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Association for Creatine Deficiencies Expands Patient Insights Network to Enable Data Sharing with ClinVar

Carlsbad, CA (July 27, 2018)- The Association for Creatine Deficiencies (ACD) is working with Geisinger, a National Institutes of Health (NIH) Clinical Genome Resource (ClinGen) grantee to share genetic and health information from patients affected by creatine deficiencies. This information will be collected through the ACD Patient Insights Network (PIN), a patient registry platform. Genetic variant information will be collected through the registry and shared with the ClinVar database.

Geisinger, as a ClinGen grantee, plans to help rare disease patient advocacy groups collect and share genetic test results and significance to advance genomics. Through their existing registry hosted by Invitae, ACD will participate in the Patient Data Sharing Program to have certified genetic counselors review and share their participants' de-identified genetic and health information. Registry participants are able to direct whether or not they would like their de-identified data shared. Whitnie Strauss, President for ACD commented, "It's our mission to drive research for CCDS (Cerebral Creatine Deficiency Syndromes). We believe gathering and sharing genetic information is vital to gaining a deeper understanding of GAMT, AGAT, and CTD."

Juliann Savatt, MS, LGC, genetic counselor at Geisinger said, "The ACD is the first patient advocacy organization to move forward with data sharing and we are excited for this joint venture. Data sharing can help support ClinGen's efforts to better understand the relationship between genetics and health. It can help clarify uncertain genetic testing results and provide more information about a condition to inform treatment and management."

The ACD PIN is designed to better understand the medical history and disease journey of creatine deficiency syndromes by collecting information directly from patients and caregivers from around the globe. The PIN puts patients in the driver's seat to control their information, while enabling patients to learn from others to manage their disease. By forming a network of engaged patients, the ACD and disease advocates may accurately speak on behalf of the disease community. Interested patients diagnosed with a creatine deficiency syndrome are invited to join the PIN at https://creatineinfo.org/patient-registry/.

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About ClinGen & ClinVar
ClinGen is an NIH-funded effort dedicated to identifying clinically relevant genes and variants for use in precision medicine and research. ClinVar is a freely accessible, public archive of reports of the relationships among human variations and phenotypes hosted by the National Center for Biotechnology Information (NCBI) and funded by the Intramural National Institutes of Health (NIH). ClinGen investigators work closely with NCBI regarding the development and functionality of ClinVar and to support data deposition from many sources including genetic testing laboratories, researchers, and patients.

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. CCDS are inborn errors of metabolism, which interrupt the formation or transportation of creatine and 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 the care needed to improve life quality and longevity for CCDS patients. For more information regarding ACD, please visit https://creatineinfo.org/patient-registry/.