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**PCORI Funding Awarded to Association for Creatine Deficiencies
Project to Empower CCDS Parents in Research Engagement**

Carlsbad, CA (Nov. 9, 2021)—The [Patient-Centered Outcomes Research Institute](#) (PCORI) selected the [Association for Creatine Deficiencies](#) (ACD) for a \$238,000 Eugene Washington PCORI Engagement Award for a project titled “Parents Advancing REsearch NeTworkS” (PARENTS). The two-year project will launch on Dec. 1 and will build a network enabling parents/caregivers and researchers to work in partnership on developing a core outcome set for Cerebral Creatine Deficiency Syndromes (CCDS), aligned with what really matters to CCDS families.

With the PARENTS project, ACD aims to empower parents/caregivers as experts leading research, providing ongoing support and training, to build patient-centered outcomes research (PCOR) and prepare them for future research opportunities that will give them the ability to make better-informed health decisions and improve outcomes and quality of life for patients with CCDS. Because CCDS are rare diseases, there are challenges for doctors to gain insight from more than one patient, and for families to come together to share knowledge, and with the PARENTS project, ACD intends to bring solutions to these areas.

The goals of the PARENTS project are to:

- Engage, train, and empower CCDS parent/caregivers on the entire research process, with a special focus on patient-meaningful outcomes (PMOs), a core outcome set and comparative clinical effectiveness research, that in the future will give patients and those who care for them the ability to make better-informed health decisions;
- Build a network where parents/caregivers and researchers work in partnership on developing a core outcome set for CCDS, aligned with what really matters to CCDS families;
- Build community and a collaboration platform between parents and clinician-researchers;
- Collaboratively develop a core outcome set for CCDS, aligned with what really matters to CCDS families;
- Ensure ongoing support to develop a sustainable PCOR agenda, with parents/caregivers as research partners leading future research opportunities based on what is most important for them;
- Build a set of research priorities for the CCDS community and a model replicable for other patient advocacy organizations, especially in the ultra-rare disease community.

“Because CCDS are ultra-rare diseases, as caregivers we are keenly aware of the necessity of engaging in research and that we cannot simply wait for research to move; we need to move research,” said ACD Director of Research and PARENTS project lead, Laura Trutoiu. “Research means hope for our children



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and loved ones. Parents and caregivers are experts in rare diseases and with the PCORI award we have the opportunity to drive and participate as partners in research for our loved ones.”

The PARENTS project asserts that by creating a network between parents/caregivers and researchers that will empower families, provide resources, build capacity and develop a patient-centered research agenda, the CCDS community will collaboratively move CCDS research forward. The result will be to build a strong partnership and sustain the PCOR CCDS agenda, identifying research projects that can improve outcomes that are most relevant to the CCDS families. Defining these outcomes will enable the success of any clinical trials as well as open up the potential for precision medicine (n=1 trials) to help the CCDS community.

With ACD taking the lead on this project overall, project leads are: Dr. Sylvia Gerda Stockler-Ipsiroglu, Investigator, BC Children's Hospital Program Director in Biochemical Diseases at BC Children's Hospital as scientific co-lead; Dr. Beth Potter, Associate Professor at University of Ottawa and holder of the University Research Chair in Health Services for Children with Rare Diseases; and Dr. Audrey Thurm, Director of Neurodevelopmental and Behavioral Phenotyping Service in the Office of the Clinical Director at the National Institute of Mental Health. The project team supporting the project leads consists of: Patient Engagement Advisor and PCORI Ambassador Maureen Smith, Delia Apatéan, Research Coordinator at BC Children's Hospital, and ACD's Community Engagement team members.

"I am honored and grateful to participate in this most innovative project," said Dr. Stockler. "Core outcomes are needed as they are meant to be used as outcomes in every single clinical trial that ever will be done around the globe for specific conditions such as creatine deficiencies. If everybody uses the same set of outcomes, if they all assess the same thing—this will help compare if the treatment actually works or not. Additionally, when developing core outcomes sets, it is so important to understand what outcome matters the most to families, in response to potential treatments, with the ultimate goal of improved quality of life and wellbeing."

CCDS are inborn errors of metabolism, which interrupt the formation or transportation of creatine. Creatine helps supply energy to all cells in the body. Symptoms may include intellectual delays, gross motor delays, expressive speech and language delay, autistic-like behavior, hyperactivity, seizures, projectile vomiting in infancy, and failure to thrive. ACD is dedicated to the three CCDS: Creatine Transporter Deficiency (CTD), Guanidinoacetate Methyltransferase (GAMT) Deficiency, and L-Arginine: Glycine Amidinotransferase (AGAT) Deficiency.

The Patient-Centered Outcomes Research Institute (PCORI) is an independent, nonprofit organization authorized by Congress in 2010 to fund research that will provide patients, their caregivers, and clinicians with the evidence-based information needed to make better-informed healthcare decisions. Eugene Washington PCORI Engagement Awards support projects that encourage active, meaningful involvement of patients, caregivers, clinicians, and other healthcare stakeholders as integral members of the patient-centered outcomes research/comparative clinical effectiveness research enterprise. [Learn more.](#)

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



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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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