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Association for Creatine Deficiencies Selects Dr. Olivier Braissant for Gene Therapy Advancement Award

Carlsbad, CA (July 15, 2021)—The Association for Creatine Deficiencies (ACD) has awarded Olivier Braissant, Ph.D., the first 2021 quarterly Gene Therapy Advancement Award (GTA) for $10,000.

After parents and families raised $50,000 in 2019 to support gene therapy research, ACD started 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, development of tools among labs that are pursuing gene therapies, and fostering a collaborative environment to find gene therapy solutions for creatine deficiencies. The consortium meets on a quarterly basis as a group to discuss their research projects and provide peer expertise.

In May 2021, ACD awarded the GTA grant to Braissant to enable testing of viral vector mediated gene therapy in a novel rat model of Creatine Transporter Deficiency (CTD). With this award, Braissant's lab will be able to test a viral vector to treat CTD in mutant rats, measure correction of creatine levels in the brain and determine if behavioral issues can also be also improved by this method of gene therapy. The study also aims to provide insights to help with choosing the best applicable protocol to be used in humans. For instance, Braissant will look at identifying the best delivery route, best dose of AAV vectors, and best time of delivery.

Braissant is an associate professor in the Service of Clinical Chemistry at the Department of Laboratory Medicine and Pathology from Lausanne University Hospital (CHUV). A specialist in the development and metabolism of the nervous system, Braissant focuses his research on inborn errors of metabolism affecting brain development. He is recognized as an expert on two groups of rare genetic diseases affecting the brain: creatine deficiency syndromes and urea cycle diseases.

"Dr. Braissant's lab has made significant contributions to the study of creatine deficiency disorders. We are excited to fund this new effort in his lab and hope to witness the impact of gene therapy in resolving CTD. These types of studies undoubtedly lay the foundation for future success in patients," said ACD Director of Research, Laura Trutoiu.

The Gene Therapy Advancement Award is part of an overall research strategy into which ACD is making significant investments. In March, ACD also announced the funding of $115,000 for four researchers who are studying Cerebral Creatine Deficiency Syndromes (CCDS). Three of these research studies are focused on CTD with two focused on drug repurposing, and the fourth is studying potential GAMT

treatments. ACD has also privately funded an additional CCDS research project; more details will be available in the coming year.

Additionally, ACD has arranged for biobanking at the Coriell Institute to make working with patient samples easy for researchers worldwide by collecting and storing cells from patients with CCDS. There are now 36 patient samples in the Coriell catalog available for researchers to access, and this year there has been an increase in the number of researchers requesting patient samples. Such tools enable researchers to understand the unique nature of patient mutations and their impact on function, laying the foundation for future personalized therapies. For more information on how to submit patient samples to Coriell, please visit: creatineinfo.org/coriell.

ACD was established in 2012, by parents with children diagnosed with a CCDS. The organization's 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, ACD can help with research grants, biosamples, survey data, and other collaboration opportunities. ACD is a proud member and grantee of the Rare As One network supported by the Chan Zuckerberg Initiative.

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 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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