# ACD Awards $145,000 to Advance Creatine Deficiency Research at Six Research Centers

*Researchers at six institutions funded by Association for Creatine Deficiencies to research creatine transporter deficiency, GAMT deficiency and AGAT deficiency*

2025 ACD Fellowship Awardees

CARLSBAD, CA, UNITED STATES, February 12, 2025 /EINPresswire.com/ -- The Association for Creatine Deficiencies (ACD) is pleased to announce the funding of six new research projects. Five 2025 ACD Fellowship Awards will provide one year of funding to support early-career researchers whose projects aim to advance scientific understanding and therapeutic development for Cerebral Creatine Deficiency Syndromes (CCDS). An additional grant, co-funded by Uplifting Athletes, builds upon the advancements of a prior ACD fellowship grant in the Schlebach lab at Purdue University. In total, ACD has awarded $145,000 to support these research projects.

The researchers whose projects are funded include:

> These exceptional projects represent crucial steps forward in CCDS research, with the potential to directly impact the development of future treatments."
>
> *Heidi Wallis*

ACD Fellow, Dr. Israel Abebe Admasu, Boston Children's Hospital (In the lab of Dr. Michela Fagiolini)
Dr. Admasu's work addresses the need for precise tools to evaluate emerging therapies for CCDS. His project establishes a non-invasive method to measure brain signals as biomarkers of disease severity, providing objective assessments for new treatments. "I believe that my research will establish a critical framework for evaluating the potential impact and efficacy of upcoming therapeutic approaches targeting creatine deficiency

syndromes," said Dr. Admasu.

ACD Fellow, Aleksander Bogoniewski, UCLA (In the lab of Dr. Gerald Lipshutz)
Bogoniewski's research employs human neurons and brain organoids to investigate the neurodevelopmental impact of CCDS mutations. Additionally, his work evaluates adeno-associated virus (AAV) gene therapies and compares them to existing treatments. "I believe our work will advance our understanding of CCDS by exploring neurodevelopment through a humanized model system. Furthermore, our research on gene addition and the evaluation of current clinical treatments has the potential to drive the development of new therapeutic methodologies," he explained.

2025 Young Investigator Awardee

ACD Fellow, Dr. Chin-Yi Chen, Virginia Tech (In the lab of Dr. Cheng-Chia Wu)
Dr. Chen's work explores the use of focused ultrasound (FUS) to enhance creatine delivery in patients with Creatine Transporter Deficiency, under the mentorship of Dr. Wu at Virginia Tech and Dr. Seth Berger at Children's National Hospital. Using both patient-derived cells and a mouse model, her research aims to improve creatine uptake in the brain. "I am deeply committed to investigating the molecular mechanisms underlying rare neurological diseases, with a particular focus on accelerating treatments for these conditions," she said.

ACD Fellow, Alex Edwin, Stanford University (In the lab of Dr. Thomas Montine)
Edwin's research focuses on the development of a small-molecule prodrug therapy for creatine transporter deficiency. His approach chemically modifies creatine to allow it to enter neurons independently of the impaired transporter, where it is enzymatically released. "Our research will provide a strong foundation towards identifying an effective therapeutic for creatine transporter deficiency," he stated.

ACD Fellow, Tesla Peretti, Queen's University (In the lab of Dr. Jagdeep Walia)
Peretti's research focuses on gene replacement therapy for AGAT deficiency and the safety evaluation of a promising GAMT deficiency treatment. Her project aims to restore creatine synthesis in the brain and assess toxicity before advancing to clinical trials. "I believe my work will be an important step in understanding CCDS, specifically AGAT and GAMT deficiencies, and

bring us closer to discovering a potential cure through gene replacement therapy," she stated.

ACD and Uplifting Athletes co-funded Young Investigator, Dr. Charles Kuntz, Purdue University (In the lab of Dr. Jonathan Schlebach)
Dr. Kuntz is a dedicated researcher in the Schlebach lab. This lab has been at the forefront of studying membrane protein folding and expression, with a focus on understanding and identifying small molecule correctors that stabilize these critical structures. Through innovative in silico screening, they have successfully identified correctors that stabilize various conformations of the SLC6A8 transporter. Currently, they are validating these findings in the lab using engineered cell lines expressing patient mutations. This groundbreaking work holds the promise of discovering novel correctors that could stabilize mutant SLC6A8 transporters, paving the way for potential treatments for this disorder.

"These exceptional projects represent crucial steps forward in CCDS research, with the potential to directly impact the development of future treatments," said ACD Executive Director Heidi Wallis. "We are honored to support these talented researchers, whose work brings hope to families affected by these disorders."

The ACD Fellowship program funds high-potential research aimed at advancing CCDS treatment options. Many previous fellowship recipients have continued in CCDS research, expanding their work and securing additional funding to further develop their findings. Applications for the 2026 ACD Fellowship Grants will open in fall 2025. For more information about ACD's research funding opportunities, visit www.creatineinfo.org.

Uplifting Athletes is a nonprofit organization that unites student-athletes nationwide and professional athlete ambassadors with their local rare disease communities.

The Young Investigator Draft is inspired by the NFL Draft but shifts the focus from selecting potential talent on the football field to recognizing the next generation of promising young medical researchers in rare diseases. To learn more about Uplifting Athletes, visit www.upliftingathletes.org.

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About ACD: The Association for Creatine Deficiencies' mission is to eliminate the challenges of CCDS. ACD is committed to providing patient, family, and public education to advocate for early intervention through newborn screening, and to promote and fund medical research for treatments and cures for Cerebral Creatine Deficiency Syndromes. Because CCDS mimic symptoms of other medical conditions, patients are often first diagnosed with autism, cerebral palsy, epilepsy, and other disorders. Proper diagnosis and early intervention are critical to establishing interventions needed to improve life quality and longevity for the CCDS patient. For more information regarding ACD, please visit creatineinfo.org.

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