



CreatineInfo Registry Tips Sheet

creatineinfo.iamrare.org

Here are some tips that can help you prepare for the registry. We will update this page regularly. Once you join the registry, you will have the option to receive email reminders and tips by email or your preferred contact method.

If you have any questions or need assistance, please contact registry@creatineinfo.org and the Registry Coordinator will assist you.

Update: March 2021

Surveys available now

Initial Surveys

Initial Surveys collect the baseline information of the participants.

Here you will be asked about:

- 1) **Caregiver Contact Information**
- 2) **Participant Profile** (Socio-demographics)
- 3) **Diagnosis** (Prior diagnosis, initial symptoms, clinical tests, other diagnoses)

Please note that more in-depth surveys related to specific symptoms and interventions will follow.

In the *Diagnosis survey*, you will be asked to upload your genetic testing report. Genetic information is very important for this study and to help research. If you need help getting your genetic testing report, the ACD can assist you (e.g., connect you with a genetic counselor).

You will also see an additional survey, the **ClinGen Patient Data Sharing** Informed Consent, where you will have the opportunity to opt in to participate in the ClinGen Patient Data Sharing Program, to share the Study Participant's pseudonymized (no personal information will be shared) genetic and health data to improve patient care and genetic testing.

To learn more about ClinGen and where data will be shared, please visit

https://bit.ly/GC_Databases

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Upcoming Surveys

The upcoming surveys will include questions about **symptoms and interventions**, including behavior, seizures, cardiac and gastrointestinal conditions, as well as medical test reports and treatments.

Here are some tips that can help you gather all the information needed for the next batch of surveys:

- Collect all diagnosis documents and test results (as an example, if a biochemical test was done, you will be asked about the creatine levels at the time, or if an EEG was done, you will be asked if the result was normal or abnormal.)
- Make a list of treatments, medications, and diet, current and past.
- If you are unsure of tests or symptom treatments and have an upcoming appointment with a physician, please request all available results.