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FOR IMMEDIATE RELEASE

ACD AWARDS OVER \$100,000 TO CEREBRAL CREATINE DEFICIENCY SYNDROMES RESEARCHERS

Carlsbad, CA (February 1, 2021)— Today, [the Association for Creatine Deficiencies](#) (ACD) announced the funding of a total of \$115,000 for researchers studying Cerebral Creatine Deficiency Syndromes (CCDS). The awards include three newly established ACD Fellowships totaling \$90,000 that will fund early career researchers, and a \$25,000 award for Dr. Nicola Longo.

Dr. Charles Kuntz (in the lab of Dr. Jonathan Schlebach), Dr. Peter Axerio-Cilies (in the lab of Dr. Sylvia Stockler), and Alex Lee (in the lab of Dr. Andreas Schulze) have been selected to each receive a \$30,000 ACD Fellowship Award for 2021-2022. The overarching goal of the ACD Fellowship program is to fund opportunities which have the potential for translational success. To this end, each of the awardees is focused on advancing therapeutic discoveries for creatine deficiency disorders.

Along with Dr. Jonathan Schlebach, Dr. Charles Kuntz will conduct virtual screening of different patient gene mutations. This work is expected to shed light on the functional impact of patient mutations and explores potential avenues for therapeutic rescue. In keeping with ACD's mission to advance translational therapies, Dr. Stockler and Dr. Axerio-Cilies will work on screening compound libraries on cells with various patient mutations.

ACD also welcomes Alex Lee, a graduate student in Dr. Andreas Schulze's lab, to the cerebral creatine deficiency disorders community. The ACD fellowship is expected to further Alex's work on understanding regulatory mechanisms for genes critical to the creatine synthesis and transport pathway.

Dr. Nicola Longo is awarded a \$25,000 grant that will go towards the discovery of an inhibitor of the synthesis of guanidinoacetate. The goal is to find a potential drug which, given with creatine supplements, can normalize the brain chemistry of GAMT patients, thus reducing the frequency of seizures and facilitating intellectual development. The current grant will be used for screening potential inhibitors in cells.

Dr. Laura Trutoiu, ACD's Director of Research, described the newly launched fellowship program as a critical step in broadening research avenues for creatine deficiencies. "Funding these stellar new and established researchers ensures that we have the brightest minds committed to finding treatments for creatine deficiencies," said Dr. Trutoiu. "We are enthusiastic to see their involvement in our community and their career accomplishments grow."

To support research, ACD works with research and science partners to fund research grants, distribute biosamples, and collect survey and patient registry data, among other collaboration opportunities. 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/.



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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 promote and fund 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 <http://www.creatineinfo.org>.