

PITT HOPKINS NEWSLETTER - July 2024

Pitt Hopkins Family Conference & Scientific Symposium

June 20-22, 2024 - Denver, CO



CONFERENCE RECAP: June 20-22, 2024

Denver, CO

The 2024 conference was our biggest one yet. Nearly 300 people from 11 different countries made their way to Denver, Colorado for an emotional reunion filled with research updates, understanding, and much-needed connection. It was so amazing to be with these incredible Pitt Hopkins families – our hope, our fuel, our family. It's hard to explain the special beauty of our Conference, but some of our families did a pretty great job. Here's what they had to say:

"Currently waiting in LAX to board our flight back to New Zealand. The conference feels life-changing to us, it was worth every minute of travel and every dollar! Thank you so much to the foundation & the board for the amazing organization & to all the families who attended! You're all family now! Even the ones we didn't get to meet yet! We are already planning to attend again next time and hopefully bring our other two children! Take care everyone, big love!" – Megan Nielsen, New Zealand

"I wanna give a huge shout to the Foundation members and all you special ladies for making these conferences happen! This was our first conference, and we will be trying to attend every one after this! I truly feel like we're part of the family. Not only are you all my role models and mentors now, but you're my friends. Life feels different... it's not as scary and I left feeling hopeful and accepted! Thank you ladies for all your hard work and dedication to making this journey a bit easier for the rest of us! Can't wait to meet again!" – Annie Dart, Wyoming

"Thank you so much for the best experience!! It was our very first conference and let me tell you it was like I've known all of you my whole life!! We felt all the love from everyone. My little Nayaah had the time of her life. Thank you again for making all this happen, we can't wait for the next one. We miss our Pitt family so much!" – Chaveli Rodriguez, Utah

"Dr. Jessica Duis, thank you for talking with me, and picking up the phone after a busy weekend at the conference. Thank you for your continued guidance, support, and knowledge. I'm so fortunate to have had the opportunity to have you on Finn's care team. I am sure that I am speaking for many Pitt Hopkins kiddos and families when I say that I appreciate you!" – Baillee Valstead, Colorado

"My heart is so full! Our second conference was nothing short of perfect. Thank you for organizing this for us. My biggest takeaway this year was the Pitt family aspect, including the Pitt sibling community specifically. All of our kids are like cousins and this is a lovely family reunion. Watching the panel of siblings made me realize how important it is for our other kids to support each other especially as they get older. These are relationships that will likely last their lifetime. This makes me happy. I love you guys!" – Chelsea Lopez, Nevada

We also want to share some resources for you that may be helpful from the conference. You can access this [document](#) with links and information. And find [recordings](#) here. Please note, the videos are password protected for confidentiality purposes - you will need to email us at information@pitt Hopkins.org to get the password if you would like to access those.

[Conference Info & Recordings](#)

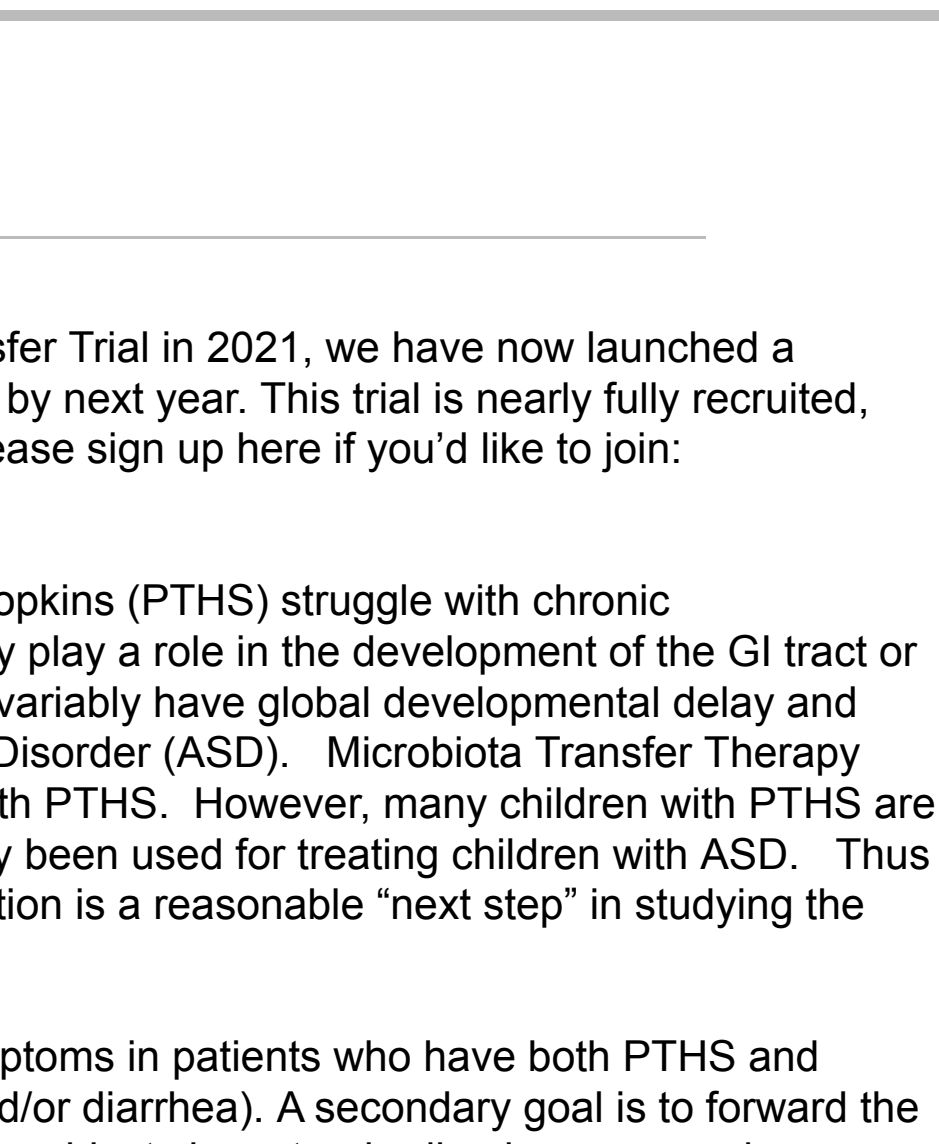
NEUREN PHARMACEUTICALS

We are extremely excited to share positive top-line results from our second ever clinical trial -- Neuren molecule NNZ-2591.

"Improvements were seen in clinically important aspects of Pitt Hopkins syndrome, including communication, social interaction, cognition and motor abilities."

Thank you to all the families who participated and to Neuren for making it happen!

[Read more here.](#)



MTT CLINICAL TRIAL UPDATE

Following the success of our first Microbiota Transfer Trial in 2021, we have now launched a second trial in the hopes of gaining FDA Approval by next year. This trial is nearly fully recruited, with a few spots still open for those interested! Please sign up here if you'd like to join:

<https://pitt Hopkins.org/mtt2/>

MORE ABOUT MTT: Many individuals with Pitt Hopkins (PTHS) struggle with chronic gastrointestinal dysfunction, suggesting TCF4 may play a role in the development of the GI tract or enteric nervous system. Individuals with PTHS invariably have global developmental delay and many also meet the criteria for Autistic Spectrum Disorder (ASD). Microbiota Transfer Therapy (MTT) has not previously been used for people with PTHS. However, many children with PTHS are also diagnosed with ASD, and MTT has previously been used for treating children with ASD. Thus this PTHS population with associated gut dysfunction is a reasonable "next step" in studying the efficacy of MTT.

The primary goal of this study is to reduce GI symptoms in patients who have both PTHS and chronic gastrointestinal problems (constipation and/or diarrhea). A secondary goal is to forward the field of PTHS research by characterizing the study subjects in a standardized manner and evaluating for genotype-phenotype correlations.

AWARENESS DAY 2024

PITT HOPKINS RESEARCH FOUNDATION
AWARENESS DAY 2024 SHIRT FUNDRAISER



To order click on the link below:
<https://www.jeanmckay.com/pitt-hopkins>

All orders must be placed by August 31st or prices will go up.
Orders will end August 20th.

ALL orders will ship by September 11th to arrive by September 18th...
AWARENESS DAY!



There is so much research to continue. There are funds that MUST be raised. This, my friends and families is up to US. Please join me in supporting the PTHS Research Foundation this September and let's bring more awareness to Pitt Hopkins. When you purchase your shirts, know that you are supporting research, you are supporting a better future for ALL of our kids! It takes a village. Please join and be a part of that village TODAY!

Awareness day 2024 will be here before you know it - so get your gear ordered now!

SHOP NOW

To order click on the link below:
<https://www.jeanmckay.com/pitt-hopkins>

ORDER TODAY!

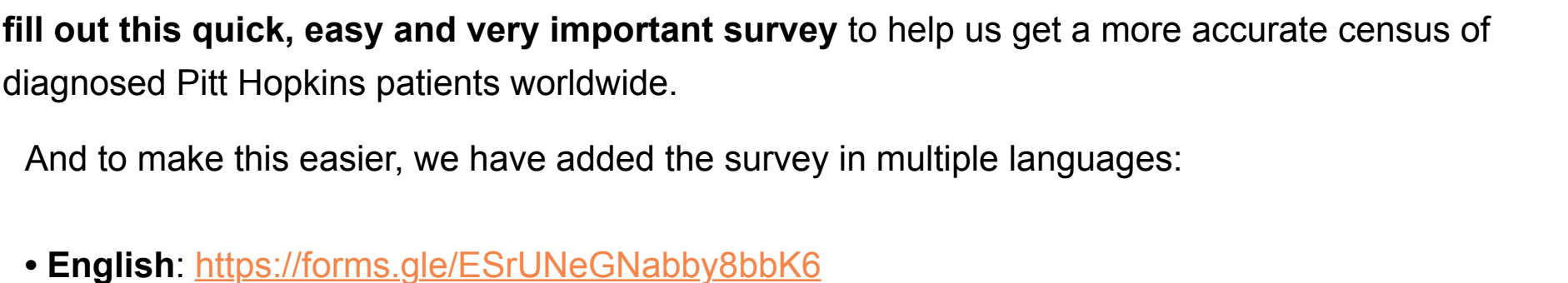
PITT HOPKINS CENSUS

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.

As of June 30, 2024, we have **1,426 individuals in the world** registered! Some quick and interesting facts:

- **1,363 diagnosed with Pitt Hopkins syndrome**
- **49 diagnosed with Pitt Hopkins-Like syndrome (1 or 2)**
- **14 clinical diagnosis of Pitt Hopkins Syndrome**
- **67 different countries**
- **48 states in the USA**
- **3 months old is the youngest registered individual**
- **51 years old is the oldest registered individual**

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



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.

And to make this easier, we have added the survey in multiple languages:

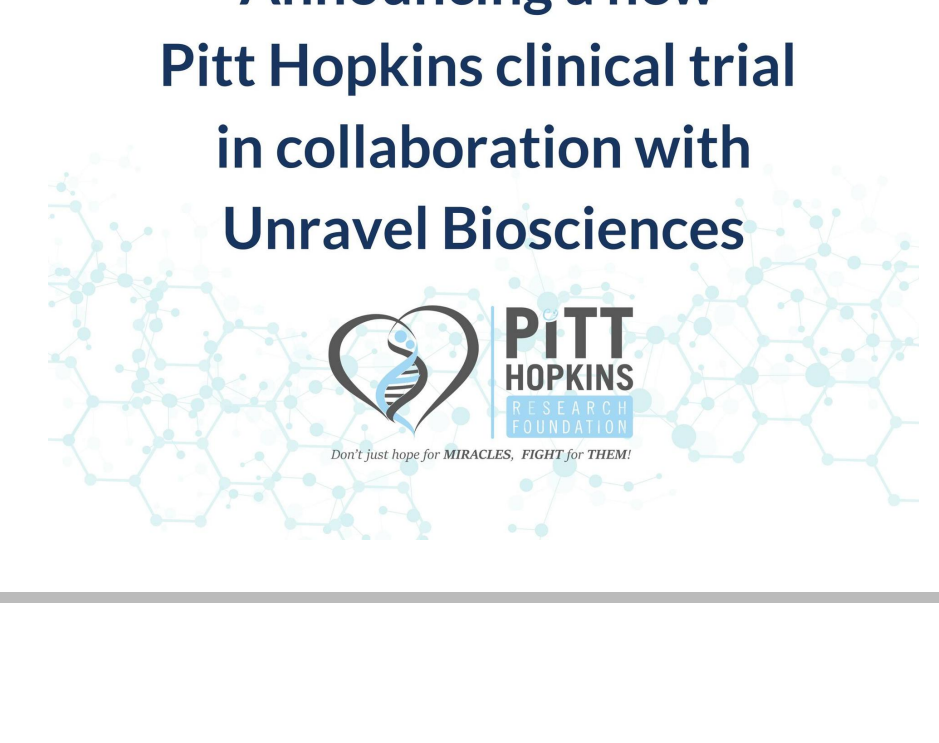
- **English:** <https://forms.gle/ESrUNeGNabby8bbK6>
- **French:** <https://forms.gle/AeFbdQsSSBUoJ57>
- **Portuguese:** <https://forms.gle/yvC5VnfSsAbbnLc7>
- **Spanish:** <https://forms.gle/3ysjo6GAm9Lor7V4A>

FILL OUT SURVEY NOW

TAKES LESS THAN 3 MINUTES!

ANNOUNCING A NEW CLINICAL TRIAL WITH UNRAVEL BIOSCIENCES

We are thrilled to announce the Pitt Hopkins Research Foundation's third ever clinical trial in conjunction with Unravel Biosciences.



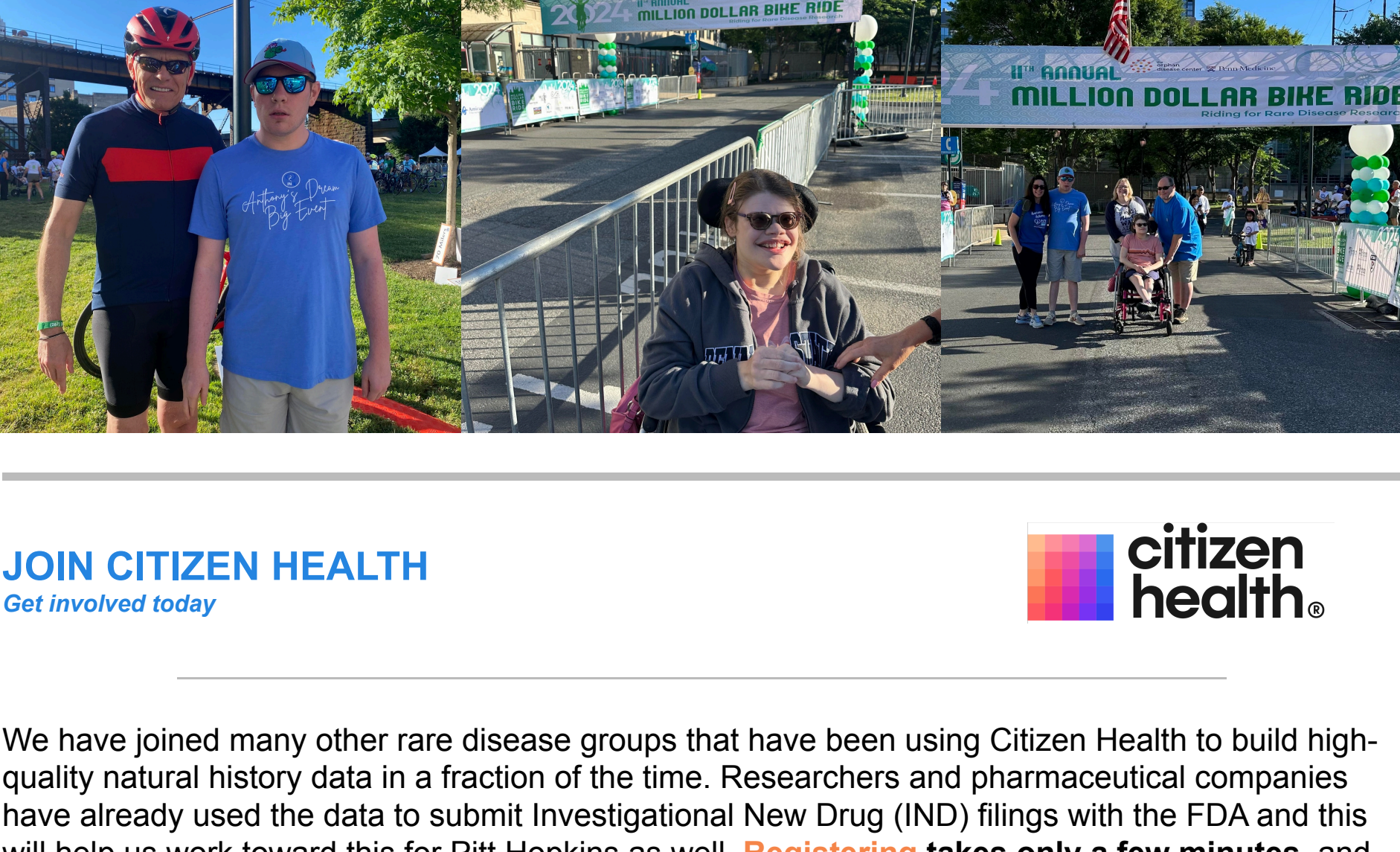
We will partner with the Unravel team to explore the use of Vorinostat, an already FDA-approved small molecule that has shown promise in other rare diseases. This study will evaluate efficacy and safety in a small cohort in Colombia.

[Read more here.](#)

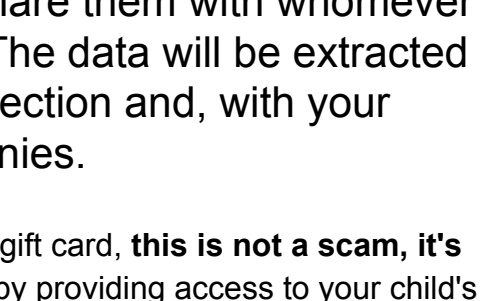
MILLION DOLLAR BIKE RIDE

June 8, 2024 – We had such a great morning at the Million Dollar Bike Ride 2024. A huge Thank You to the families who participated and fundraised - **we are so appreciative of you all representing and raising \$56,000 for Pitt Hopkins research!**

If you are a researcher interested in applying for the 2024 grant, watch our social media for announcements and check out milliondollarbikeride.org for more information.



JOIN CITIZEN HEALTH



We have joined many other rare disease groups that have been using Citizen Health 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.

*Please note: Pitt parents - if you recently received an email from Citizen Health with info about a gift card, this is not a scam, it's for real! You are receiving a gift card because you participated in the digital natural history study by providing access to your child's medical records. If you join Citizen Health and choose to put your de-identified data towards vital research efforts, and your data is licensed to a commercial researcher, you're eligible to share in that revenue! Pitt Hopkins patients included in the latest commercial research efforts are eligible to receive up to \$400 each.

If you haven't joined and are interested in finding out more information, please visit our website at: <https://pitt Hopkins.org/citizenhealth/>

[Get started here!](#)

JOIN PARENTS, WE NEED YOU!

• **Join the Pitt Hopkins Registry**

We are proud to have partnered with CoRDS of Stanford for this reason, including helping us gather information 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 Citizen Health**

Join our community on the 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](#)



Our Contact Information
 {{(Organization Name)}}
 {{(Organization Address)}}
 {{(Organization Phone)}}
 {{(Organization Website)}}
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