

Envision the Cure - Embrace the Challenge

Rare Disease and the Coronavirus Pandemic



**how does covid-19
impact people with
CADASIL?**

HELP DOCTORS FIND OUT BY
COMPLETING AN ONLINE SURVEY



The NIH is interested in how COVID-19 is affecting people with CADASIL. The survey is 30 minutes long and is for individuals with CADASIL who either have or have not had COVID-19. To participate **before the study ends on Oct 31**, please visit:

https://redcap.link/covid_in_communities

The survey is in both **English** and **Spanish**. If other languages are needed, please email COVIDinCommunities@nhlbi.nih.gov. Additional information about this rare disease study can be found on the [NIH website](#). Dr. Kozel and her team hope to have 500 CADASIL participants!



Million Dollar Bike Ride 2020 results in \$82,000 CADASIL research grant! CADASIL Community is amazing!

The Penn Medicine Orphan Disease Center 2020 Million Dollar Bike Ride was a virtual event this year due to COVID-19. That did not stop **Team CADASIL** participants and supporters from raising over \$50,000 to support CADASIL research! In addition, UPenn matched \$30,000 dollar-for-dollar and they are in the process of reviewing CADASIL grant applications for the 2021 \$82,000 CADASIL research grant! Thank you!


Meet [Dr. Beth Kozel, MD, PhD](#), a [clinical geneticist and principal investigator of the COVID-19 Rare Disease study](#) currently looking for CADASIL patients to participate in their research study. Watch this **CADASIL care** [webcast](#) by Dr. Kozel on genetic testing for CADASIL on our [website](#).

CADASIL care

**Genetic Testing for CADASIL:
When, why and how to test**

Beth Kozel, MD, PhD
Clinical Geneticist, Lasker Clinical Research Scholar
National Heart, Lung, and Blood Institute (NHLBI)

Do you have loved ones who have not joined the **CADASIL Family Registry** and




GenomeConnect
The ClinGen Patient Partner

**ClinGen's Patient Data Sharing Program:
Leveraging Data Sharing Experience from GenomeConnect
to Broaden Patient Data Sharing Efforts**

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ClinGen
Clinical Genome Resource

Background


The Clinical Genome Resource (ClinGen)

- Broad sharing of genomic and phenotypic data informs variant classification and gene-disease validity
- ClinGen has facilitated patient data sharing through its patient registry, GenomeConnect, since 2014.

GenomeConnect

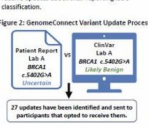
- Open to anyone who had genetic testing regardless of diagnosis or test results.
- Patients have been a source of novel genomic data and additional case level information through GenomeConnect.

Figure 1: GenomeConnect ClinVar Submissions



- Data sharing can provide participants a means to receive updates about their reporting lab's classification.

Figure 2: GenomeConnect Variant Update Process



- Given GenomeConnect's success in engaging patients data sharing, ClinGen aimed to expand data sharing to registries managed by parent advocacy organizations.

ClinGen is funded through the following grants: 1U49HG008214, 1U49HG008448, 1U49HG008450. GenomeConnect is built on the ReportConnect® which covered thousands of tested patients.

Patient Data Sharing Program

Figure 3: Patient Data Sharing Program Process

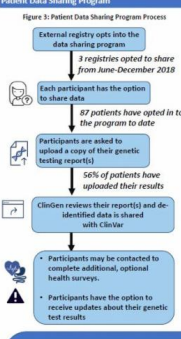
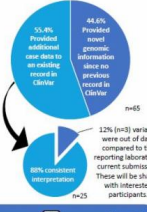



Figure 4: Variants Shared by Patient Data Sharing Participants



Patient data sharing contributes valuable information to the public knowledge base that may not be otherwise available, benefiting both patients and the genetics community.

Take a picture with your phone to learn more.



data sharing@clin-gen.org
@GenomeConnect

[cureCADASIL](http://cureCADASIL.com) | info@cureCADASIL.org | Website

STAY CONNECTED

