



PITT HOPKINS

RESEARCH FOUNDATION

Connecting families. Driving research. Changing lives.

World-wide Family Registry • Pitt Hopkins Clinics & Medical Care • Cutting-edge Research • Advocacy & Awareness

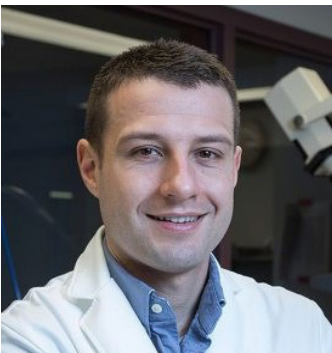
Our Vision and Pathway to a Cure *for Pitt Hopkins Syndrome*

mission

to support research dedicated to finding a treatment, and an eventual cure for Pitt Hopkins Syndrome and other similar disorders.



WE ARE BOLD. AND SO ARE OUR GOALS.



“The warp speed pace of [the PHRF] and research is simply unbelievable. Having the best universities in the country immersed in a friendly competition to be the one that cracks PTHS is an incredible feat for any foundation, let alone one that services an ultra rare syndrome like Pitt Hopkins.”

Dr. Andrew Kennedy | Assistant Professor of Chemistry & Biochemistry | Bates College



“We (scientists) don’t know of another group that has accomplished as much as you have in 4-5 years. You’ve accomplished more per patient than any parent organization many of us have ever dealt with.”

*Craig Powell, M.D., Ph.D. | Chair, Department of Neurobiology
The University of Alabama at Birmingham School of Medicine
Director, Civitan International Research Center*

What would you do if your child **could not speak?**
Or walk?

Or suffered with debilitating anxiety, seizures, gut issues, or motoric impairment.

What if they **depended on you to do everything** for them, even as they became an adult?

You would do anything to help them.

You would fight.

Now imagine a child that has all of those issues, rolled into **one devastating disease.**

It's called **Pitt Hopkins Syndrome...**

and this is what our **children are fighting,**
every single day. We need an army as strong and brave as
they are to tackle it. **To take it down for good.**

Join us in making
Pitt Hopkins **history...**

Our History and Why We Do This

The Pitt Hopkins Research Foundation (PHRF) was launched in 2012 to drive research toward a cure for the agonizing symptoms of Pitt Hopkins Syndrome. In the summer of 2013, the PHRF secured its 501(c)(3) status and became a nonprofit organization. We are proud to maintain our annual gold-level status, through GuideStar, demonstrating our commitment to transparency.

We are the only organization in the world devoted to funding research into Pitt Hopkins. Because the gene was only discovered in 2007, the research landscape was nonexistent when we started. Since our founding, we have awarded nearly \$3,000,000 to labs all over the world.

Our efforts have shown dramatic symptom reversal in mice, attracting attention from university and pharmaceutical scientists, as well as the National Institute of Mental Health.

An average of 95 percent of every dollar donated to the PHRF goes directly to our research initiatives. Our Foundation is powered entirely by volunteers, so that we can focus every penny on what matters most: helping our children as quickly as possible.

The Pitt Hopkins Research Foundation is defining the landscape of Pitt Hopkins research. We set the research strategy by proactively identifying and monitoring promising therapeutic areas, seeking out scientific and industry partnerships and working closely with them to advance programs.

We believe Pitt Hopkins can be cured, but time matters. A rigorous, coordinated, and agile research agenda will get us there as quickly as possible.

Our ultimate goal is to close our doors for good...

by finding a cure.



Pitt Hopkins Syndrome

is a rare, neurodevelopmental disorder caused by a mutation of the Tcf4 gene on the 18th chromosome.

Our children suffer from significant developmental issues, problems with motor coordination (ataxia) and balance, breathing abnormalities, and seizures.

Most never talk. Many never walk.

Pitt Hopkins affects all races and both genders equally. Currently there are approximately 1000+ people in the world diagnosed with this rare syndrome, however we believe there are many more who remain undiagnosed or misdiagnosed.

Pitt Hopkins syndrome is considered an Autism Spectrum Disorder. Because we know the actual gene that causes it, scientists believe that curing Pitt Hopkins syndrome will unlock answers for other neurological disorders.

The NIMH has supported our seed grants with more than \$5,000,000 in additional research dollars. They know, as do our scientists, how important this work is for finding a cure, not just for our children, but for other neurological diseases such as Autism, Schizophrenia, and Alzheimer's Disease.



Research is the future.
A cure is possible.
But, we need your support.

Key focus areas are:

- continue to transition from basic research to transformational, applied discoveries
- fund future clinical trials
- fund the study of clinical phenotyping/ outcome measures and potential biomarkers
- fund small molecule drug discovery program targeting TCF4 pathway
- fund additional clinicians and clinical researchers
- fund and broaden our gene therapy programs

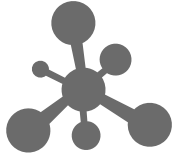
We don't just hope for miracles. We *fight* for them.

Join us and be part of the *miracle*.



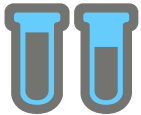
Science shows us a cure is possible and we have a solid plan to help get us there. But we can't do it alone. We will need the commitment and the support of generous donors around the world.

*We invite you to learn about
our strategic research plan.*



Cure

Our ultimate goal is to cure Pitt Hopkins, and close the doors of our Foundation for good. Currently we are exploring two approaches that attack the root cause of the disorder: TCF4. These approaches, pursued in parallel, are applicable to all TCF4 mutations and deletions.



Gene therapy - GENE THERAPY IS OUR LEAD PROGRAM. The concept behind gene therapy is simple: Delivery of healthy copies of the TCF4 gene to compensate for the faulty ones. We are fortunate to have some of the leading researchers in the world with regards to gene therapy tackling this research.

Protein replacement - We are working with researchers to deliver TCF4 proteins to the brain. This approach is already working in other disorders.



Treat

Although focused on CURE we also invest in small molecule treatments that have the potential to improve symptoms and quality of life. We are preparing to launch 3 clinical trials in this arena. Because time is precious, and our children need help now, we are also actively pursuing ALREADY FDA APPROVED COMPOUNDS that could significantly impact symptoms.



Enable

Currently, The Pitt Hopkins Research Foundation is single handedly responsible for creating the conditions that will enable the most impactful pharmaceutical and biotech industry investment in Pitt Hopkins research. To this end we have invested in:

1. Collecting human cells from patients to test new therapies in the lab.
2. Opening Pitt Hopkins Clinics so clinicians may gain a greater understanding of the disease.
3. Launching a comprehensive Registry/Natural History Study so that clinicians and pharmaceutical companies have a greater understanding of disease progression.



Understand

Science and technology is more promising than ever before. Our investments – and the investments of many other groups have lead to this critical and incredible juncture. We will continue to invest in the basic science to learn all we can about TCF4, novel therapeutic approaches and the latest scientific breakthroughs.

We are focused on two things: maximizing your dollars – and ours, and healing our children as quickly as possible.

Why care about Pitt Hopkins?



Pitt Hopkins Syndrome is a single gene disorder.

We know exactly what gene causes it....

Our research goes beyond rare. Single gene disorders like Pitt Hopkins are science's best hope of finding cures for diseases with no specific origin like Alzheimer's, Autism and epilepsy. Tcf4 is important to all of us.

It is also implicated in liver disease, corneal disease and schizophrenia.

Around the world, our partner labs and scientists are making life-changing discoveries.

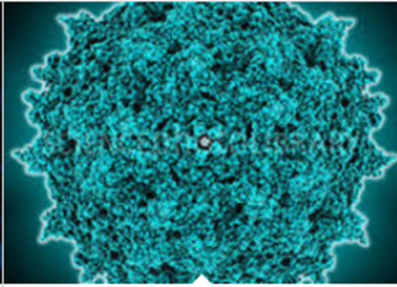
A cure is possible. **Help transform lives.**

Together, we will make Pitt Hopkins history.

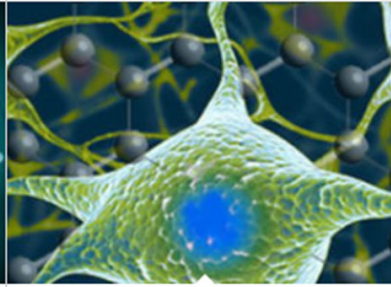
Current* sponsored research, funded by our generous donors.



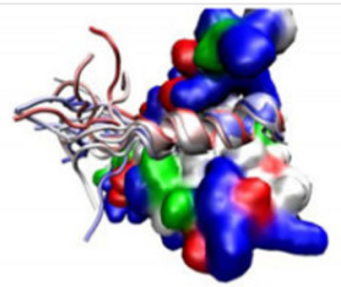
A human Pitt Hopkins Syndrome model for therapeutic proof of principle



PTHS Late Onset Gene Therapy Grant



Epigenetic Therapy to Treat Pitt-Hopkins Syndrome (PTHS)



Kindal Kivisto Award for Promising Young Researchers



Single Concentration Re-Purposing Screen...



A Model of Pitt-Hopkins Syndrome, and Efficacy Testing



Protein Replacement Therapy Grant



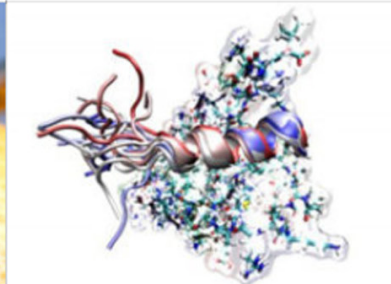
Drug Repurposing Project to Identify Potential Drug Candidates



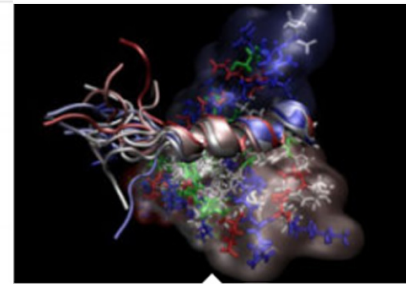
Ann D. Bornstein Gene Therapy Grant



Pharmacological Rescue and Screening-Pitt Hopkins Model



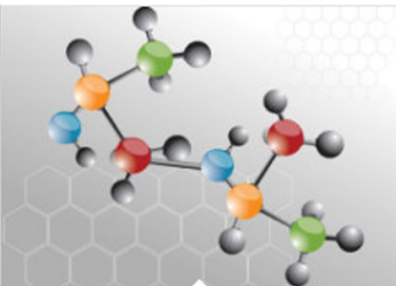
Identification of Direct Target Genes of TCF4 in Neurons



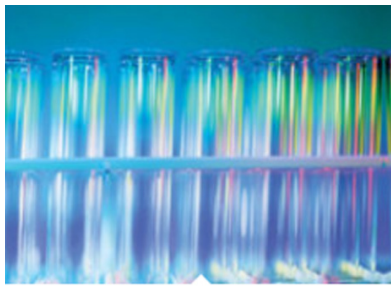
Characterization and Generation of PTHS Model Mice



Pilot Study to Identify Small Molecule Activators of TCF4



Sarah Huffman Award for Promising Young Researchers



Drug Validation Initiative-Compound Testing in Mice

Visit pitthopkins.org/research to learn more.



2018 PHRF Family Conference & Science Symposium

The Pitt Hopkins Research Foundation is committed to directly funding the most promising research available in the world to help find a treatment and ultimately a cure for Pitt Hopkins Syndrome.

Since 2012, the Pitt Hopkins Research Foundation has funded nearly \$3,000,000 in research grants. That funding has helped scientists create mouse models and neuronal stem cells to test drugs which could help ameliorate symptoms. Additionally, our scientists discovered two therapeutics that have reversed the symptoms of PTHS in mice. **We are working hard to bring these drugs to human trials, with your help, our hopes could be reality.**

Since we know the gene that causes Pitt Hopkins Syndrome (TCF4), we are in an important place to fund meaningful research. Research that could give insight to other learning, memory and motor disorders with no known cause, such as Alzheimer's, Autism, Epilepsy and Parkinson's disease.

Our community and children need your support.

DONOR INFORMATION (please print)

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 THE ADDRESS ABOVE IS MY: Home Business
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Please return this form to:
 Pitt Hopkins Research Foundation
 PO Box 6257 • Plymouth, MA 02362
 email: phrf-fundraising@pitt Hopkins.org

All funds will be deposited with the Pitt Hopkins Research Foundation.
 Your donation is tax deductible to the fullest extent of the law.

YES, I WISH TO SUPPORT A PATHWAY TO A CURE FOR PITT HOPKINS SYNDROME.

\$25,000 (Platinum Supporter) \$5,000 (Silver Supporter)
 \$7,500 (Gold Supporter) \$1,000 (Bronze Supporter)
 Other amount \$ _____
 Donate online here: pitt Hopkins.org/donate

My check, **payable to the Pitt Hopkins Research Foundation**, is enclosed.
 Please charge my credit card:
 AmEx Discover MasterCard VISA
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(This signature authorizes charges to above card number)

I pledge \$ _____. Please mail me a pledge statement.
 Matching gift company, if applicable: _____



PITT HOPKINS

RESEARCH FOUNDATION

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