD patients with a therapeutic solution to deliver creatine in the brain.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Audrey Mittelman

Department of Child Neurology
Hospices Civils de Lyon

Audrey Mittelman is a junior child neurologist who will finish her residency in November. She will then join the team of Pr Vincent Des Portes in the department of Child Neurology of Lyon Hospital.

Fanny Mochel

Professor of genetics Sorbonne University

She received her MD in Genetics in 2005 at the University Paris Descartes, her PhD in Neuroscience in 2010 at Sorbonne University and is board certified in inborn errors of metabolism.
Prof. Mochel leads the French reference centers on Neurometabolic diseases and Leukodystrophies in adults and co-leads a research team dedicated to Neurometabolism and neuroimmunity at the Paris Brain Institute of La Pitié-Salpêtrière University Hospital in Paris. She is chair of the Adult Metabolic Group of the Society for the Study of Inborn Errors of Metabolism (SSIEM), council member of the SSIEM, and co-chair of the French society for inborn of errors of metabolism in adults.
Her research focuses on the characterization and treatment of brain energy deficiencies in neurometabolic and neurodegenerative diseases, as well as novel treatments targeting metabolism and innate immunity in leukodystrophies. Her major areas of expertise are the identification of neurometabolic biomarkers in vitro(metabolomics) and in vivo (metabolic imaging) as well as therapeutic approaches targeting the Krebs cycle.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Rima Nabbout, MD, PhD

Professor of Pediatric Neurology
University Paris Cité

Rima Nabbout, MD, PhD, is professor of paediatric neurology at Université Paris cité and director of the French Reference Centre for Rare Epilepsies at Necker-Enfants Malades, Paris, France.
She is member of EJP-RD (European Joint Programme on Rare Diseases), leading the WP20 on innovation in clinical trials in RD; of ERN (European Reference Networks) EpiCARE steering committee and research council and of IRDiRC (International Rare Diseases Research Consortium).. She is president of the scientific committee of the BNDMR (Banque Nationale de Données Maladies Rares), coordinator of the task force on transition of the International League Against Epilepsy, and core member of 2 task forces on nosology and Big Data.
Professor Nabbout's research at Institut Imagine (Director of the chair of Epilepsy and leading the epilepsy program at INSERM U1163, Translational Research for Neurological Diseases) is focused on childhood rare epilepsies, aiming to syndromes' delineation, deep phenotyping, defining disease impact and underlying mechanisms. She is principal investigator in clinical trials on rare epilepsies with development of patient-centred end-points and innovative methodologies for personalized therapies. She has authored more than 300 peer-reviewed papers and received Horizons 2020, FP7, national grants, and philanthropic funds, mainly from FAMA Swiss funds.

Marzia Pasquali, PhD, FACMG

Medical Director, Biochemical Genetics &
Newborn Screening Section Chief

Dr. Pasquali is a Professor of Pathology, the Program Director of the ACGME accredited Fellowship program in Clinical Biochemical Genetics at the University of Utah School of Medicine, and the Section Chief and Medical Director of Biochemical Genetics at ARUP Laboratories. Dr. Pasquali earned her degrees of Doctor in Pharmaceutical Chemistry and Technology and Pharmacy Doctor at the University of Parma School of Pharmacy in Italy. She trained in Clinical Biochemical Genetics at Emory University, in Atlanta, Georgia where later served as the co-director of the Biochemical Genetics Laboratory. Dr. Pasquali is board certified in Clinical Biochemical Genetics. She is a member of the Society for Inherited Metabolic Disorders, the American College of Medical Genetics and Genomics, and several other professional societies. Her research interests are newborn screening, disorders of carnitine and creatine metabolism and transport, and lysosomal storage disorders.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Gajja Salomons

Professor of Molecular Biology of Neurometabolic Disorders
Vrije Universiteit Amsterdam, Amsterdam

Dr. Salomons is professor of the Department of Clinical Chemistry and head of the merged Laboratory Genetic Metabolic Diseases (GMD) at the University Medical Centers in Amsterdam, location AMC, Amsterdam, The Netherlands. This lab provides diagnostics and research on (inborn) errors of metabolism on the following levels: metabolomics (metabolites in body fluids), enzymes, genetics (DNA/RNA) and is specialized in functional studies in overexpression models to classify genetic variants. The combined expertise on massaspectrometry, biochemistry, cell models and genetics is an unique feature of this laboratory. The overall aim of the GMD is unraveling the cause of metabolic disorders, development of diagnostic approaches and therapies for specific (inborn) errors of metabolism. Salomons is member of the expertise center for creatine defects in Amsterdam and she has an special interest in creatine metabolism and transport for which Salomons received several grants and prizes.

Prof. Andreas Schultze

Hospital for Sick Children and University of Toronto

Dr. Andreas Schulze is a metabolic pediatrician and professor in the Departments of Paediatrics and Biochemistry at University of Toronto. He is Medical Director of the Newborn Screening Program at The Hospital for Sick Children (SickKids) Toronto and Senior Associate Scientist at the SickKids Research Institute.
Schulze receive a medical diploma and a doctorate in medicine from the Faculty of Medicine at Leipzig University in 1987. After completing graduate training and PhD in Physiological Biochemistry under the supervision of Dr. Hans-Joachim Boehme and Dr. Eberhard Hoffmann (1987-1992), Schulze pursued postgraduate clinical training in Pediatrics at the University Children's Hospital in Heidelberg under Dr. Hans-Juergen Bremer and Dr. Georg F. Hoffmann (1992-1999). Schulze defended a Professorial Thesis
(Habilitation) and received the Venia Legendi from the Ruprecht-Karls University Heidelberg in 2004. He is board certified in Physiological Biochemistry (1993) and in Pediatrics (1999).
Since 2007, Schulze works as clinician scientist at The Hospital for Sick Children in Toronto. As a clinician he takes care of children with inborn errors of metabolism and oversees the SickKids Newborn Screening Program. As a scientist, he established a research group and a research laboratory at the SickKids Research Institute. Dr. Schulze's research is centered around creatine deficiency syndromes and regulation of creatine homeostasis. His research encompasses the metabolism of arginine, ornithine,
and guanidino compounds, and includes small molecule drug discovery.

CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Nicolas Tournier

PhD, PharmD

Université Paris-Saclay

Nicolas Tournier (PhD, PharmD) is hospital pharmacist and pharmacologist at Université Paris-Saclay.

He leads the Pharmacological Neuroimaging group of the BioMaps research unit (CEA/Inserm/CNRS/Université Paris-Saclay). The main aim of this group is to promote the clinical translation of original imaging techniques to improve our comprehension of brain function, in health and disease. The team has ample expertise in pharmacological imaging to study the mechanism and effects of CNS drugs to the brain in preclinical disease models and patients.

Jiddeke van de Kamp, MD, PhD

Consultant Clinical Genetics

Amsterdam University Medical Center, VU Medical Center

Dr. Jiddeke van de Kamp is a consultant at the department of Clinical Genetics of the Amsterdam University Medical Center. She did her PhD in 2014 on X-linked creatine transporter deficiency under supervision of professor Gajja Salomons and dr Grazia Mancini. This work focuses on the clinical features in male patients and female carriers and the evaluation of arginine and glycine supplementation treatment. It also includes an hypothesis on the pathophysiology of creatine transporter deficiency.
She is a member of the creatine expertise team at the Amsterdam University Medical Center, location VU Medical Center.

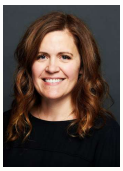
CCDS INTERNATIONAL SYMPOSIUM - SPEAKERS

Alizé Vivès

Medical Innovation Project Manager, Ad Scientam

Alizé Vivès is Medical Innovation Project Manager at Ad Scientiam. Alizé is pharmacist (PharmD) and graduated from the Strategy and Management of International Business master of ESSEC Business School. She worked for a few years in conducting patient studies to generate real-life data. She participated in various projects on chronic and rare diseases, promoting the patient voice to the scientific community. She then decided to specialize in Digital Health and joined Ad Scientiam to discover new digital biomarkers to empower patients and clinicians within patient journeys. She works closely with medical experts,patients & patients advocacy groups to design innovative digital solutions. She mainly works on projects in Neurosciences, Rare diseases and Psychiatry.

Ad Scientiam is a French MedTech company, based in Paris. It was created in 2013, in the Brain Institute in Paris. With a team of more than 65 people with various expertise, Ad Scientiam is specialized in the development of connected medical devices, adapted to the monitoring of chronic diseases, including rare or neurological ones. These new tools allow patients and caregivers to monitor the evolution of their main symptoms at any time, and from their home, using their smartphone. As an example, Ad Scientiam launched in 2018 MSCopilot®, the first CE-marked software medical device for the self-assessment of multiple sclerosis patients. New devices are currently being validated in neuroscience, rare diseases and mental disorders.



Heidi Wallis, BS

Executive Director

Association for Creatine Deficiencies

Heidi is the Executive Director of the Association for Creatine Deficiencies and the parent of two children with GAMT Deficiency, a creatine synthesis disorder. Prior to her current role, Heidi advocated for creatine deficiencies as an ACD volunteer board member from 2015-2021. Her background is in Business Management and before working for ACD she was as a grant analyst and project manager in the Utah Public Health Lab Newborn Screening Informatics program. Heidi serves on the Utah Newborn Screening Advisory Committee, the Utah team of the Mountain States Regional Genetics Network, the ClinGen DAPC Working Group, and the ClinGen CCDS Variant Curation Expert Panel. Heidi's vision is that one day all creatine deficiencies will be diagnosed at birth, through routine newborn screening, and will be treated with an effective and appropriate treatment before the onset of symptoms.

The Association for Creatine Deficiency (ACD) is an international nonprofit organization dedicated to the three Cerebral Creatine Deficiency Syndromes: CTD, GAMT, AGAT.

CCDS INTERNATIONAL SYMPOSIUM - SPONSORS

CERES BRAIN THERAPEUTICS

ultragenyx pharmaceutical

Maladies rares Héréditaires du Métabolisme
Filière nationale de santé

DéfiScience
Maladies Rares du Neurodéveloppement
FILIÈRE NATIONALE DE SANTÉ

ape Association des Parents d'Elèves de l'Ecole Internationale Bilingue

CCDS INTERNATIONAL SYMPOSIUM - PARIS 2023

INFORMATIONS

TRANSFER FROM THE AIRPORT

Ride takes at least 60mn from Charles de Gaulle, depending on time of the day.

Taxi: you must take an official taxi at the airport (there is sign) in order to benefit from the flat rates for direct taxi rides between Paris and both airports.

From the airport to the hotel (right bank)

- €55 between Paris - Charles de Gaulle airport and Paris right bank;
- €41 between Paris-Orly airport and Paris right bank;

Bus. Roissy Bus from the Roissy Charles de Gaulle to Paris Opéra

Also Uber, Connectotransfers....

SYMPOSIUM VENUE

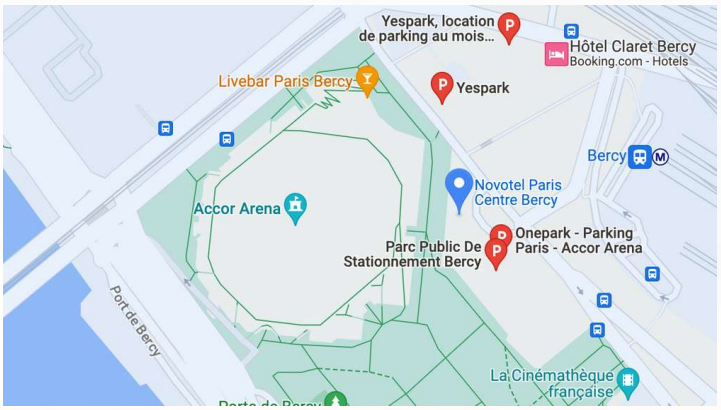
NOVOTEL BERCY

85 rue de Bercy

75012 Paris

métro :Bercy

tel: +33 1 43 42 30 00



CHILDCARE

drop off between 8.30am and 9.00am

pick up between lunch break from 12.30pm to 2.00pm

ends at 6.00pm on Friday and 4.00pm on Saturday

MEALS

Breakfast : for Novotel residents, in the main dining room from 7.00am (no room service available)

Buffet lunch in the main dining room between 12.30pm and 2.00pm

Gala dinner on Friday, please meet at 7.15pm in front of the hotel

CCDS INTERNATIONAL SYMPOSIUM - PARIS 2023

Association Xtraordinaire

www.xtraordinaire.org

Maladies rares
du neurodéveloppement
liées au chromosome X

Association de familles

Dis, Maman...
Je suis Xtraordinaire, c'est ça ?

Partageons
nos expériences
pour mieux les aider