

Creatine Deficiency Research Center Funded by ACD Launches at University of Utah

CARLSBAD, CA, UNITED STATES, March 20, 2023 /EINPresswire.com/ -- The [Association for Creatine Deficiencies](#) (ACD) today announced the launch of the new Creatine Deficiency Research Center at the [University of Utah](#), which has been set up with funding support from ACD.

ACD has committed \$240,000 in 2023 funds toward this multi-year initiative at the University of Utah, which, in conjunction with [ARUP Laboratories](#), seeks to discover new diagnostic tools and therapies for Cerebral Creatine Deficiency Syndromes (CCDS), inborn errors of metabolism, which interrupt the formation or transport of creatine. Creatine is essential to sustain the high energy levels needed for muscle and brain development.



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“The launch of the Creatine Deficiency Research Center is a major milestone for our community and we are extremely pleased to see these important research projects move forward with full-time staff dedicated to working on solutions to treat and diagnose creatine deficiency syndromes,” said Heidi Wallis, ACD executive director. “This initiative at the University of Utah is being led by an outstanding team of scientists and we are very thankful for their dedication and commitment to helping improve the quality of life of those affected by creatine deficiencies.”

The Creatine Deficiency Research Center will be led by Marzia Pasquali, PhD, FACMG, section chief of ARUP’s Biochemical Genetics, and Nicola Longo, MD, PhD, a pediatric medical geneticist, chief of the Medical Genetics division at University of Utah Health, and a member of Biochemical Genetics at ARUP. They are joined by Steven Baker, MD, PhD, an associate medical director of Transfusion Medicine at ARUP who also does basic science research in the Department of Pathology at the University of Utah, and Filippo Ingoglia, PhD, ARUP medical director of

Biochemical Genetics.

CTD is caused by mutations in the SLC6A8 gene. While patients with CTD have the necessary AGAT and GAMT enzymes to form creatine, the creatine transporter does not function properly. This results in normal creatine in the bloodstream, but not in the brain and muscles. Individuals with CTD struggle with symptoms including intellectual disability, speech delay, seizures, behavioral challenges and low muscle tone. Many CTD patients are also diagnosed with autism. There is currently no proven therapy to treat CTD.

In late 2022, ACD launched a fundraising campaign with a goal to raise \$250,000, to fund the Creatine Deficiency Research Center among other research projects. This goal was exceeded, with a total of \$255,825 raised by January 2023, enabling the funding of this new groundbreaking research project. "A big part of our mission at ACD is to promote and fund medical research for treatments and cures for Cerebral Creatine Deficiency Syndromes, and we want to thank each and every one of our generous donors whose support has made it possible for us to put that mission into action as we move forward with the Creatine Deficiency Research Center," said Dan Coller, ACD board chair.

The Creatine Deficiency Research Center will support two full-time research scientists and will be overseen by four CCDS expert mentors who will engage in groundbreaking research projects aimed at treatment and diagnosis for creatine deficiencies, with the first project led by Baker, expected to begin in the summer of 2023, focused on testing methods such as enzyme therapy, gene therapy, and CRISPR editing to induce expression of the enzyme AGAT in neurons to permit the internal synthesis of creatine in the neurons themselves.

"Enabling all neurons to synthesize creatine may be the key to replenishing brain creatine levels in CTD patients," said Baker. "This research we are embarking on with the formation of the Creatine Deficiency Research Center has the potential to pave the way for development of a treatment for patients with CTD."

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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