



## Envision the Cure - Embrace the Challenge

### Rare Disease Resources for Coronavirus Pandemic

While everyone is affected by the coronavirus pandemic, we understand those with CADASIL and other underlying diseases may have extra concern. Although there is not specific research or guidance for CADASIL, we hope these [rare disease resources](#) from our partner Global Genes are helpful. Please stay healthy.

### Million Dollar Bike Ride 2020 goes virtual!

After much deliberation, the Penn Medicine Orphan Disease Center has decided to host a virtual Million Dollar Bike Ride, instead of gathering face-to-face on June 13th. Their efforts to support rare disease is ongoing and they remain committed to the 2020 MDBR Research Grant Program. Team CADASIL will participate in the virtual event and continue to raise up to \$30,000 to be matched dollar for dollar by UPenn to support a CADASIL research grant.

Ride, run, or walk the week of June 8th and post your picture on social media with the hash tags #cureCADASIL #PennMedMDBR2020 [Your support is appreciated!](#)

Do you have loved ones who have not joined the CADASIL Family Registry and the ClinGen program? Encourage them to join today! [www.cureCADASILFamilyRegistry.com](http://www.cureCADASILFamilyRegistry.com)

### CADASIL Poets and Authors

Families affected by CADASIL have amazing ways of expressing themselves and making a difference for this rare disease. A New Jersey couple has been writing and performing poetry for many years. Please visit our website to learn more about [David and Eloise's use of poetry](#) during their journey with CADASIL.

CADASIL champion [Michael Kennedy](#) has authored two books whose proceeds support CADASIL awareness, education and research. Michael has hosted numerous fundraisers, co-chairs Team CADASIL in the Million Dollar Bike Ride and amazingly finds time to

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

Juliana M. Savetti, Danielle B. Azzariti, W. Andrew Faucett, David H. Ledbetter, Vanessa Rieger Miller, Emily Paton, Heidi L. Rehm, et al.

**Background**

The Clinical Genome Resource (ClinGen) is a global consortium of researchers and clinicians working to understand the genetic basis of rare diseases. ClinGen has facilitated patient data sharing through its patient registry, GenomeConnect, since 2014.

**Figure 1: GenomeConnect ClinVar Submissions**

46% of ClinVar submissions are from ClinGen participants. 51% of ClinGen participants have submitted at least one variant to ClinVar. n=959

**Figure 2: GenomeConnect Variant Update Process**

27 updates have been identified and sent to participants that opted to receive them.

**Figure 3: Patient Data Sharing Program Process**

External registry opts into the data sharing program. 3 registries opted to share from June-December 2019. Each participant has the option to share data. 87 patients have opted in to the program to date. Participants are asked to upload a copy of their genetic testing report(s). 56% of patients have uploaded their results. ClinGen reviews their report(s) and de-identified data is shared with ClinVar. Participants may be contacted to complete additional, optional health surveys. Participants have the option to receive updates about their genetic test results.

**Figure 4: Variants Shared by Patient Data Sharing Participants**

55.9% of variants are from ClinGen participants. 44.6% of variants are from ClinGen participants. 12% (n=3) variants were out of date compared to the reporting laboratory's current submission. These will be shared with interested participants. 88% of variants are from ClinGen participants. n=23

**Take a picture with your phone to join more.**

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

GenomeConnect is funded through the following grants: 1U54HG006514, 1U54HG006515, 1U54HG006516. GenomeConnect is built on the PatientOverseas (a wholly owned subsidiary of Invitae) platform.

writing children's books with a purpose.

**Thank you Eloise, David, and Michael!**

## cureCADASIL Leadership Expansion, with a Focus in Drug Development

It is with great pleasure that the Board of Trustees announces the appointment of 2 new Trustees. Joining the [Board of Trustees](#) are Lisa Pugliese (Board Member, expertise in clinical trial development and management) and Debra Robinson (Board Member, expertise in global drug program development).

In addition to the transition of Jane Gunther from Trustee to President, this expansion represents a shift for cureCADASIL, bringing drug development talent, expertise, and energy to the table that will support and focus our research and clinical initiatives. Thank you for your support as our organization continues to evolve to envision a cure for CADASIL and take the necessary steps to make a cure a reality!



### 2019 MDBR Research Grant: Pericyte Contractility in CADASIL



Read details provided by Dr. Fabrice Dabertrand ([University of Colorado at Denver](#)) on his research grant titled [Pericyte Contractility in CADASIL](#).

Let's do it again in 2020!! Donate to Team CADASIL and help us reach \$30,000 to be matched dollar for dollar by UPenn!

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