

ClinGen Data Sharing Informed Consent

Thank you for your interest in the ClinGen Data Sharing Program. Data sharing helps increase understanding of a condition's features, which may help identify possible interventions and treatments. By sharing your information, you are driving research forward.

What is the ClinGen Data Sharing Program?

The ACD is working with the Clinical Genome Resource (ClinGen) Data Sharing Program to give you the choice to share the Study Participant's pseudonymized genetic and health data with others who will use it to improve patient care and genetic testing.

"Study Participant" refers to the person diagnosed with Cerebral Creatine Deficiency Syndromes (CCDS).

ClinGen is a National Institutes of Health (NIH)-funded project aiming to build a resource that defines the impact of genes and genetic changes on health. This effort relies on data sharing.

How will my information be shared?

If you choose to give the ClinGen Patient Data Sharing Program access to Study Participant's individual genetic and health information that you share with ACD, ClinGen will have access to the Study Participant's genetic testing report that you uploaded and the health history you share via surveys.

- If you did not upload the Study Participant's report, you will have an opportunity to upload it in the future.
- If you no longer have a copy of their testing, the ClinGen team can help you request one.

Once the Study Participant's report is shared, a ClinGen Patient Data Sharing Program team member will collect information about the genetic change(s) that were found from the report and will pseudonymize and share the Study Participant's genetic and health data with approved users and open and controlled-access databases including:

- ClinVar, a National Center for Biotechnology Information database of genetic changes and their relationship to human health.
- Pseudonymized information from ACD will also be available to inform other ClinGen activities to understand the relationship between genes and health.
- To learn more about where data would be shared go to https://bit.ly/GC_Databases (in a separate tab or window)

No personal identifying information will be shared. The ClinGen Patient Data Sharing Program also will have access to your email address from the Caregiver contact information survey in order to contact you with questions about your genetic test results and health surveys.

How can I choose to receive updates from ClinGen?

If you choose to give the ClinGen Patient Data Sharing Program access to the Participant's individual genetic and health information, you also have the option to receive updates about the Study Participant's genetic test results from the ClinGen team. It is important to remember:

- As we learn more about genetic changes, their relationship to health might change.
- Some updates might impact the Study Participant's medical care while others will not.
- Such updates are rare, so most participants will not receive any. It is not possible for the ClinGen Patient Data Sharing Program to identify all updates related to Study Participants' genetic test results.

If you choose to receive updates, you might be contacted by the ClinGen Patient Data Sharing Program team if we learn about potential updates to the Study Participant's genetic test results. ClinGen Patient Data Sharing Program would contact you and direct you back to the Study Participant's doctor or a healthcare provider in your area to discuss this more. Please remember to provide your email in the Caregiver contact information survey so that your contact information is on file.

CHECK all that apply (Yes or No)

I agree to give ClinGen access to the Study Participant's individual genetic and health information. ClinGen will then share the Study Participant's pseudonymized data with other approved users and open and controlled-access databases. **NO personal identifying information will be shared.**

Yes ___ No ___ *

(If yes)

Would you like ClinGen to contact you via email about potential updates to the Study Participant's genetic testing results? Please remember to complete the Caregiver contact information survey so that your email contact information is on file. Yes___ No_____*