

## What is a patient registry?

A patient registry is a centralized, secure place to store medical information, test results, tissue samples, and blood samples so that ADCY5 families and researchers can easily manage this information.

## Why is a patient registry important?

Having this information organized in one place makes it much easier for doctors and researchers to study the ADCY5 gene mutation. The more information that is collected on the ADCY5 gene mutation, the more information researchers have to study. This will hopefully lead to new discoveries and treatments that can help the ADCY5 gene mutation families.

Since diagnosis of ADCY5 mutations are rare, few patients have been identified. As a result, it is critical that every patient is registered to gather the maximum amount of information. As research progresses and clinical trials become possible, the information needed by researchers will already have been gathered.



**ADCY5.ORG** supports and provides resources to the ADCY5 community. The organization's goal is to support affected families, support researchers and clinicians to find treatments and someday a cure for ADCY5-related dyskinesia.

### Contact Us

Email: [info@adc5.org](mailto:info@adc5.org)

Website: <http://www.adcy5.org/>

Facebook: Search for "ADCY5"  
+1.858.882.7420

## Our Partner



RARE Science is a non-profit research organization whose mission is to accelerate finding therapeutics for children with rare disease through a common computational platform that enables data sharing across all stakeholders.

E-mail: [info@rarscience.org](mailto:info@rarscience.org)

Web: [www.rarscience.org](http://www.rarscience.org)



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## THE ADCY5 INTERNATIONAL PATIENT REGISTRY

*The First Step in Finding A Cure*

Connecting Stakeholders  
Across the Globe

## How do I join the ADCY5 Patient Registry?

The first step is to contact us so that a medical records expert can help you collect your medical information from doctors, hospitals, and other healthcare providers. You will need to sign a release so that the clinical research organization can retrieve your records for you and load them into the registry. Please email us [info@adc5.org](mailto:info@adc5.org).

## What will we do with the data?

The data and samples are stored and managed in accordance with international standards and applicable privacy laws. The registry will be set up so that the patient or the patient's family has control of who can access the medical information. Data from medical records will then be standardized into a common format and stored anonymously in a database. The database will ensure privacy, but will enable clinicians and researchers to analyze individual patient data and data from the whole ADCY5 group. The more patients analyzed, the better the disease will be understood.



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*Moving towards a cure one child at a time.*

## What is ADCY5-related Dyskinesia?

A mutation in the ADCY5 gene, results in ADCY5-related dyskinesia, a motion disorder that causes involuntary muscle movement such as tics and spasms. Not every patient has the same level of physical symptoms. It initially affects infants to teenagers. Infants with severe cases may experience a delay in motor milestones.

ADCY5-related dyskinesia is often misdiagnosed as another motion disorder such as cerebral palsy, epilepsy or mitochondrial disease. Because there are not many known cases, researchers and clinicians have hard time identifying the disease. It is currently diagnosed through genetic testing and it is difficult to find medicines to treat or cure patients. As more individuals are gathered through a registry, diagnosis and treatments can be found sooner.

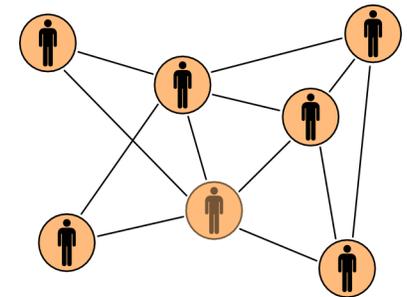
*Coming together is the beginning.  
Keeping together is progress.  
Working together is success.*

*-- Henry Ford*

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## Seven Benefits of having a Patient Registry

1. **Brings together the ADCY5 community for support and resources.**
2. **Provides a secure central location to organize and compare medical records.**
3. **Brings awareness to the disorder to drive faster diagnosis.**
4. **The medical information helps clinical researchers understand the disease and identify potential treatments.**
5. **Unites patients and families around the world.**
6. **Simplifies the set up of ADCY5 clinical trials.**
7. **Leads to collaboration and improved care, even in the absence of an immediate cure.**



**Every family is an important piece of the puzzle.**