BUILDING STRENGTH
changing lives

CONTACT: Laura Trutoiu,
Director of Research
auract@creatineinfo.org

FOR IMMEDIATE RELEASE

Association for Creatine Deficiencies (ACD) Awards Dr. Gerald Lipshutz Ph.D., with their Gene Therapy Advancement Award (GTA)

Carlsbad, CA (October 2, 2020)—The Association for Creatine Deficiencies (ACD) is excited to award Dr. Gerald Lipshutz, Ph.D., with the second ACD Gene Therapy Advancement (GTA) award of $10,000.

The ACD was established in 2012, by parents with children diagnosed with a Cerebral Creatine Deficiency Syndrome (CCDS). Our mission is to drive and facilitate research for treatments and cures for creatine deficiencies as well as to provide patient, family, and public education, and to advocate for early intervention through newborn screening. To support research, the ACD can help with research grants, biosamples, survey data, and other collaboration opportunities. We are a proud member and grantee of the Rare as One network supported by the Chan Zuckerberg Initiative.

In the winter of 2019, the CCDS patient community raised an initial fund of $50,000 to be used towards gene therapy research efforts. With this support in mind, the ACD established the CCDS Gene Therapy Consortium in 2020 to foster concentrated efforts into gene therapy for creatine deficiencies. The mission of the consortium is to facilitate the timely sharing of information and development tools among labs that are pursuing gene therapies for creatine deficiencies. We believe that by building a collaborative environment and supporting shareable tools through grants we can shorten the timeline and effort required to find gene therapy solutions for creatine deficiencies. The consortium meets on a quarterly basis as a group to discuss the latest research and provide peer expertise.

“Bringing all researchers to the table to share their experience and insight is key to our mission of supporting multiple research avenues for all three CCDS. Dr Lipshutz’s pursuit of gene therapy for GAMT deficiency is much needed for our community, and we are happy to be able to support his work and look forward to seeing future results” said Laura Trutoiu, ACD Director of Research.

In October 2020, the ACD awarded the second GTA grant to Dr. Gerry Lipshutz for his ongoing efforts in gene therapy for Guanidinoacetate Methyltransferase (GAMT) deficiency disease. GAMT deficiency is one of the two common cerebral creatine deficiency syndromes arising due to autosomal recessive mutations in the GAMT gene. Symptom onset typically occurs before three years of age including features of autism and self-mutilation along with intellectual disability and seizures. The current standard for therapy is life-long and includes oral creatine to replenish cerebral creatine along with supplementation of ornithine and restriction of dietary arginine and protein.

Dr. Lipshutz and his lab have developed a viral vector based gene therapy for human GAMT that is currently in the early stages of testing in preclinical mouse models. Proceeds of the award will be utilized by Dr. Lipshutz to conduct measurements of guanidinoacetate (GAA) and creatine in bio samples collected from GAMT deficient mice that have received adeno associated viral vector (AAV:GAMT) based GAMT gene therapy. In addition to metabolomic measurements, animals will also be subjected to an array of behavioral tests that help determine the efficacy of GAMT gene therapy.

“We have been able to develop a gene therapy approach for GAMT deficiency and thus far it has shown biochemical efficacy in a

mouse model of the disorder. Having these additional funds will help us in performing behavioral studies with the mice to see if we can both identify a behavioral abnormality and also determine if it is resolved with this gene therapy approach. Interacting with the family representatives from the Association for Creatine Deficiencies has been a wonderful experience and working together I hope that in the not too distant future we can bring forth a new and efficacious therapy to patients with GAMT deficiency." - Dr. Lipshutz

ABOUT DR. GERALD LIPSHUTZ

Dr. Gerald Lipshutz is a Professor in Residence at UCLA, having been on faculty for 16 years. He has primary appointments in Surgery and in Molecular and Medical Pharmacology and is also a member of the Intellectual and Developmental Disabilities Research Center at UCLA. As a physician-scientist he runs an NIH-funded laboratory in gene and cell therapy in addition to his clinical effort. His main research interests have been in developing gene and cell therapies for rare metabolic disorders of the liver, primarily urea cycle disorders. Specifically his effort has focused on arginase deficiency and carbamoyl phosphate synthetase 1 deficiency. While both disorders lead to intellectual disabilities, the mechanism of neurologic injury is quite different. It is the finding of some similar biochemical abnormalities in arginase deficiency (specifically elevated guanidinoacetate) that led Gerry to begin to investigate gene therapy approaches for GAMT deficiency and the creatine family of disorders more broadly.

ABOUT THE ASSOCIATION FOR CREATINE DEFICIENCIES

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 screening and treatments needed to improve life quality and longevity for the CCDS patient. To learn more about creatine deficiencies please see our educational video or visit http://www.creatineinfo.org.