



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.

Saturday June 13th 2020
IS NOW VIRTUAL
We may not be able to ride together on June 13th, but we will still hit the road for research!

So the Ride is Virtual, Now What?
Between now and June 13th, show your support for the MDBR by riding (safely and socially distanced) or help us by fundraising (like to help those in need). We will still post – and in solidarity – to other rare diseases and money for rare disease research. This means we want to see your training progress, milestones and achievements over the next several weeks! Use #PennMedMDBR2020 to post your photos and other updates on social media and tag the MDBR!

What Happens on June 13th?
On June 13th, have fun social media to stay updated on cyclist progress, fundraising status, and encouragement from the CDC. Be sure to share your progress on social media by using #PennMedMDBR2020, and tag the MDBR (handles below).

Are We Still Fundraising?
Fundraising continues! The CDC remains committed to offering matching funds for teams who raise \$20,000. Let's help them reach their goal so we can fund rare disease research across the globe!

Whether you're riding 13, 34 or 72 miles in your neighborhood, living room, or in spirit, we hope you will join us for the 7th Annual Million Dollar Bike Ride!
Circumstances may keep us from riding together in Philadelphia, but our commitment to the research must go on!

The Million Dollar Bike Ride for Cystic Disease Research
@cureCADASIL @cureCADASIL @cureCADASIL Penn Medicine

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

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

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

Julian M. Savett, Danielle R. Azzafro, W. Andrew Paucetti, David H. Ledbetter, Vanessa Rangan Mitter, Emily Falero, Heidi L. Rabinov, Joel Shouler, Leo Rogers, Sandra Pineda, Laura Tricoli, Jo Anne Vidali, Erin Rooney Riggs, Christa Lise Martin

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

40% Pathogenic variant data in review	51% Benign variant submissions, most are previous record in ClinVar
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27 updates have been identified and sent to participants that opted to receive them.

Figure 2: GenomeConnect Variant Update Process

Participating Lab A (BRCA1, BRCA2) vs. ClinGen (BRCA1, BRCA2) Library Design

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Figure 3: Patient Data Sharing Program Process

- External registry opts into the data sharing program
- 3 registries opted to share from June-December 2018
- 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

55-6% Provided additional case data to an existing record in ClinVar	54.0% Provided information about new problems record in ClinVar
88% consistent with previous	12% (n=3) variants were out of date compared to the reporting laboratory's current submission. These will be shared with interested participants.

Patent 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 join the program.

debarand@clin-gen.org @GenomeConnect

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 write wonderful 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!

Donate to Team CADASIL

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STAY CONNECTED

