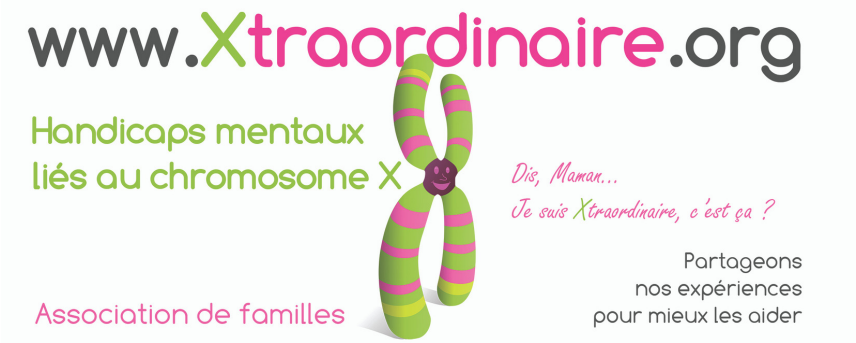
Inborn Cerebral Creatine Deficiency Syndromes Symposium

CTD GAMT AGAT

September 6-7th, 2019
Rotterdam, The Netherlands

SYMPOSIUM PROGRAM GUIDE

Thank you to Xtraordinaire, for organizing the 2019 Inborn Cerebral Creatine Deficiency Syndromes Symposium, and initiating this unique opportunity for the scientific and medical communities, the experts, and the patients' families to meet and exchange on CCDS.



A SPECIAL THANK YOU

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Symposium Agenda

Venue: De Doelen
Room: Van Weelde zaal
Address: Schouwburgplein 50 Rotterdam, Netherlands

Friday, September, 6th:

13.30	**Registration / Welcome Coffee** **@ Willem Burger Foyer**
	Introduction Session (15.30/16.00)
	Welcome by Gajja Salomons
15.30	**Cerebral Creatine Deficiency Syndromes and Diagnosis** *Gajja Salomons / Abel Thijs*
15.45	**Patient Perspectives** *Xtraordinaire / ACD*
	Session 1: Signs and Symptoms of Cerebral Creatine Deficiency Syndromes (16.00/19:30)
Chair:	**Carole Chehowah**
16.00	**Observational Study / Databases / Prevalence:** Vigilan: Observational Study of Male CTD Patients. *Judith Miller* Databases and Family Perspectives *Xtraordinaire, ACD* Prevalence in France & Netherlands *Aurore Curie / Gajja Salomons*
16.45	**The Basics of Behaviour:** **On Cognition, Development and Functioning.** *Sylvia Huisman*
17.30	Break
17.50	**Importance of Early Diagnosis: Feasibility of Newborn Screening for GAMT Deficiency.** *Marzia Pasquali*
18.15	**AGAT - The Therapeutical Target for GAMT Deficiency (and more?)** *Andreas Schulze*
18.40	**Brain Magnetic Resonance Spectroscopy and Creatine Measurements.** *Petra Pouwels*
19.05	**Q&As**
19.30	**Family Gathering and Discussion Time**
20.30	**Dinner Reception** **Stadbrasserie, Kruisstraat** (connected with de Doelen)

Saturday, September, 7th:

8.00 **Coffee @ Willem Burger Foyer**

9.00 **Creatine Transporter Deficiency in Females**
Jiddeke van de Kamp

Session 2: Animal Models for Cerebral Creatine Deficiency Syndromes (9.15/11.50)

Chair: **Jiddeke van de Kamp**

9.15 **Creatine Deficiency Mouse Models (AGAT, GAMT)**
Arend Heerschap

9.40 **A New Knock-in Rat Model of Creatine Transporter Deficiency**
Olivier Braissant

10.15 **Translational Phenotypes and Biomarkers of Brain Function for Creatine Transporter Deficiency**
Laura Baroncelli

10.40 Coffee break

11.00 **The Sum of All Parts? Effects of Neurotransmitter-Specific Crt Knockouts on Learning.**
Matthew Skelton

11.25 **CTD Mouse Model Experiences and Therapeutic Options.**
Ton de Grauw

11.50 **Lunch at Stadbrasserie,**
Kruisstraat
(Connected with de Doelen)

Session 3: Supplementation Treatment in Cerebral Creatine Deficiency and NBS (13.00/14.00)

Chair: **Monique Williams**

13.00 **Treatment Outcomes of Cerebral Creatine Deficiency Syndromes.**
Saadet Andrews

Epilepsy in Cerebral Creatine Deficiency Syndromes.
Saadet Andrews

13.40 ***Q&As***

Continued—Saturday, September, 7th:

Session 4: Development of New Treatment in Creatine Transporter Deficiency (14.00/18.00)

Chair: **Olivier Braissant**

14.00 **Drug Development for Neurodevelopmental Disorders: Lessons Learned from Fragile X Syndrome.**
Vincent des Portes / Aurore Curie.

Clinical Trials for Rare Disease: Current Pitfalls and New Perspectives.
Vincent des Portes / Aurore Curie.

14.40 **Novel Molecules for the Therapy of Creatine Transporter Deficiency**
Maurizio Balestrino

15.05 **Dodecyl Creatine Ester-Loaded Nanoemulsion as a Promising Therapy for Creatine Transporter Deficiency**
Aloise Mabondzo

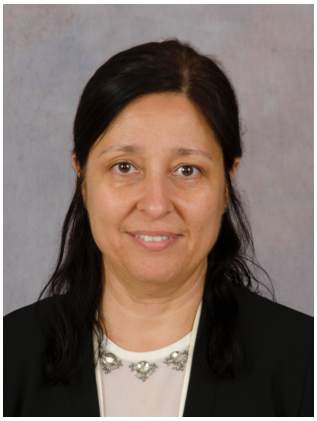
15.30 **Rescue by 4-Phenylbutyrate of Several Misfolded Creatine Transporter -1 Variants Linked to Creatine Transporter Deficiency Syndrome.**
Sonja Sucic

15.55 **Panel Discussion / Q&As**

16.30 **Families Debriefing**

17.30 **Farewell Reception @ StadBrasserie**

Dr. Saadet Andrews, MD, PhD, FCCMG, FRCPC



Dr. Andrews is a metabolic staff physician at The Hospital for Sick Children. She is an Associate Professor of Pediatrics at The University of Toronto. She finished medical school at the Aegean University, Izmir, Turkey. She completed her pediatric residency at the University of Istanbul, Turkey and Biochemical Genetics Fellowship, at the University of British Columbia, Vancouver, Canada. She finished her PhD“ Creatine deficiency syndrome: contributions to selective and newborn screening andtreatment“ at the Free University Amsterdam, The Netherlands. Her researchinterests include creatine deficiency disorders, pyridoxine dependent epilepsy,inherited neurotransmitter disorders.

Maurizio Balestrino

Maurizio Balestrino was born in Genova, Italy, the same city where Christopher Columbus was born. He received a Degree in Medicine and Surgery, with honors , in 1980 and a 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 learned 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 senior researcher in the Department of Neuroscience, Ophthalmology and Genetics of the University of Genova where 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 in these fields.

Laura Baroncelli

Dr. Baroncelli graduated in Biology from 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-yearpost-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. Recently, 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. Her scientific production was highly fruitful leading to the publication of 36 original research papers in international peer-reviewed journals (H index: 18). She was awarded personal fundings by Fondazione Roma, LUMOS Pharma, Italian Ministry of Health, Lejeune Foundation and Telethon for the study of creatine-related disorders. She is also Academic Editor of Neural Plasticity and Scientific Report, and reviewer for various international journals and national agencies.

Olivier Braissant, PhD

PhD thesis in biology, University of Lausanne 1990-1994 (Institute of Animal Biology, Prof W.Wahli; Molecular Biology/peroxisome proliferator activated receptors). Postdoc, University of Lausanne 1990-1994 (Institute of Animal Biology, Prof W.Wahli; Molecular Biology/ peroxisome proliferator activated receptors).

- Since 1997 at the Department of Clinical Chemistry (CDC), University
  - Research responsible: 1997-2004
  - Privat Docent & Master of Teaching and Research: 2004
  - Head of research unit: 2004-2009
  - Head of research section / Head of laboratory diagnosis: 2009
  - Associate Professor: 2019.

At CDC, 1997-2019: Head of the neurometabolic unit, working one IEM Affecting brain development, In Particular Deficiencies creatine, urea cycle diseases and toxicity of ammonium for CNS, and organic acidurias.

Aurore CURIE, MD, PhD

- Assistant Professor at the University of Lyon and at Femme-Mère-Enfant University Hospital, Hospice-Civils-de-Lyon, France
- Co-head of the Reference Center for Intellectual Disability from rare causes in Lyon, Hospice-Civils-de-Lyon, France
- Coordinator of the Inter University Diploma (DIU) on Neurodevelopmental Disorders
- Training: Masters in Molecular Biology, Cellular and Molecular Development Mechanisms, and in Neuroscience, Doctor in Medicine, PhD in Neuroscience, Post-doctorate at the Martinos Center for Biomedical Imaging (Harvard university, MIT, MGH), University Diploma on Interpretation of Clinical Trials.

Aurore Curie is a child neurologist (MD, PhD) at the Child Neurology Department of Lyon Hospital and the Reference Center for Intellectual Disability (ID) from rare causes. She is affiliated to the Institute of Cognitive Sciences Marc Jeannerod (ISC, CNRS UMR 5304) and also part of the DéfiScience national network for rare diseases of brain development and ID. 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 plateform “Cognitoscope” (ISC). 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.

Ton DeGrauw, MD, PhD
Professor of Pediatrics and Neurology
Emory School of Medicine

Antonius (Ton) DeGrauw, MD, PhD is a pediatric neurologist and Professor of Neurology and Pediatrics at Emory University. He graduated from the University of Nijmegen in the Netherlands and trained at Cincinnati Children’s Hospital. He was the Director of Neurology at Cincinnati Children’s from 1994-2011. From 2011-2017 he was Chief of Neurosciences at Children’s Healthcare of Atlanta and Director of Neurology in the Department of Pediatrics at Emory University School of Medicine. His research has involved the effects of neurometabolic disorders and epilepsy on the developing brain resulting in 100 peer reviewed publications.

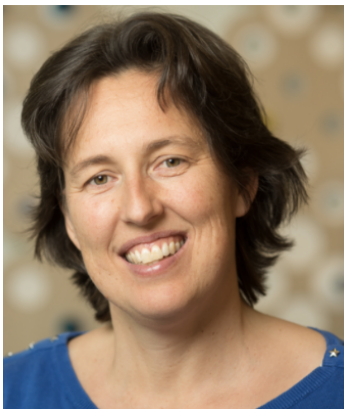
He has a longstanding interest in energy metabolism of the brain and specifically he has focused on creatine deficiency syndromes after he and his collaborators were the first to find several patients with Creatine Transporter (CrT) Deficiency in the early 2000’s. He was instrumental in developing CrT knockout model and showing that treatment for this condition may be possible.

Arend Heerschap
Full professor

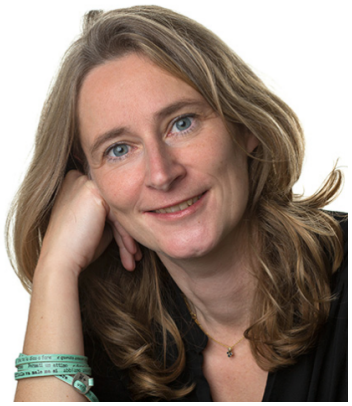
Arend Heerschap has a graduate degree in Biochemistry, did his PhD in Biophysical Chemistry on NMR in nucleic acids and worked for Philips Medical Systems before joining the Radiology department on research in Magnetic Resonance. Since 1998 he is full professor at this department and head of the Biomedical MR research group.

Research is focused on translational research, from in vitro NMR studies of tissues to MR of patients. Fundamental biomedical problems and the advancement of MR as a diagnostic tool are addressed. Main research topics are (1) Energy metabolism in skeletal muscle and brain and (2) Biology, diagnosis and treatment of cancer (prostate, brain, liver, etc). Using transgenic technology, vascular and cellular contrast agents, and natural abundance and labelled spin systems MR is applied in clinical research, diagnostics and in studies of animal models. High resolution and MAS NMR are applied in study of body fluids and biopsies. Dedicated MR systems for research on these objects have been implemented over the years.

Sylvia Huisman, MD, PhD
Zodiak, Prinsenstichting, Purmerend
Amsterdam Expertise Center for Neurodevelopmental Disorders, Amsterdam University Medical Center



Sylvia Huisman is a Physician for Individuals with Intellectual Disabilities (Dutch: AVG = Arts voor Verstandelijk Gehandicapten). Sylvia has a special interest in challenging behavior in people with intellectual disabilities and genetic syndromes and did her PhD on Self-injurious Behavior in Cornelia de Lange syndrome (CdLS). In her research she demonstrates that genetic entities are very interesting and promising to study behavior, and how detailed interdisciplinary phenotyping may lead to better understand pathogenetic molecular mechanisms. She works at the expert outpatient clinic Zodiak (Prinsenstichting), and at the Centre for Consultancy and Expertise (CCE). Sylvia is medical director of the expert clinic for adults with Cornelia de Lange syndrome, Rubinstein-Taybi syndrome, Pitt-Hopkins syndrome and Marshall-Smith syndrome at the Amsterdam Expertise Center for Neurodevelopmental Disorders (AECO) at the Amsterdam University Medical Center. She is involved in Waihonapedia, an online platform owned by patient organizations, to share information between families, professionals and researchers.



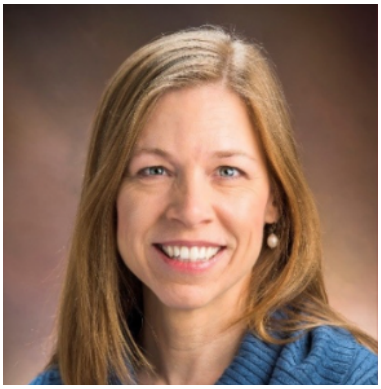
Jiddeke van de Kamp, MD, PhD
Consultant Clinical GeneticsAmsterdam University Medical Center, VU Medical Center, Amsterdam, Netherlands

Dr. Jiddeke van de Kamp is a consultant at the department of Clinical Genetics of the Amsterdam University Medical Center. She did her PhD 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.

Aloïse Mabondzo, HDR
CEA, Immunoanalysis and Pharmacology Department, Joliot Institute

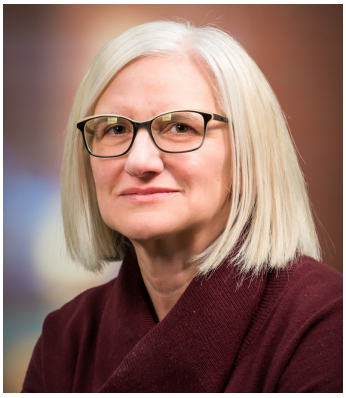
Aloïse Mabondzo, joined the CEA, the Life Science Division in May of 1998 as the leader of a Neurovascular Pharmacology Lab with a strong focus on in vitro blood-brain barrier models and pathophysiology of the brain.

Over the last 10 years, responsibilities have included managing of contractual studies (including interaction with industrial partners, protocol writing, GLP compliance, managing of technical personnel, writing expert reports), the tutoring of students and looking for research grants. He has conducted more than 100 contractual studies with industrial partners. Several PhD and post-doctoral positions have been part of his team. He has obtained numerous academic research grants. These financial supports have allowed and still make possible the development of research programs in the neuroscience field: drug transport-metabolism-the development of innovative cellular and animal model of Alzheimer's disease, nanotoxicology, X-linked creatine transporter deficiency disease. He is author or co-author of 52 articles in peer reviewed journals, 2 patients.



Judith Miller, PhD
clinical psychologist

Judith Miller, PhD, is a clinical psychologist with 25 years' experience in developmental disorders. She has a joint appointment as Assistant Professor in both the Psychiatry and Pediatrics departments at the Children's Hospital of Philadelphia (CHOP), which is affiliated with the Perelman School of Medicine at the University of Pennsylvania. She is also the Clinical Training Director at the Center for Autism Research, and the Autism Director for the Leadership in Education in Neurodevelopmental Disorders (LEND) program at CHOP. She was introduced to Creatine Transporter Deficiency (CTD) by Lumos Pharma, and was asked to help determine first signs, natural history, and possible outcome measures. She has recently published results on the first signs of CTD and how families came to an accurate diagnosis. She is currently the coordinating Principal Investigator for the Vigilan Observational Study.



Marzia Pasquali, PhD, FACMG
Medical Director, Biochemical Genetics and Newborn Screening Section Head, Biochemical Genetics

Dr. Pasquali is a professor of Pathology and Co-Director of the Fellowship training programin Clinical Biochemical Genetics at the University of Utah School of Medicine. Dr. Pasquali earned her degrees of doctor in pharmaceutical chemistry and technology and pharmacy doctor at the University of Parma School of Pharmacy. Dr. Pasquali trained in 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.

Pr Vincent des Portes, MD, PhD

Head of the department of child neurology, University Hospital HFME, Lyon, France
Professor of Pediatrics, Medical school Lyon Sud, University Lyon 1, France.

Training: MD (1994): Clinical training in Pediatrics (1989 – 1994) & Clinical Fellowship (1998_2001), APHP, Paris PhD (1994-1998): Genetics of X-Linked Intellectual disabilities, Pr J Chelly, Pr A Kahn, Cochin, France; Post-doctoral Position (2002): research fellow, The MIND Institute, Sacramento, Davis Univ. CA, USA. Prize from the French Academia of Science, 1998: human genetics and pathology (works on the DCX gene).

Teaching: Coordinator of two Inter University Diploma (DIU) in the field of NDD: Neuropsychologie Education Pédagogie; Déficience Intellectuelle. Scientific expert, START Project: translational Teaching in NDD (funded by CNSA, ARS IDF/ARA)

Research: Unit L2C2, Institute for Cognitive Science – Marc Jeannerod, (ISC-MJ), Lyon, France.

- Coordinator of a national grant, Ministry of Health (PHRC national 2008), still ongoing, that led to 15 publications «Neuronal Networks and cognitive processes in X-Linked Intellectual Disabilities».
- Investigator (PI for France) in 5 Multisite pharmacological trials in Fragile X Syndrome.

Publications: 148 citations in PubMed. SIGAPS: 1346 H index: 43 (Google Scholar)

Petra Pouwels, PhD

Assistant Professor, Department of Radiology and Nuclear MedicineAmsterdam UMC, location VUmc, Amsterdam, the Netherlands

Dr. Petra Pouwels is an NMR spectroscopist by training, and she works as MR physicist at the department of Radiology and Nuclear Medicine at Amsterdam UMC. After her PhD at Utrecht University in the field of high resolution NMR spectroscopy, she became a post-doctoral fellow in the field of human clinical MR Spectroscopy at the Max Planck Institute for biophysical chemistry in Göttingen, Germany (Prof. Jens Frahm).

Since then she is employed as MR physicist at Amsterdam UMC, location VUmc. She expanded her expertise of in vivo MR spectroscopy with several quantitative MRI techniques in neurology including structural MRI, functional MRI, and diffusion tensor imaging. She has a special interest in metabolic disorders, white matter disorders in children, and multiple sclerosis, and supervises all MR spectroscopy examinations both for research and clinical purposes.

Gajja Salomons, PhD
Professor of Molecular Biology of Neurometabolic Disorders
VU Medical Center, Netherlands
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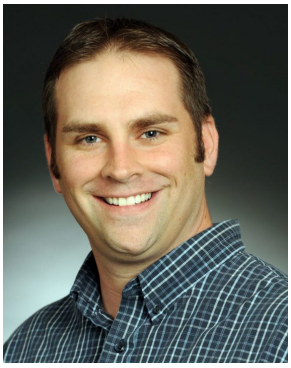
Dr. Gajja Salomons is a professor at the Department of Clinical Chemistry and also head of. the Metabolic Laboratory of the VU University Medical Center in Amsterdam, Netherlands. The laboratory provides diagnostics on inborn errors of metabolism on the following levels: metabolites, enzymes, DNA/RNA as well as on the functional level. Her research is embedded in the Neuroscience Campus Amsterdam and in the Center for Childhood White Matter Disorders led by Prof Marjo van der Knaap. The overall research aim of the Metabolic Laboratory is on unraveling the cause of neurometabolic disorders, development of diagnostic approaches and translational research. The main interests are the cerebral creatine deficiency syndromes, 2-hydroxyglutaric acidurias, disorders in polyol metabolism, and most recently on aminoacyl tRNA synthetase defects. Gajja Salomons and her team co-author on more than 170 peer-reviewed papers including papers reporting the underlying genetic defect of these disorders. Her laboratory functions as a worldwide referral center. Professor Salomons is honorary secretary of the Society for the Study of Inborn Errors of Metabolism and she is communicating editor of the Journal of Inherited Metabolic Disease.

Andreas Schulze, MD, PhD, FRCPC
Professor, Pediatrics and Biochemistry University of Toronto, Head of Metabolic Genetics Medical Director, Newborn Screening Program and Senior Associate Scientist at the Research Institute for the Hospital for Sick Children

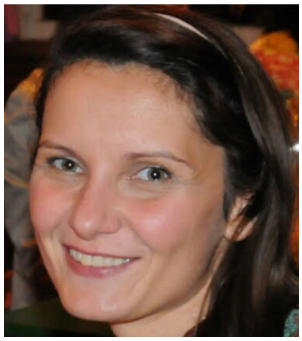
Dr. Schulze received his MD and PhD in Biochemistry at the University of Leipzig, Germany. He has worked at the Hospital for Sick Children in Toronto, Canada since 2007 and has established his own research group with a focus on Cerebral Creatine Deficiency Syndromes, Regulation of Creatine Synthesis, Pathophysiology of Guanidino Compounds, and Small Molecule Treatments. He is Director of the Newborn Screening Program at the Hospital for Sick Children, as well as the Section Head of Metabolic Genetics. Dr. Schulze was the first to report and describe the full biochemical spectrum in GAMT Deficiency and has an ongoing interest in advancing research of creatine deficiencies.



Matthew Skelton, PhD

Assistant Professor, Department of Pediatrics
University of Cincinnati Division of Neurology
Cincinnati Children's Hospital

Matthew Skelton earned his PhD in Developmental Biology from the University of Cincinnati and completed a Postdoctoral Fellowship with Cincinnati Children's Research Foundation. He is an Assistant Professor of Neurology at Cincinnati Children's. His dissertation work focused on the effects of developmental exposure to the drug of abuse 3,4+-methylenedioxymethamphetamine (commonly known as ecstasy). As a post-doctoral fellow in the lab of Dr. Michael Williams, Dr. Skelton developed and validated the behavioral phenotype of the Creatine Transporter (Crt) knockout mouse. This mouse is an invaluable tool that is used to answer important biological questions about CTD and to test potential therapies for this disorder. The focus of Dr. Skelton's work is to better understand what happens to brain cells that do not have creatine and how these cells contribute to the cognitive deficits observed in the mice. His lab uses a variety of techniques, from molecular biology to electrophysiology to behavioral testing, to answer these questions. He has been NIH funded and authored over 5 peer-reviewed manuscripts



Sonja Sucic

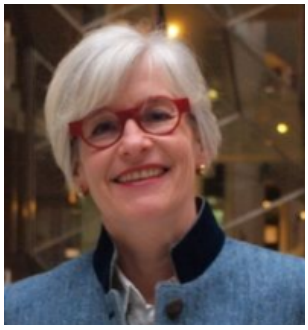
Associate Professor at the Institute of Pharmacology, Medical University of Vienna. PhD in the field of Pharmacology, at the School of Biomedical Sciences , University of Queensland, Brisbane, Australia. Advisor: Associate Professor Lesley Bryan-Lluka. Title: "The importance of the second transmembrane domain and the first intracellular loop in noradrenaline transporter function". Sonja ´s research focuses on solute carrier 6 (SLC6) transporters: their protein folding, ER export/trafficking mechanisms and pharmacological rescue of misfolded variants linked to various pathological conditions (e.g. infantile/juvenile Parkinson´s disease and dystonia, creatine transporter deficiency and epilepsy). In addition, other ongoing projects in the lab investigate molecular structure-function relationships of monoamine neurotransmitter transporters, in particular their interactions with antidepressant and psychostimulant drugs.

Abel Thijs, MD, PhD

Abel Thijs MD, PhD, is a specialist in internal medicine, with special interest in vascular medicine and inborn errors of metabolism. He is a member of the creatine team of AmsterdamUMC. He is a deputy head of the department of internal medicine and director of the residency program on internal medicine of this medical center.

Monique Williams, MD, PhD

Pediatrician, Center for lysosomal and metabolic disorders, Sophia Children's Hospital ErasmusMC, Rotterdam, The Netherlands Expertise in Organic acidurias, and Urea cycle disorder, Porphyria and Vascular Lipid disorders. Spokeswoman of EIMD scientific board. Member of the Neonatal Screening Advisory Board for metabolic disorders (ANS-MZ) in The Netherlands. Member of the Creatine Expertise Center AmsterdamUMC.



Dr. Williams received her MD and PhD at Leyden University (1994). Training as a Pediatrician and Metabolic specialty in Rotterdam (2003) and Paris(2004-JM Saudubray). Initially secondary creatine deficiencies drew her attention, her work at the laboratory Metabolic Laboratory of the VU University Medical Center in Amsterdam (2014-2019), together with the upcoming expansion of neonatal screening for GAMT deficiency led to membership of the Creatine Expertise Center AmsterdamUMC (previously: VUmc). Together with Dr. M Mulder (metabolic colleague-AmsterdamUMC) medical and care guidelines are being established for GAMT deficiency, initiated by the VKS (Dutch Metabolic Patient Organization). Dr. Williams is physician to a number of GAMT deficient patients and co-author of publications on GAMT deficiency.

Jnana Therapeutics

www.jnanatx.com

Jnana Therapeutics is pushing the frontiers of knowledge and technology to decipher the language of cellular metabolite communication and unlock the solute carrier (SLC) transporters as a therapeutic target class. We are focused on advancing differentiated therapeutics for Crohn's disease and ulcerative colitis that act by modulating guy barrier function or innate immune pathways. In parallel, we are pursuing therapeutics modulating novel SLC transporter targets in other diseases of great unmet need, including first-in-class therapies for currently untreated monogenetic neurological disease which includes Creatine Transporter Deficiency (CTD).

Ultragenyx

www.ultragenyx.com.

Ultragenyx is a biopharmaceutical company committed to bringing to patients novel products for the treatment of rare and ultra-rare diseases, with a focus on serious, debilitating genetic diseases. Founded in 2010, the company has rapidly built a diverse portfolio of approved therapies and product candidates aimed at addressing diseases with high unmet medical need and clear biology, for which there are no approved therapies.The company is led by a management team experienced in the development and commercialization of rare disease therapeutics. Ultragenyx's strategy is predicated upon time and cost-efficient drug development, with the goal of delivering safe and effective therapies to patients with the utmost urgency.

Xtraordinaire

www.xtraordinaire.org

Xtraordinaire is a French-based non-profit association founded in 2006 by families (parents of extraordinary children) dedicated to X-linked intellectual disability rare diseases. Xtraordinaire's main goals are to assist patient families, foster solidarity between families, organize events, work closely with the experts and research teams, participate in research programs, raise funds, and improve awareness and visibility on rare diseases.

The Creatine Transporter Deficiency branch is the most active within Xtraordinaire, and represents 60+ families.

Association for Creatine Deficiencies

www.creatineinfo.org

The Association for Creatine Deficiencies (ACD) is a non-profit 501(c)(3), founded in 2012, whose aim is to eliminate the challenges for patients and families living with a Cerebral Creatine Deficiency Syndrome through disease education and patient advocacy. Their mission is to educate and raise awareness of CCDS among the medical community, as well as the general public, and to advocate on behalf of families and patients living with CCDS. ACD has 400+ members in their support group, 20+ volunteers, and eight international scientists and researchers that are part of their Scientific and Medical Advisory Board.