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FOR IMMEDIATE RELEASE

Association for Creatine Deficiencies Receives $450,000 CZI Rare As One Grant

Carlsbad, CA (Feb. 3, 2020)—The Association for Creatine Deficiencies (ACD) today announced that the organization has been selected to join the Chan Zuckerberg Initiative (CZI) Rare As One Network and awarded a $450,000 grant.

ACD is one of 30 organizations selected to be a part of the Rare As One Network. CZI is providing these organizations with funding, training, community mentorship, and capacity-building resources. The grant will serve to build capacity for the organization; build upon a research network for Cerebral Creatine Deficiency Syndromes (CCDS) by working with the patient community and researchers and clinicians; and hosting an international scientific meeting to convene the research network.

“No one is more committed to finding cures for rare diseases than the patients and families of those affected by these disorders,” said Priscilla Chan, Co-Founder & Co-CEO of CZI. “We are proud to support patient-led organizations as they pursue diagnoses, information, and treatment options in partnership with researchers and clinicians.”

This opportunity is part of the Rare As One Project’s goal to lift up the work that patient communities are doing to accelerate research and drive progress against rare diseases, which affect approximately 400 million people worldwide. The program will provide funding, tools, and support and training to selected organizations.

“As parents, we can’t afford to be bystanders in the research process waiting on developments from the medical field,” said ACD Director of Research Laura Trutoiu. “Our organization is committed to being an active player in research, and being part of the Rare As One Network is a once-in-a-lifetime opportunity for us to speed up the development and implementation of truly collaborative research efforts.”

The ACD’s end goal is to identify effective treatments for all three CCDS, ultimately ensuring that any children with a creatine deficiency are diagnosed at birth and receive prompt treatment. Guanidinoacetate methyltransferase deficiency (GAMT) and L-Arginine:glycine amidinotransferase (AGAT) deficiencies are treatable, but most newborn screening programs still do not screen for them at birth. Creatine Transporter Deficiency (CTD) is currently untreatable.

“Getting clinical trials underway as soon as possible is crucial for our children to have hope for a better future,” added Trutoiu. “A critical step for research growth for the ACD is to centralize patient data, and the Rare As One grant will help make that happen.”

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The collaborative research network the ACD is growing is supported by a Scientific and Medical Advisory Board consisting of 11 doctors from around the world who contribute to the organization's mission with their commitment to the community and vast knowledge of molecular and biochemical genetics research translation to clinical application. The ACD is particularly grateful to have the guidance of the Scientific and Medical Advisory Board President Dr. Nicola Longo as lead researcher in the Rare As One Network project, along with Dr. Saadet Andrews as lead clinician. Dr. Longo and Dr. Andrews will work together with Trutoiu and the ACD to guide the growth of the organization's collaborative research network competencies.

The ACD is a parent-led advocacy organization that provides patient and family support and public education to advocate for the earliest possible diagnoses of CCDS. The organization is committed to fostering a symbiotic collaboration where researchers and parents work together. With three rare diseases under one umbrella, rather than splitting resources up to focus on each disease separately as commonly happens in the rare disease community, ACD is leveraging the power of a stronger, united community to drive research.

"CCDS are rare and we are a small, growing patient advocacy organization. Until now, we have not had the resources to work on longer term strategic plans," said ACD President Heidi Wallis. "With the support of the Rare As One Network, we will be able to tackle a long list of strategic goals, getting us closer to our vision of the best possible treatments and outcomes for all individuals born with a Cerebral Creatine Deficiency Syndrome."

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 http://www.creatineinfo.org.

About the Chan Zuckerberg Initiative: Founded by Dr. Priscilla Chan and Mark Zuckerberg in 2015, the Chan Zuckerberg Initiative (CZI) is a new kind of philanthropy that's leveraging technology to help solve some of the world's toughest challenges — from eradicating disease, to improving education, to reforming the criminal justice system. Across three core Initiative focus areas of Science, Education, and Justice & Opportunity, CZI is pairing engineering with grant-making, impact investing, and policy and advocacy work to help build an inclusive, just, and healthy future for everyone. For more information, please visit www.chanzuckerberg.com.

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