

Conference Agenda

Thursday, July 11th

9:00 AM	REGISTRATION BEGINS		
12:15 PM	group photo for children/presenters (meet in registration area)		
12:30-12:45	welcome and introductions		
12:45-1:45	general assembly presentation Dr. Juan Pascual: University of Texas Southwestern research updates Room A		
1:45-2:00	break and transition time		
2:00-2:45	small group breakout session 1 (choose one)		
	Dr. Juan Pascual and Dr. Charles Roe: present and future clinical trials, patient registry Room A	Dr. Jörg Klepper: Glut1 Deficiency in early childhood B	Kris Engelstad and Dr. Veronica Hinton: Glut1 Deficiency in tweens, teens, and young adults C
	meet the experts: Dr. Jackie Montes, Dr. Cigdem Akman, Stacey Bessone		
2:45-3:00	break and transition time		
3:00-3:45	small group breakout session 2 (choose one)		
	Dr. Eric Kossoff: ketogenic diet therapies and their applications Room A	Dr. Dominic D'Agostino and Dr. Raffaele Pilla: ketone ester research B	CoRDS: Liz Donohue international rare disease patient registry C
	meet the experts: Dr. Veronica Hinton, Kris Engelstad		
3:45-4:00	break and transition time		
4:00-5:00 webcast time slot	general assembly presentation Dr. Jörg Klepper: global clinical research updates Room A		
5:00-6:00 webcast time slot	summary presentations from Dr. Pascual, Dr. Kossoff, and Drs. Montes/Hinton for the webcast audience: 15 minutes each, optional for attendees		
6:30 pm	social gathering: meet, greet, and eat! Family Photo Fun Booth!		



Conference Agenda

Friday, July 12th

8:30-9:00	foundation updates		
9:00-10:00	general assembly presentation Dr. Eric Kossoff: advances in the ketogenic diet		Room A
10:00-10:15	break and transition time		
10:15-11:00	small group breakout session 3 (choose one)		
Room A	Dr. Cigdem Akman: epilepsy in Glut1 Deficiency	Stacey Bessone, RD, LD: best practices for the ketogenic diet	Dr. Veronica Hinton: learning and behavior across the ages
	meet the experts: Dr. Juan Pascual, Dr. Charles Roe, Dorothy Kelly, Ana Hernandez, Dr. Jörg Klepper		
11:00-11:15	break and transition time		
11:15-12:00	small group breakout session 4 (choose one)		
Room A	Dr. Eric Kossoff: ketogenic diet therapies and their applications	Dr. Jörg Klepper: Glut1 Deficiency in early childhood	Kris Engelstad and Dr. Veronica Hinton: Glut1 Deficiency in tweens, teens, and young adults
	meet the experts: Dr. Dominic D'Agostino, Dr. Raffaele Pilla		
12:00-1:00	LUNCH BUFFET PROVIDED		
1:00-2:00	general assembly presentation Dr. Jackie Montes and Dr. Veronica Hinton: Columbia University research updates		Room A
2:00-2:15	break and transition time		
2:15-3:00	small group breakout session 5 (choose one)		
Room A	Dr. Juan Pascual: upcoming clinical trials and patient registry	Dr. Dominic D'Agostino and Dr. Raffaele Pilla: ketone ester research	Nutricia: cooking demonstration and recipe ideas
	meet the experts: Dr. Eric Kossoff & Stacey Bessone (team session), Dr. Cigdem Akman		
3:00-3:15	break and transition time		
3:15-4:00	small group breakout session 6 (choose one)		
Room A	Dr. Dominic D'Agostino and Dr. Raffaele Pilla: ketone ester research	Dr. Cigdem Akman: epilepsy in Glut1 Deficiency	Stacey Bessone, RD, LD: best practices for the ketogenic diet
	meet the experts: Dr. Jörg Klepper, Kris Engelstad, Dr. Veronica Hinton, Dr. Jackie Montes		
4:00-4:15	break and transition time		
4:15-5:00	small group breakout session 7 (choose one)		
Room A	Dr. Juan Pascual: upcoming clinical trials and patient registry	Dr. Eric Kossoff: ketogenic diet therapies and their applications	Dr. Veronica Hinton: learning and behavior across the ages
7:00-8:30	poolside social time		

Conference Offerings



Whole Group Presentations:

- Dr. Juan Pascual – UT Southwestern research updates
- Dr. Jörg Klepper – global clinical and research updates
- Dr. Eric Kossoff – ketogenic diet advances
- Dr. Jackie Montes and Dr. Veronica Hinton - Columbia research updates

Breakout Sessions:

- Dr. Juan Pascual and Dr. Charles Roe** – upcoming clinical trials and patient registry
- Dr. Eric Kossoff** – differences in ketogenic diet therapies and their applications
- Dr. Jörg Klepper** – Glut1 Deficiency in early childhood
- Kris Engelstad and Dr. Veronica Hinton** – Glut1 Deficiency in tweens, teens, and young adults
- Dr. Dominic D’Agostino and Dr. Raffele Pilla** – ketone ester research
- Dr. Veronica Hinton** – learning and behavior across the ages
- Dr. Cigdem Akman** – understanding the underlying mechanisms of epilepsy in Glut1 Deficiency
- Stacey Bessone, RD LD** – best practices for the ketogenic diet
- Nutricia/Ketocal** – cooking demonstration and recipe ideas
- CoRDS** – Coordination of Rare Diseases at Sanford: updates on international rare disease registry efforts

Meet the Experts:

time slots where experts will be available for individual discussions, Q & A

Other:

- Nutricia and CoRDS will be available throughout the conference with materials and Q & A
- A volunteer-staffed Children’s Activity Room will be available during all conference sessions
- A live webcast will be broadcast from 4:00-6:00 pm on Thursday evening

Glut1 Deficiency Foundation Conference Presenters



Dr. Cigdem Akman

Cigdem Inan Akman, MD
Director of Pediatric Epilepsy
Associate Professor of Clinical Neurology and Pediatric
Division of Child Neurology
Columbia University Medical Center
New York, New York



Dr. Akman is Associate Professor of Clinical Neurology and Pediatrics of the Division of Pediatric Neurology, Departments of Neurology and Pediatrics, College of Physicians & Surgeons of Columbia University.

Dr. Akman was born and educated in Ankara, Turkey where she graduated with Honors from the School of Medicine, Ankara University. Dr. Akman completed her training in Child Neurology at State University of New York; Clinical neurophysiology & Epilepsy at Boston Children's Hospital, Harvard Medical School. Subsequently, Dr Akman joined the Division of Pediatric Neurology and Pediatric Epilepsy at Columbia Presbyterian Medical Center and participated in clinical education and patients' care until 2008. Between 2008 and 2010, Dr Akman worked as the Director of Clinical Epilepsy Research at Texas Children's Hospital, Baylor College of Medicine. Dr Akman recently re-joined the Division of Pediatric Neurology and Epilepsy at Columbia as the Director of Pediatric Epilepsy.

Dr. Akman's clinical research has focused on cognitive and behavioral problems associated with epilepsy, imaging modalities used for epilepsy diagnosis and epilepsy surgery. Dr Akman also has published in the area of clinical neurophysiology to describe the specific EEG patterns in critically ill children. Her clinical practice focuses on infantile and childhood epilepsy; epilepsy surgery for the diagnosis of childhood refractory epilepsy.



Stacey Bessone, RD, LD

Stacey K. Bessone, RD, LD/N Ketogenic Dietitian
All Children's Hospital / Johns Hopkins Medicine, St Petersburg, Florida

Stacey is originally from Tampa, Florida. She is a graduate of Florida International University in Miami, Florida and has been a Registered Dietitian for almost 17 years. All of her clinical practice has been in the area of Pediatrics.

Currently, she is the Ketogenic Dietitian at All Children's Hospital in St Petersburg, Florida. Her practice encompasses inpatient diet initiations as well as in and out patient follow up consultations. Stacey covers the hospital based Ketogenic clinic in the Epileptology department. She also provides nutrition education for the Modified Atkins Diet and other diets under the Ketogenic Diet umbrella.

Stacey presents education on the Ketogenic Diet to all staff and residents at All Children's Hospital as well as to medical professionals in the community. Stacey presents education on the Ketogenic Diet to all staff and residents at All Children's Hospital as well as to medical professionals in the community.



Glut1 Deficiency Foundation Conference Presenters



Dr. Dominic D'Agostino

Dominic D'Agostino, PhD
Assistant Professor
Hyperbaric Biomedical Research Laboratory
Department of Molecular Pharmacology and Physiology
Morsani College of Medicine
University of South Florida
Tampa, Florida

Dr. D'Agostino's laboratory develops and tests metabolic therapies for seizures disorders, neurodegenerative diseases and cancer. They investigate the mechanism of these pathologies with a variety of in vivo and in vitro techniques, including radio-telemetry (EEG, EMG, ECG), electrophysiology, fluorescence microscopy, confocal microscopy, atomic force microscopy (AFM), biochemical assays and in vivo bioluminescence imaging. Current studies are focused on optimizing the anticonvulsant effects of the ketogenic diet, ketone esters and alternative fuels as a metabolic strategy to prevent seizures and enhance brain function.

2004-2006 Postdoctoral Fellow, Department of Neuroscience, Cell Biology and Physiology), Wright State University Boonshoft School of Medicine, Dayton, OH
2006-2008 Postdoctoral Fellow, Molecular Pharmacology and Physiology, Morsani College of Medicine; University of South Florida, Tampa FL
2008-present Assistant Professor, Molecular Pharmacology and Physiology, Morsani College of Medicine; University of South Florida, Tampa FL

Rutgers University, New Brunswick, NJ	BS	1994-1998	Biological Sciences, Nutrition
Rutgers University, Robert Wood Johnson Medical School, New Brunswick, NJ	Ph.D.	1999-2004	Physiology, Neuroscience



Dr. Darryl De Vivo

Darryl C. De Vivo, MD
Sidney Carter Professor of Neurology and Pediatrics
Co-Director, Motor Neuron Center
Associate Chairman for Pediatric Neurosciences and Developmental Neurobiology
Founding Director of the Colleen Giblin Research Laboratories for Pediatric Neurology
Columbia University Medical Center
New York, New York

Dr. De Vivo's planned presentation will be done by Dr. Veronica Hinton and Dr. Jacqueline Montes.

[Glut1 Deficiency Foundation Medical Advisory Board Member and Chair](#)

Dr. Darryl C. De Vivo, M.D., is the Sidney Carter Professor of Neurology, Professor of Pediatrics, and Director Emeritus (1979-2000) of the Pediatric Neurology Service at Columbia University Medical Center in New York City. He also is the Associate Chairman for Pediatric Neurosciences and Developmental Neurobiology, the Founding Director of the Colleen Giblin Research Laboratories for Pediatric Neurology at Columbia University Medical Center and the Co-Director of the Center for Motor Neuron Biology and Disease (MNC). Dr. De Vivo was a Director for Neurology and President of the American Board of Psychiatry and Neurology, Secretary of the American Academy of Neurology, and President of the Child Neurology Society. He completed postgraduate training in medicine, pediatrics, and neurology at the Massachusetts Memorial Hospital and the Massachusetts General Hospital, as well as at the National Institutes of Health where he was a Clinical Associate in Neurology. He has published more than 400 original articles and reviews, lectures extensively in the U.S. and abroad, serves on several editorial boards and advisory committees, and is a former associate editor for Rudolph's Textbook of Pediatrics. He is a current editor of "Neuromuscular disorders in Infancy, Childhood and Adolescence: A Clinician's Approach", recognized as the standard reference in the field.

Research Interests: Genetically-determined metabolic diseases that affect the developing brain and neuromuscular system. Specific disorders include Glut1 Deficiency, mitochondrial diseases, epilepsy, Duchenne muscular dystrophy, spinal muscular atrophy and other pediatric neuromuscular diseases.

Clinical Interests: Child neurology, neuromuscular disorders, neurometabolic disorders and neurogenetics.



Glut1 Deficiency Foundation Conference Presenters



Kris Engelstad

Kristin Engelstad, MS, CGC
Clinical Coordinator and Genetic Counselor
Department of Neurology
Columbia University Medical Center
New York, New York

[Glut1 Deficiency Foundation Medical Advisory Board Member](#)

Kristin Engelstad is a Clinical Coordinator and Genetic Counselor for Pediatric Neurology at Columbia University. She has been the coordinator for the Natural History Study of Glut-1 Deficiency Syndrome for over 10 years and serves as a liaison between the families, the local physician and the research team. She also coordinates Natural History Studies and Clinical trials for patients with mitochondrial disorders. She is involved with a contact registry and biobank for patients with mitochondrial disorders. Recently, she received a Masters Degree in Human Genetics and now serves as a Genetic Counselor for pediatric neurological disorders.

Ms. Engelstad has been involved with several publications in Glut1 Deficiency Syndrome and in mitochondrial disorders. She has worked closely with Dr. De Vivo for many years and has been privileged to work with and meet many families with a child with Glut1 Deficiency Syndrome.

Ms. Engelstad received her BS in Biology at the University of Iowa. She attended graduate school at Sarah Lawrence College in Bronxville, New York and received a Masters of Human Genetics. Her research interests include Glut1 Deficiency Syndrome and mitochondrial disorders.



Dr. Veronica Hinton

Veronica J Hinton, PhD.
Associate Professor of Clinical Neuropsychology
G.H. Sergievsky Center & Department of Neurology
Columbia University Medical Center
New York, New York

Veronica J. Hinton is an Associate Professor of Clinical Neuropsychology in the G.H. Sergievsky Center and the Department of Neurology. She is a developmental neuropsychologist who specializes in working with children with neurodevelopmental delays. Dr. Hinton is an active clinical researcher who has published on the neuropsychological profiles associated with a variety of pediatric disorders. Her primary research interests include examining the neurobehavioral profile associated with disorders of known genetic etiology. Dr. Hinton has worked closely with Dr. De Vivo for many years and has been privileged to work with and meet many families with a child with Glut1 Deficiency Syndrome.

Dr. Hinton received her BS in Biology and Psychology at Columbia University. She attended graduate school at the City University of New York, and received a PhD in Psychology with a specialization in Neuropsychology in 1994. During her graduate training, she was awarded a fellowship at the Center for Developmental Neuroscience affiliated with the New York State Institute for Basic Research in Developmental Disabilities. Dr. Hinton completed a clinical internship in neuropsychology at Hillside Hospital and the Long Island Jewish Hospital in Hillside, NY, and post-doctoral training in developmental psychobiology at Columbia University and the New York State Psychiatric Institute. She was appointed to the faculty of the Sergievsky Center and the Department of Neurology of Columbia University in 1996.

Glut1 Deficiency Foundation Conference Presenters



Dr. Eric Kossoff

Eric H. Kossoff, M.D.
Associate Professor, Neurology and Pediatrics
Medical Director, Ketogenic Diet Center
Johns Hopkins Hospital
Baltimore, Maryland

[Glut1 Deficiency Foundation Medical Advisory Board Member](#)

Dr. Kossoff is originally from New York and received his medical degree from SUNY at Buffalo School of Medicine. He went on to complete a residency in Pediatrics at Eastern Virginia Medical School in Norfolk, Virginia. He completed a second residency in Child Neurology and a fellowship in Pediatric Epilepsy at The Johns Hopkins Hospital in Baltimore.

Now an Associate Professor of Neurology and Pediatrics at Johns Hopkins Hospital, Dr. Kossoff focuses on the diagnosis and treatment of childhood seizures and epilepsy, particularly treatments other than medications such as diet, electrical brain stimulation, and surgery. Specific interests include the ketogenic diet, the modified Atkins diet for children and adults (created in 2003), infantile spasms, hemispherectomy, the interaction of migraine headaches with epilepsy, and Sturge-Weber syndrome.

Dr. Kossoff is also very involved in teaching and is the Director of the Pediatric Neurology Residency Program at Johns Hopkins. He is a coauthor of the books: Treatment of Pediatric Neurologic Disorders and the 5th edition of Ketogenic Diets.



Dr. Jörg Klepper

PD Dr. med. Jörg Klepper
Children's Hospital Aschaffenburg
Klinik für Kinder- und Jugendmedizin
Aschaffenburg, Germany

[Glut1 Deficiency Foundation Medical Advisory Board Member](#)

Dr. Klepper is Head of the Department of Child and Adolescent Medicine at the Children's Hospital Aschaffenburg in Aschaffenburg, Germany.

Medical degree	Frankfurt and Würzburg University, Germany
Paediatric training	Würzburg und Essen University, Germany
Postdoc fellowship	Columbia University, New York, USA
Consultant for paediatric neurology	Essen University, Germany
Lecturer for Paediatrics	Essen University Würzburg University

Dr. Klepper's main fields of work are disorders of brain energy metabolism such as GLUT1 deficiency syndrome or pyruvate dehydrogenase deficiency. Both disorders are associated with intractable epilepsy and respond to the ketogenic diet. Research interests are mechanisms and adverse effects of the diet, as well as establishing international protocols for the use of the diet in epilepsy and metabolic disorders.

Glut1 Deficiency Foundation Conference Presenters



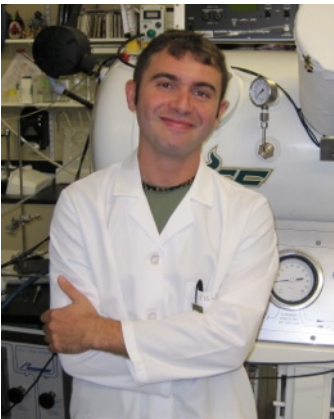
Dr. Jacqueline Montes

Jacqueline Montes, PT, EdD, NCS
Researcher, Lecturer, Physical Therapist
Spinal Muscular Atrophy Clinical Research Center
Department of Neurology
Columbia University Medical Center
New York, New York

Jacqueline Montes, PT, EdD, NCS obtained her Bachelors degree and physical therapy certification at Hunter College here in New York City. She also received a Masters of the Arts (MA) and a Doctorate of Education (EdD) in Motor Learning and Motor Control from Columbia University's Teachers College. She has been a certified Neurologic Clinical Specialist by the American Board of Physical Therapy Specialties since 2005.

Dr. Montes has worked at the Columbia University Medical Center since 1994 first joining the Physical Therapy Department rotating through specialties such as Pulmonary, Orthopedics and Wheelchair Clinic. In 1997 her focus and specialization became the care of acute neurological and neurosurgical patients. In 2003, Dr. Montes joined the multidisciplinary team at the Eleanor and Lou Gehrig MDA ALS Research Center at Columbia University. Her work included evaluating and treating patients with motor neuron disease, as well as coordinating their care at home, rehabilitative services, durable medical equipment and mobility aids. She joined the SMA research team and pediatric neuromuscular clinic in 2007.

Dr. Montes has acted as coordinator and/or evaluator for phase I, II and III clinical and observational trials. She has also led numerous research projects, including industry and NIH-sponsored single and multi-center clinical studies. She has acted as lead evaluator for multi-center trials, training physical therapists from across the nation on obtaining outcome measures, and she has provided ongoing training and quality control to evaluators from other institutions. Her work has resulted in publications and presentations at international meetings. She has authored manuscripts and abstracts on outcome measure validation, weakness, falls, and fatigue.



Dr. Raffaele Pilla

Dept of Molecular Pharmacology and Physiology
Morsani College of Medicine
University of South Florida
Tampa, Florida

Dept. of Medicine and Surgery
University of Salerno
Baronissi (Salerno) - Italy

Service of Oncology
Public Hospital Fatebenefratelli
Benevento - Italy

Raffaele Pilla, Pharm.D., Ph.D., Doctor Europaeus, received his Master's degree in Pharmacy at G. d'Annunzio University in Chieti-Pescara, Italy in 2005, where he also served internships at the Cell Physiology Laboratory and Molecular Biology Laboratory. Prior to his degree, he was an Erasmus Student at Faculté de Pharmacie de Reims in Reims, France. He received his Doctor Europaeus in 2010 from Pitié-Salpêtrière Institute in Paris, France. Also in 2010, he received his Ph.D. in Biochemistry, Physiology, and Pathology of Muscle at G. d'Annunzio University in Chieti-Pescara, Italy. He was hired as a Postdoctoral Scholar in the Department of Pharmacology and Physiology at the University of South Florida in Tampa, on two research grants respectively funded by the Office of Naval Research (US Navy) and Divers' Alert Network. He has written and lectured widely on his experiences.

Dr. Pilla has been involved in ongoing research at the University of South Florida with the use of ketone esters. The initial work, funded by the Office of Naval Research, focused on using ketone esters to prevent CNS oxygen toxicity. Dr. Pilla is working to coordinate efforts in the United States and Italy to expand the ketone ester research to other conditions such as epilepsy and cancer and its use as an alternative metabolic fuel.

Dr. Pilla has also served extensively in various international medical missions, orphan and homeless programs, emergency medicine rescue and training, and animal rescue and fostering. He is fluent in Italian, English, French, and Spanish. He has a special interest in journalism and photography, and he enjoys sailing, SCUBA diving, soccer, volleyball, jogging, and motorcycles.



Glut1 Deficiency Foundation Conference Presenters



Dr. Juan Pascual

Director and Principal Investigator
Rare Brain Disorders Clinic and Laboratory
Department of Neurology and Neurotherapeutics
UT Southwestern Medical Center
Dallas, Texas

Glut1 Deficiency Foundation Medical Advisory Board Member

Juan M. Pascual holds appointments to the faculty of the Departments of Neurology and Neurotherapeutics, Physiology and Pediatrics and is Director of the Rare Brain Disorders Clinic and Laboratory. He is also a member of the Division of Pediatric Neurology, of the graduate programs in Neuroscience and Integrative Biology, and of the postgraduate clinical training programs in Neurology, Pediatric Neurology, Pediatrics and Medical Genetics.

Dr. Pascual received his M.D. degree from the Universidad de Granada, Spain (founded in 1526 by Emperor Charles V over the school established by the Nasrid Sultan of Granada in 1349). He received his Ph.D. degree in Molecular Physiology and Biophysics from Baylor College of Medicine in Houston, Texas. His postdoctoral research was conducted at the Center for Molecular Recognition, College of Physicians and Surgeons of Columbia University and, later, at the Colleen Giblin Research Laboratories for Pediatric Neurology at the same institution. He also obtained residency training in Pediatrics at Washington University School of Medicine- St. Louis Children's Hospital and in Neurology and Pediatric Neurology at the Neurological Institute of New York- Columbia University Medical Center. He received certification in Neurology with Special Qualification in Child Neurology from the American Board of Psychiatry and Neurology.

Research Interests

- Brain metabolism
- Clinical trials for neurological and genetic disorders
- Complex, rare and undiagnosed diseases
- Functional brain imaging
- Human brain development
- Mitochondrial disorders
- Neural excitability: synaptic transmission
- Neurodegenerative diseases of children
- Neurogenetics
- Sentience, autonomy and sources of truth

Clinical Interests

- Complex, Rare and Undiagnosed Brain Disorders
- Mitochondrial Disorders
- Neurodegenerative Diseases of Children
- Neurogenetics
- Neurological and Genetic Disorders



Dr. Charles Roe

Clinical Professor and Investigator
Department of Neurology and Neurotherapeutics
UT Southwestern Medical Center
Dallas, Texas

Dr. Roe was a professor of pediatrics and chief of the division of "Genetics and Metabolism" at Duke University Medical Center for 25 years until 1995. He was then the Medical Director and developed the Institute of Metabolic Disease at Baylor University Medical Center from 1995 until 2009. He has been a member of the Department of Neurology and Neurotherapeutics at UT Southwestern Medical Center in Dallas since 2009. During his career, he also studied in the Department of Biochemistry at Brandeis University for 2 years.

He has had more than 40 years of experience in Clinical Investigations with a long-term relationship with the U.S. Food and Drug Administration (FDA), and has authored over 200 peer-reviewed manuscripts and book chapters. He has spent the last 20 years developing and applying the concept of "anaplerotic diet therapy" using triheptanoin with patients and in animal studies.



Glut1 Deficiency Foundation Conference Presenters



Nutricia, the makers of KetoCal, specializes in the delivery of advanced medical nutrition. Nutricia representatives will conduct a cooking demonstration, supply product samples, share recipe ideas, and be available throughout the conference to answer questions.



The Coordination of Rare Diseases at Sanford registry is an international and central registry of individuals of any age who have been diagnosed with a rare disease or an uncommon disease with unknown prevalence (undiagnosed individuals are also eligible for enrollment).

The registry collects basic contact and diagnostic information from participants. Any researcher with the appropriate approval can request de-identified information from the registry that might be helpful to the research they are conducting.

By enrolling in CoRDS, participants will be notified of opportunities to participate in clinical trials and other research. Liz Donohue, Director, will present information on the registry efforts and will be available throughout the conference to answer questions and enroll patients.



Glut1 Deficiency Foundation Conference Exhibitors



Innovation in Nutrition

