CCDS Scientific + Patient SYMPOSIUM

ACD ASSOCIATION FOR CREATINE DEFICIENCIES

2022 CCDS Scientific & Patient Symposium

Program Agenda

Sunday, June 26th

All times below are in Mountain Time

7:00 - 8:00 AM	**Breakfast**
8:00 - 8:30 AM	Kids Camp Dropoff
8:30 - 8:45 AM	Opening Remarks and Session Overview, Sangeetha Iyer
	Session I: Advances in therapeutic development for CCDS
8:45 - 9:00 AM	Identifying the mechanism by which creatine represses expression of AGAT, Alex Lee
9:00 - 9:15 AM	Towards the Discovery of Small Molecules that Restore Function to Defective SLC6A8 Creatine Transporter Variants, Charles Kuntz
9:15 - 9:30 AM	A high throughput screening assay for CTD drug discovery, Manuel Cernigoj
9:30 - 9:45 AM	Modeling CTD using Brain Organoids Reveals Molecular Targets Associated with Inherited Intellectual Deficiency and Acquired Neurodegenerative Diseases, Léa Broca-Brisson
9:45 - 10:15 AM	Q&A/Coffee Break
	Session 2: Computational and other approaches to understanding patient mutations
10:15 - 10:30 AM (Virtual)	Integrating computational and experimental evidence for the characterization of single amino acid variants of the creatine transporter (SLC6A8), Evandro Ferrada
10:30 - 10:45 AM (Virtual)	Discovery of Small Molecules that Restore Function to Defective SLC6A8 Creatine Transporter Variants, Peter Axerio-Cilies
10:45 - 11:00 AM (Virtual)	In vitro effectiveness of a prodrug of the creatine precursor guanidinoacetic acid against block of the creatine transporter, Maurizio Balestrino
11:00 - 11:15 AM	Q&A/Coffee Break
11:15 - 12:00 PM	**Keynote Session: Combined Brain, Terry Jo Bichell**
12:00 - 1:00 PM	**Lunch**
	Session 3: Exploring models of CCDS
1:00 - 1:15 PM	Small molecule therapy for GAMT deficiency, Nicola Longo
1:15 - 1:30 PM	Mice with a P544L mutation in the Slc6a8 gene have reduced brain Cr, spatial learning deficits, and impaired nest building, Matthew Skelton
1:30 - 1:45 PM (Virtual)	Creatine transporter deficiency impairs stress adaptation and brain energetics homeostasis, Hong-Ru Chen
1:45 - 2:00 PM	Q&A/Coffee Break

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	Session 4: Towards Clinical Trials for CCDS
2:00 - 2:15 PM	For a Clinical gene therapy trial for Creatine deficiency disorders in future: What parents shall be aware of?, Jagdeep Walia
2:15 - 3:00 PM	Clinical trial and outcomes: A Panel discussion Panelists: Melanie Brandabur, Andreas Schulze, Saadet Andrews, and Aurore Curie
3:00 - 3:15 PM	ACD's Vision for the Future, Sangeetha Iyer
3:15 - 4:00 PM	**Coffee and Connections**
4:00 - 5:00 PM	Special ACD Warrior of Hope Session, Chris Nikic - "1% Better - The Road to Full Potential"
5:30 - 9:00 PM	SuperHero Celebration (Drinks 5:30, Dinner & Entertainment 6:00-9:00)

Monday, June 27th

7:00 - 8:00 AM	**Breakfast**
8:00 - 8:30 AM	Kids Camp Dropoff
8:30 - 8:45 AM	Evolving landscape of newborn screening for creatine deficiency syndromes, Marzia Pasquali
	Session 5: Gene Therapy Approaches for CCDS
8:45 - 9:00 AM (Virtual)	Females' characterization in the Slc6a8Y389C rat model of CTD, Lara Duran-Trio
9:00 - 9:15 AM (Virtual)	AAV strategy to treat creatine transporter deficiency in the Slc6a8Y389C/y KI rat, Gabriella Fernandes-Pires
9:15 - 9:30 AM (Virtual)	Creatine Transporter Deficiency: the long journey to successful therapy, Laura Baroncelli
9:30 - 9:45 AM	AAV-Based Gene Therapy Restores Cerebral Creatine, Reduces Guanidinoacetate, and Resolves of Behavioral Abnormalities in a Mouse Model of GAMT Deficiency, Gerald Lipshutz
9:45 - 10:15 AM	Q&A/Coffee Break
	Session 6: Diagnosis, genotypic and clinical features of CCDS patients
10:15 - 10:30 AM	Recognizing Females with Creatine Transporter Deficiency Syndrome, Kim Cecil
10:30 - 10:45 AM	Universal GAMT Newborn Screening in New York State: The First Three Years, Denise Kay
10:45 - 11:00 AM	The ClinGen Variant Curation Expert Panel for Cerebral Creatine Deficiency Disorders: Classifying the clinical significance of variants in GATM, GAMT, and SLC6A8, Jenny Goldstein
11:00 - 11:15 AM	Q&A/Coffee Break

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11:15 - 11:30 AM	ACD Special Interest Session: Probably Genetic, Abby D'Cruz
11:30 - 11:45 AM	ACD Special Interest Session: Xtraordinaire - Focus on CTD Families in France, Carole Chehowah
11:45 - 1:00 PM	**Lunch**
	Session 7: Learnings from CCDS Patient families
1:00 - 1:15 PM	Creatine Transporter Deficiency: bridging the gap between science and society, Kim Soesbergen
1:15 - 1:30 PM	Measuring family adjustments and accommodations in Intellectual and Developmental Disorders, Judith Miller
1:30 - 1:45 PM	Oral medications and creatine transporter deficiency (CTD): Impact of clinical symptoms and perspectives of caregivers and clinicians, Melanie Brandabur
1:45 - 2:00 PM	The CreatineInfo Registry: Demographics and Initial Findings, Laura Voss
2:00 - 2:15 PM	Outcome measures for clinical trial in Creatine Transporter Deficiency, Aurore Curie
2:15 - 2:30 PM	Q&A/Coffee Break
2:30 - 3:00 PM	Patient Meaningful Outcomes for CCDS Part I and Part II, Sangeetha Iyer and Heidi Wallis
3:00 - 3:15 PM	ACD Closing Remarks: The year ahead and beyond, Heidi Wallis
3:15 - 4:00 PM	**Coffee and Connections**
4:00 - 5:00 PM	Parent and Caregiver Focus Session: Practical Behavior Management Strategies; Sarah Buchanan, Carmen B. Pingree
5:30 - 9:00 PM	Awards Dinner (Drinks 5:30, Dinner & Entertainment 6:00-9:00)