PITT HOPKINS RESEARCH FOUNDATION

Don't just hope for MIRACLES, FIGHT for THEM!

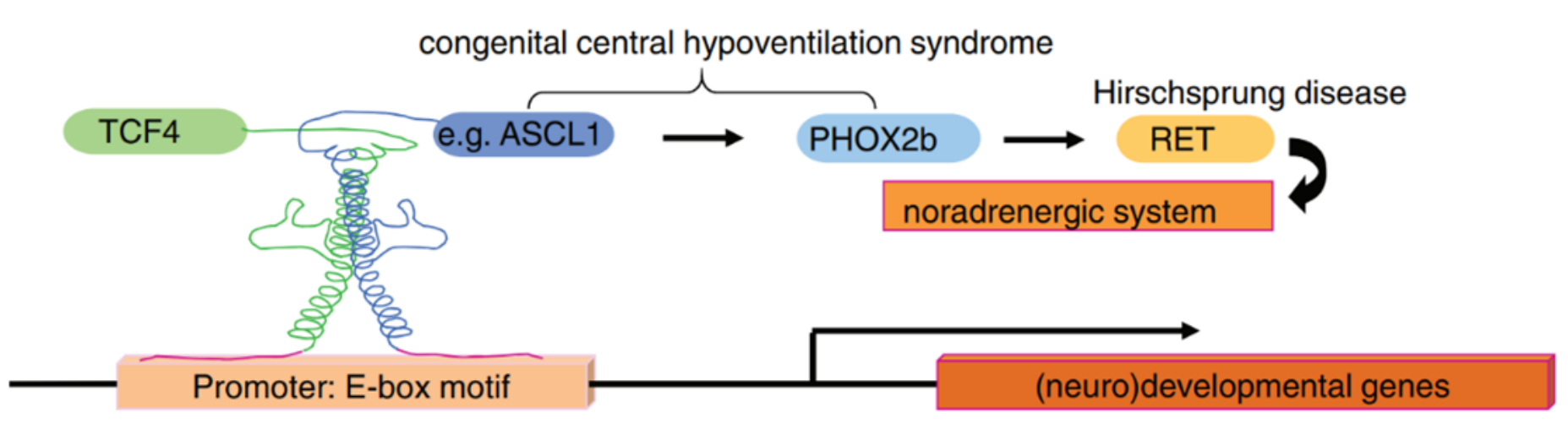
PITT HOPKINS NEWSLETTER - February 2024

2024 CONFERENCE
Denver, CO

Less than 5 months away! We are incredibly excited for you to join us at our family conference and science symposium in the beautiful Rocky Mountains. The hotel link has been updated (note, we had to change venues) to start booking your rooms and registration will open very soon. Check out our website for all the latest info!

Click here to visit the conference website!

TCF4, The Gene Behind Pitt-Hopkins Syndrome
Perla Sandoval & Allison Brown, Authors
November 29, 2023



La Jolla Labs wrote a wonderful article in *Genome*, detailing Pitt Hopkins Syndrome.

Pitt-Hopkins syndrome is a rare, genetic, neurodevelopmental disorder caused by haploinsufficiency of TCF4 that leads to several different physiological complications including intellectual disabilities and developmental delays.

Introduction

Pitt-Hopkins syndrome (PTHS) is an ultra-rare genetic disorder affecting at least 1,286 people worldwide. PTHS is caused by a mutation in the TCF4 gene, which impairs TCF4 protein function, causing a number of negative downstream effects. Patients with this condition will often experience developmental delays, GI issues, intellectual disabilities, seizures, breathing problems, problems with their motor skills, distinctive facial features, and may also struggle with social and communication issues associated with autism spectrum disorder [1]. Currently, there is no cure for PTHS, however, some symptoms can be managed through specialized health care and personalized education plans [2]. In this article, we will dig into the clinical and genetic details of PTHS, as well as the history and future of PTHS and PTHS patient care.

CONTINUE READING HERE

Zoom call with Dr. James Adams
February 6, 2024

Join us
Tuesday, February 6th at 8:00 p.m. EST
for a chat with Dr. James Adams!

Tune in for the details on our newest clinical trial, starting recruitment this month!
Also hear more about his work with Pitt Hopkins and where your fundraising dollars go.

You will need to register in advance for this call here:
https://us06web.zoom.us/meeting/register/tZctcumspjopE9FhafP2C-KlmhtyO6CoFLK5

Once confirmed, you will receive an email with the Zoom link for Tuesday. A recording will be sent to everyone who has registered following the call.

Register Here

TUESDAY, FEBRUARY 6, 2024
8:00 PM EST
Join our live Zoom call with
Dr. James Adams
Why?
Dr. Adams and ASU conducted the Microbiota Transfer Therapy, the first ever clinical trial for Pitt Hopkins Syndrome. He will help explain how it went and where we are headed.
Who is Dr. Adams?
President Professor at Arizona State University, where he directs the Autism/Asperger's Research Program
Why Join the Zoom?
PTHS parents can hear directly from our researcher
Understand his work
See where your fundraising money goes
Gain insight on ways to fundraise going forward
www.pitthopkins.org
PITT HOPKINS

Rare Disease Day
February 29, 2024

Rare Disease Day February 29th, 2024 is an observance held on the last day of February to raise awareness for rare diseases and improve access to treatment and medical representation for individuals with rare diseases and their families. This year it falls on a leap year, which is actually the rarest day of the year.

To find all filters, cover pages, and all profile picture backgrounds use this https://www.rarediseaseday.org/downloads/

We will post our ideas on Facebook and Instagram soon!

NEUREN PHARMACEUTICALS
Clinical Trial Update

We are pleased to announce the Neuren Phase II study in the treatment of PTHS closed enrollment in November 2023, with top-line results from the trial expected to be available in Q2 2024.

Thank you to the families that have participated to make this possible!

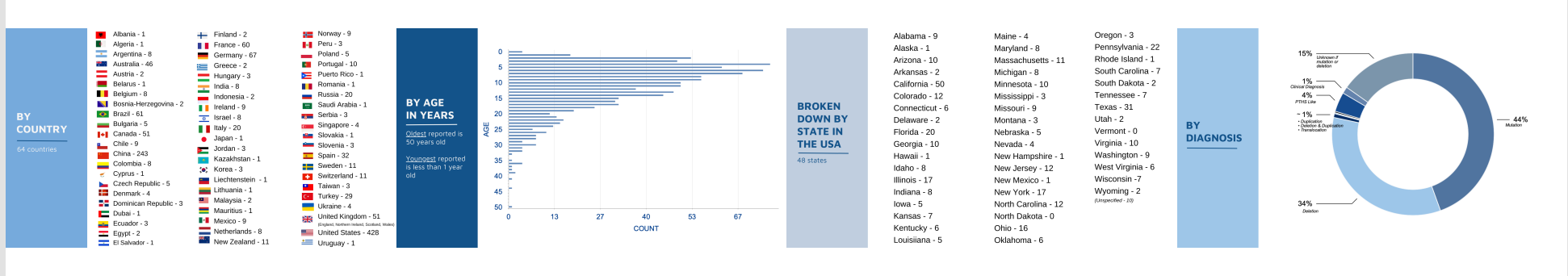
PITT HOPKINS CENSUS
Please join!

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.

As of December 31, 2023, we have **1,318 individuals in the world** registered! Some quick and interesting facts:

- **1,263 diagnosed with Pitt Hopkins syndrome**
- **41 diagnosed with Pitt Hopkins-Like syndrome (1 or 2)**
- **14 clinical diagnosis of Pitt Hopkins Syndrome**
- **64 different countries**
- **48 states in the USA**
- **3 months old is the youngest registered individual**
- **50 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.pitthopkins.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/AeEFbdQsSSBUpoJ57
- **Portuguese**: https://forms.gle/yvC5VnfSsAhbnLic7
- **Spanish**: https://forms.gle/3ysjo6GAm9LoR7V4A

MILLION DOLLAR BIKE RIDE
2023 Awardee + 2024 Date Announced

The 2023 MDBR grant awardee is Dr. Brady Maher. A grant of $73,473 will be awarded to the Maher lab for *Identification of vulnerable cell types and quantification of cell type-specific differential gene expression in the Pitt-Hopkins Syndrome mouse model*. Grant details, including a lay summary, can be found here.

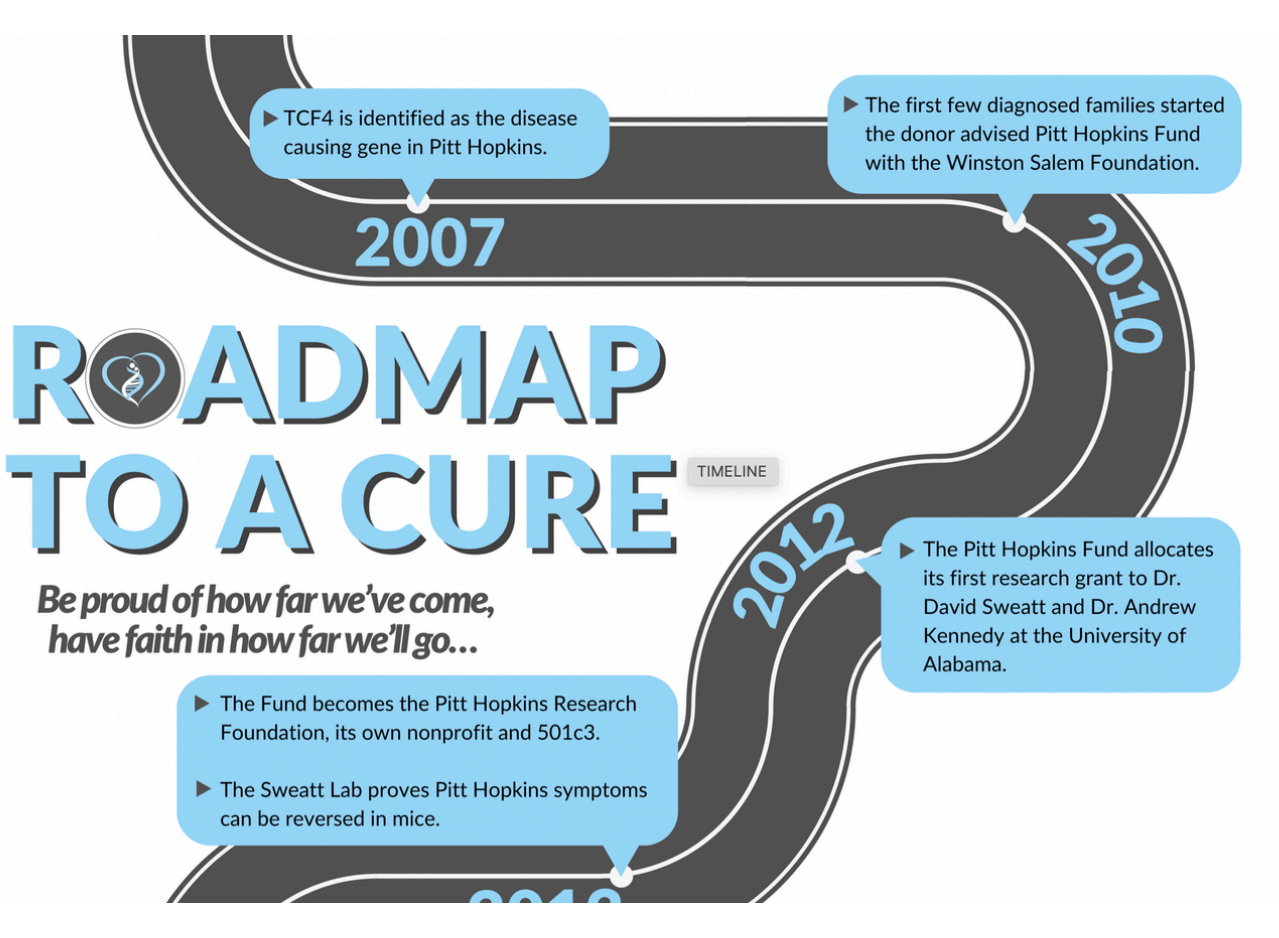
THANK YOU to everyone who helped raise money for this grant through the Million Dollar Bike Ride 2023!

Saturday, June 8, 2024 --

JOIN US IN PHILADELPHIA

for our biggest fundraiser of the year!

Roadmap to a Cure
10 Years of Progress!



TCF4 is identified as the disease causing gene in Pitt Hopkins.
The first few diagnosed families started the donor advised Pitt Hopkins Fund with the Winston Salem Foundation.
2007
2010
ROADMAP TO A CURE
TIMELINE
Be proud of how far we've come, have faith in how far we'll go...
2012
The Pitt Hopkins Fund allocates its first research grant to Dr. David Sweatt and Dr. Andrew Kennedy at the University of Alabama.
The Fund becomes the Pitt Hopkins Research Foundation, its own nonprofit and 501c3.
The Sweatt Lab proves Pitt Hopkins symptoms can be reversed in mice.

See full 10 years of progress here.

Join our Ciitizen Initiative
Get involved today

We have joined many other rare disease groups that have been using Ciitizen 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 Ciitizen
Join our community on the Ciitizen 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}}