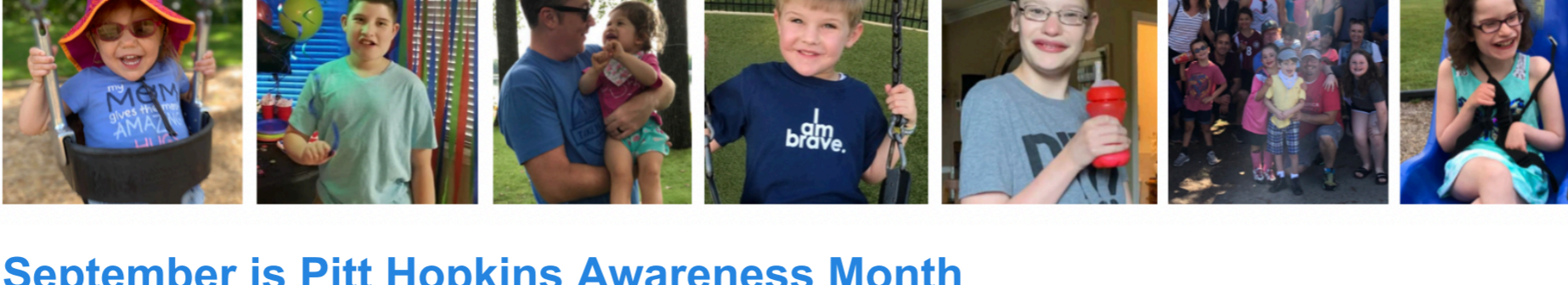


PITT HOPKINS NEWSLETTER - SEPT 2023



September is Pitt Hopkins Awareness Month

Get Involved today!

Guide to Awareness Day: 5 Ways to Get Involved!

- 1. Do a Fundraiser**
 - [Facebook Campaign](#) - Facebook is such a great tool for non-profits, with ZERO fees taken out of your donation and is an easy way to reach all your family and friends!
 - Get your school or work place involved - do a hat day/casual day
 - [Fundraiser Campaign](#)
 - Online Raffle
 - T-shirt fundraiser
 - Planning a fundraiser? Check out our new editable fundraising sponsorship [template](#).
- Check out <https://pithopkins.org/fundraise/> for more ideas on how you can do a fundraiser to support Pitt Hopkins research. Be sure to let us know what you have planned, we love to share ideas with our community!
- Join us Sunday, September 10th - Fundraising Zoom - Tips and Tricks. All things Fundraising! A zoom link will be available on our social media pages.
- 2. Get involved in Research**
 - [Fill out the registry](#): The registry is very important for gaining a better understanding of Pitt Hopkins syndrome as well as a critical tool for current and future clinical trials. Make it a priority to fill out the registry during the month of September! Get started [here](#).
 - [Join the global census](#) to help us get an accurate count of how many individuals in the world have been diagnosed with Pitt Hopkins. This quick survey only takes 2-3 minutes.
 - [Join Invitae](#) and the natural history study. More details below in the newsletter as well.
- 3. Share on social media!**
 - Share your story on social media with family and friends
 - Tag the [PHRF Facebook page](#) in your September PTHS Awareness Day posts
 - On September 18 be sure to wear your Pitt Hopkins gear and share a picture with us! When you share on Facebook, Twitter and/or Instagram, be sure to use [#pithopkins #beyondawareness](#) to show how you are going beyond awareness this month!
- 4. Get decked out in Pitt Hopkins gear!**

We have an entire Pitt Hopkins apparel collection with RareTees. 100% of all sales support Pitt Hopkins Syndrome research initiatives. [Start shopping now!](#)
- 5. Donate to transform lives!**

If you are inspired by the amazing individuals living with Pitt Hopkins syndrome, we ask you to consider making a donation to fund life-changing research today.

Donate Today

And make sure to join us on social to follow all of our updates in real time! Including research updates, clinical trial information, Sibling Saturday, Warrior Wednesday, Fundraising Friday and MORE!

Follow us now! >>> [Facebook](#), [Instagram](#), [LinkedIn](#), [Twitter](#)

La Jolla Labs Investigating the Feasibility of ASO therapy in Pitt Hopkins Syndrome

Research Update

We are thrilled to [announce](#) our new partnership on a very exciting project with [Mahzi Therapeutics](#) and [La Jolla Labs](#). This project is focusing on ASOs and their use in Pitt Hopkins syndrome (PTHS).

ASOs are an established class of drug showing increasing success in treating neurological diseases. To determine if ASOs would make good therapeutics for PTHS, the PHRF has arranged a collaboration with Mahzi Therapeutics and La Jolla Labs. Currently, this project is in the very early phases of scientific development, working to establish proper biological systems to evaluate this strategy. Once experimental conditions are worked out, this collaboration will test the ability of ASOs to restore the protein levels of TCF4 and effectively reverse the cause of PTHS.

ASOs are currently being explored in several Angelman syndrome trials and the idea of this ASO, if successful, will be to upregulate the good copy of the TCF4 thereby increasing the protein overall.



NEUREN PHARMACEUTICALS

Clinical Trial Update

The Neuren NZ-2951 Clinical Trial for Pitt Hopkins is underway, with many participants having started treatment. **All clinic locations are still recruiting, so if you are interested in participating see below:**

- Neuren site details [here](#)
- ClinicalTrials.gov details [here](#).
- Please also see [our website](#) for information.



PITT HOPKINS CENSUS

Please join!

A question that comes up often is, *how many individuals in the world live with Pitt Hopkins syndrome?* It is a difficult question to answer, but **gathering this information is vital for research**. Pharmaceutical companies are highly motivated to know this number and have asked us to help ascertain it.

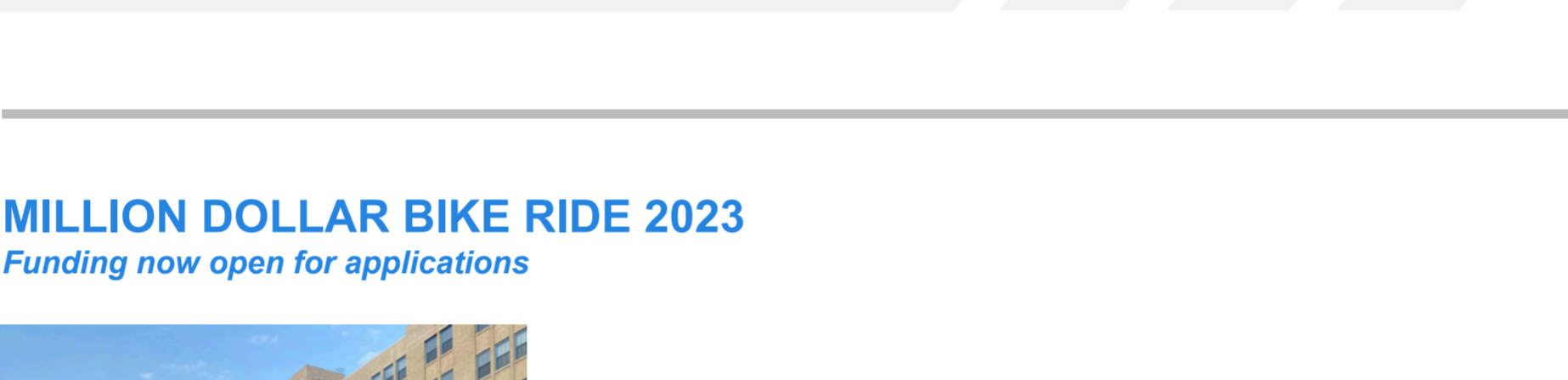
We currently have **1,188 individuals in the world** registered! We know there are many more out there, so please, if you haven't already joined - [do so now!](#) It only takes a few minutes. And to make this easier, we have added the survey in multiple languages:

- **English:** <https://forms.gle/ESrUNeGNabyBbbK6>
- **French:** <https://forms.gle/AeEFbdQsSSBUoJ57>
- **Portuguese:** <https://forms.gle/yvC5VnfSsAhbnLic7>
- **Spanish:** <https://forms.gle/3y5j06GAm9LoR7V4A>

More details about the information gathered, including a break down of countries, type of diagnosis and age, can be seen on our website www.pithopkins.org/census.

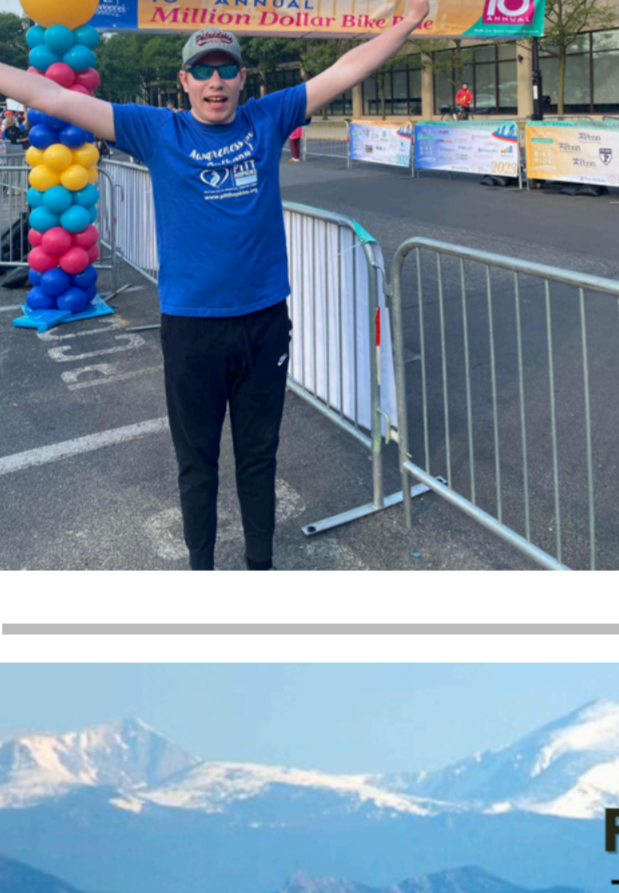
BY COUNTRY	62 countries	
Albania - 1	Finland - 2	Norway - 7
Argentina - 1	France - 56	Peru - 2
Australia - 8	Germany - 62	Poland - 5
Austria - 34	Greece - 1	Portugal - 9
Austria - 1	Hungary - 3	Puerto Rico - 1
Belarus - 1	India - 7	Romania - 1
Belgium - 6	Indonesia - 2	Russia - 18
Bosnia-Herzegovina - 1	Ireland - 8	Saudi Arabia - 1
Brazil - 60	Israel - 8	Serbia - 3
Canada - 45	Italy - 17	Singapore - 4
Chile - 9	Japan - 1	Slovenia - 3
China - 225	Jordan - 2	Spain - 29
Columbia - 4	Kazakhstan - 1	Sweden - 10
Cyprus - 1	Korea - 3	Switzerland - 11
Czech Republic - 4	Liechtenstein - 1	Taiwan - 2
Denmark - 4	Malaysia - 1	Turkey - 29
Dominican Republic - 3	Mauritius - 1	Ukraine - 4
Dubai - 1	Mexico - 8	United Kingdom - 50
Ecuador - 3	Netherlands - 8	United States - 405
Egypt - 2	New Zealand - 10	Uruguay - 1
El Salvador - 1		

Parents and/or Legal Guardians, We are asking you to please take a couple of minutes and fill out this quick, easy and very important survey to help us get a more accurate census of diagnosed Pitt Hopkins patients worldwide.



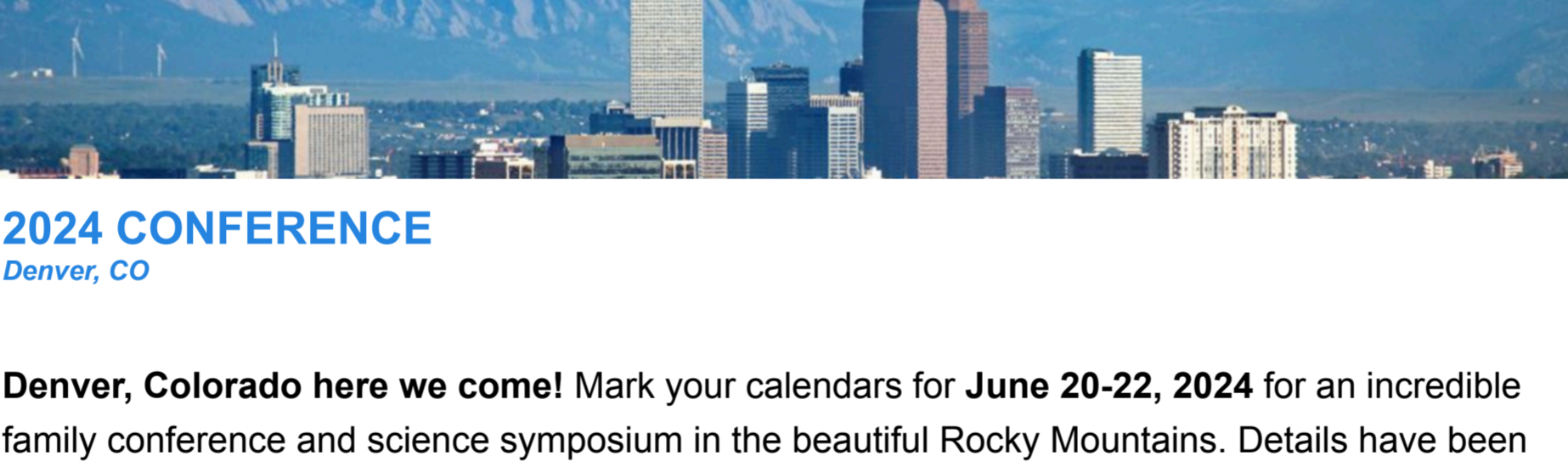
MILLION DOLLAR BIKE RIDE 2023

Funding now open for applications



THANK YOU to all of our MDBR donors for 2023. We reached our goal and now have a Pitt Hopkins research grant available!

Currently accepting Letters of Interest (due Sept 15th) for one \$73,473 pilot grant through the 2023 #MDBR Pilot Grant Program. **Apply here:** bit.ly/MDBR2023RFA. Grants are open to the international research community.



2024 CONFERENCE

Denver, CO

Denver, Colorado here we come! Mark your calendars for **June 20-22, 2024** for an incredible family conference and science symposium in the beautiful Rocky Mountains. Details have been updated on our conference website so that families can start planning for our best conference yet!

Details here: <https://pithopkins.org/pthsconference2024/>.

COMBINEDBrain - 2023 Roadshow

See locations below

Interested in participating in biomarker research?
COMBINEDBrain is traveling to a conference near you

COMBINEDBrain is a non-profit consortium of over 60 patient advocacy groups for rare, genetic neurodevelopmental disorders. They recognize the need to collect patient samples for researchers to identify biomarkers to be used to treat *idempol* treatments for our children. They are on a mission to collect ~500 samples from our member organizations over the next 6-8 months and *Insect CB Foundation* is one of them. Samples collected from our community will be stored and available to researchers across the world. Are you or one of your family members interested in participating in this exciting project for *Insect CB disorder*? Here are the basics:

Who: Any participant diagnosed with a CB Disorder or unaffected sibling.

What: COMBINEDBrain will be collecting urine samples and blood samples (processed for plasma and a finger stick) to be stored in the CB Biorepository and available for select biomarker projects as well as other interested researchers. They will also collect several online surveys to be completed by caregivers.

Where: COMBINEDBrain is collecting samples all across the country this year. Please see attached list of locations to find the closest biorepository collection to you. You may attend ANY of these meetings

Sample Size: Our goal is to collect 20 samples from each patient group

Please email us at management@pithopkins.org if you are interested in participating, and we will assist you in registering for this research opportunity.

Interested in participating in research?

COMBINEDBrain is traveling to a conference near you

GLUT1 Deficiency, August 28th 2023 (Noblesville, IN)
 IRF2BPL Foundation, Sept 22nd 2023 (Cincinnati, OH)
 KCNQ2 Cure Alliance, Sept 29th-30th 2023 (Chicago, IL)
 Rory-Belle Foundation, Oct 1st 2023 (Rogers, MN)
 USP7/Prader Willi, Oct 5th-6th 2023 (Denver, CO)
 TBR5 Community, Oct 12th-13th 2023 (San Antonio, TX)
 COMBINEDBrain Meeting, Oct 16th 2023 (Washington DC)
 FAM117A, Oct 29th (Mercer Island, WA)
 ADNP Kids Research Foundation, Oct 30th-Nov 1st (Los Angeles, CA)
 FAST Research Foundation, November (Miami, FL)
 SYNGAP1 Research Fund, Dec 1st-3rd 2023 (Orlando, FL)
 SLC6A1 Connect, Dec 1st-3rd 2023 (Orlando, FL)

Microbes for the Mind

Article featured in ASU's Alumni Magazine, ASU Thrive



We are honored to be highlighted in ASU's alumni magazine ASU Thrive this month. We are incredibly grateful for Dr. Adams Dr. Kraymalnik-Brown and ASU for conducting the Microbiota Transfer Therapy, the first ever clinical trial for Pitt Hopkins. We are now moving forward with additional studies, using a more accessible route for more individuals with Pitt Hopkins, with the hopes of bringing this treatment to the community at large.

"The results have been so promising that the team acquired the FDA's orphan drug designation and rare pediatric disease designation for Pitt-Hopkins syndrome, and following a Phase 3 study, they hope to receive FDA drug approval for their treatment."

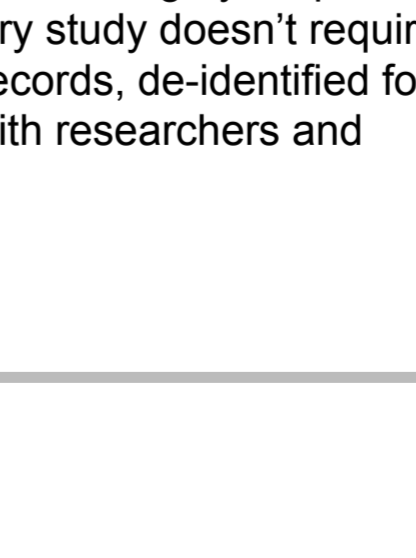
SPECIAL THANKS to our ASU cover girls Nicole and Alexandra (PTHS, 8 yo) Anderson!

Read full article [here](#).

Partnership with Invitae's Rare Patient Network

Get involved today

We are excited to share our partnership with Invitae's Rare Patient Network with you all! We have joined many other rare disease groups that have been using Citizen to build high-quality natural history data in a fraction of the time. Researchers and pharmaceutical companies have already used the data to submit Investigational New Drug (IND) filings with the FDA and this will help us work toward this for Pitt Hopkins as well.



Registering takes only a few minutes, and the Rare Patient Network team will collect all your or your loved one's medical records. You will receive full access to the records through your personal portal and can share them with whomever you choose. This natural history study doesn't require any clinical visits. The data will be extracted from your existing medical records, de-identified for your privacy and protection and, with your consent, organized to share with researchers and pharmaceutical companies.

[Get started here!](#)

PITT PARENTS, WE NEED YOU!

- **Join the Pitt Hopkins Registry**

We are proud to have partnered with CoRDS of Stanford to create this thorough and very important research initiative. The Pitt Hopkins registry is very important for many reasons, including helping us gather information for research, creating a central resource for researchers for more rapid recruitment of research participants, helping us to get an overall better picture of Pitt Hopkins syndrome and helping us connect with families. As we head toward clinical trials, this registry will also be an important tool to help with recruiting patients.

[Find out more >>](#)
- **Join the ASU Questionnaire**

We need your help in participating in a very important questionnaire with Arizona State University. We have 15 families enrolled, and we need 15 more to participate.

The Importance of this study: This will be the first formal validation of a comprehensive questionnaire for PTHS, which is needed for assessing changes in future clinical trials.

[Find out more >>](#)
- **Join Invitae**

Join our community on Invitae's Citizen Rare Patient Network today. It only takes a few minutes to sign up and you will get access to all your medical records in one place at no cost. US Patients only.

[Find out more >>](#)
- **Donate Cells to Coriell**

The NIGMS Repository is a research biobank. They collect samples from individuals with genetic diseases and make cell lines and DNA for scientists to use in their studies. These cells are being used to create iPSC lines and mini-brains (organoids) for our funded scientists to test medications on. More information on donating and how it can help PTHS research can be found here.

[Find out more >>](#)

Donate Today

