



Fall 2022 Update

When we launched our CreatineInfo Registry in March 2021, researchers and clinicians told us that 200 participants in a patient registry would be the ideal benchmark to drive patient-focused research; this would be the “magic number” to make meaningful interpretations from the data and build support from outside partners. Therefore, we set ourselves a goal of enrolling 200 CCDS patients. We are happy to report that we are 80% of the way towards reaching that goal - we currently have 160 participants enrolled in our registry! Participating in the patient registry and natural history study is an easy way that you can help drive CCDS research. If you have not yet enrolled or would like to confirm that you are up-to-date on all of your surveys, please visit <https://creatineinfo.iamrare.org>. Help us reach our goal of 200 participants by enrolling today!

In addition to growing our enrollment, we have been hard at work developing new surveys to launch in the registry. In particular, we will release a cardiovascular health survey and a seizure survey in the coming months. We have partnered with Dr. Mark Levin (cardio) and Dr. Saadet Andrews (seizure), in addition to our Family Advisory Board (FAB) to ensure these surveys are scientifically-driven while maintaining the caregiver perspective. Stay tuned for their launch!

Volunteers Needed for our Family Advisory Board (FAB)! As our patient registry grows, we want to ensure that everyone’s voice and perspective is heard. Therefore, we are expanding our FAB! In particular, we are looking for more global representation (outside of the U.S.) and more GAMT & AGAT representation. As a FAB member, your primary role is to provide feedback for the registry and for the surveys prior to their launch. The commitment level is relatively light; we hold weekend meetings every 2-3 months. If you are interested, please email Emily at registry@creatineinfo for more information.