

ACD Funds over \$100,000 for Creatine Deficiency Research Fellowships

CARLSBAD, CALIFORNIA, UNITED STATES, February 1, 2022 /EINPresswire.com/ -- Today, the [Association for Creatine Deficiencies \(ACD\)](#) announced the funding of a total of \$105,000 for early career researchers studying Cerebral Creatine Deficiency Syndromes (CCDS). The awards include continued funding for three fellowships totaling \$75,000 for researchers whose projects were initially funded last year and made progress in the first year, and one new fellowship for \$30,000.

The researchers whose projects were funded for the 2022-2023 funding period, to begin today, Feb. 1, 2022, include:

- \$30,000 each to continue collaborative Creatine Transporter Deficiency (CTD) drug repurposing precision medicine studies led by Dr. Peter Axerio-Cilies (in the lab of Dr. Sylvia Stockler at BC Children's Hospital Research Institute in Vancouver) and Dr. Charles Kuntz (in the lab of Dr. Jonathan Schleich at the University of Indiana, Bloomington)
- \$30,000 for Léa Broca-Brisson in collaboration with Dr. Aloise Mabondzo to develop organoids for CTD that allow for better disease modeling
- \$15,000 continued funding for Alex Lee (in the lab of Dr. Andreas Schulze) for basic science research to understand creatine mechanisms

The overarching goal of the ACD Fellowship program is to fund opportunities which have the potential for translational success, leading to clinical trials for creatine deficiency treatments.

Dr. Kuntz and Dr. Axerio-Cilies together with Dr. Schleich and Dr. Stockler are leading the way in identifying drugs that might rescue creatine uptake across different types of SLC6A8 mutations. The two fellowship projects complement each other in characterizing specific mutations and how they respond both in computer simulation and petri dish experiments to existing compounds. Through these studies, research is identifying potential treatments targeting specific genetic variants by identifying repurposable drugs that can restore functional activity (creatine uptake) of the dysfunctional SLC6A8 transporters. Results from the first year of these studies showed that there are existing compounds that may increase creatine transport in non-functional mutations. As the studies move forward in the second year, they aim to follow up on this finding by understanding the mechanism of rescue (expression or function), and by

collecting similar data on additional variants, followed by verifying this finding in patient fibroblasts bearing the same mutation. Additional information on Dr. Axerio-Cilies and Dr. Kuntz's drug repurposing projects is available in the December 2021 ACD [Creatine Decoded article](#).

ACD welcomes Broca-Brisson, PhD student at University Paris Saclay in France, as the newest recipient of an ACD fellowship award. Broca-Brisson's project is focused on development of human brain organoids from CTD patients as a tool for evaluating the efficacy of potential treatments. She also plans to identify measurable biomarkers as a standard for measuring the success of treatments, particularly in relation to improvement of cognitive impairments.

Lee, a graduate student in Dr. Andreas Schulze's lab, will continue his work on understanding regulatory mechanisms for genes critical to the creatine synthesis and transport pathway.

"ACD is funding ground-breaking research in creatine deficiencies. The outcome of these projects may help inform future research and even lead to treatments for CTD. We are grateful for the financial support of our donors which makes these initiatives possible and helps us move the needle towards better outcomes for all patients with CCDS," said ACD Board Chair and Director of Research Laura Trutoiu, Ph.D. "We are also grateful to and welcome the research fellows that are joining our community and working alongside us in the search for cures."

In 2021, ACD announced a five-year vision to invest \$5 million in CCDS research with the aim of having five Investigational New Drugs (INDs) for creatine deficiencies in progress with the FDA by 2026. INDs can translate into clinical trials and treatments for CTD and better treatments for GAMT. With the record-breaking 2021 Holiday Heroes campaign, ACD raised more than \$170,000 to kick off these and other research efforts.

To support its ambitious research vision, ACD works with research and science partners to fund research grants including the fellowship program, distribute biosamples, and collect survey and patient registry data, among other collaboration opportunities. The parent-led organization's research strategy is to fund, build, and share tools and resources that can accelerate the discovery of treatments for children living with creatine deficiencies. ACD's approach is collaborative and relies on open science to shorten the timelines required for development, with a focus on supporting the widest possible range of relevant research efforts in the scientific community in order to maximize the chances of getting treatments.

ACD is a proud member and grantee of the Rare As One network supported by the Chan Zuckerberg Initiative. More information about the ACD research initiative, including funding opportunities, is available at creatineinfo.org/research.

About ACD: The Association for Creatine Deficiencies was established in 2012 with the mission to eliminate the challenges of living with cerebral creatine deficiency syndrome. ACD is committed to providing patient, family, and public education to advocate for early intervention through

newborn screening, and to support and drive medical research for treatments and cures for CCDS. 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 screening and treatments needed to improve life quality and longevity for the CCDS patient. For more information regarding ACD, please visit creatineinfo.org.

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