Dear Friends,

Words can never truly express the gratitude we have for you, our donors. Your generous support helped us to build this foundation and allowed us to believe, that with courage, hard work and persistence, we could shape a better future for our children. Your donations mean so much more than money, they give all of us hope, and for that, we can never thank you enough.

From the day we started the PHRF in 2012, our foundation has worked tirelessly to reshape the future of Pitt Hopkins syndrome – from funding the most cutting edge labs, to cultivating top scientists and industry partners, to increasing international awareness and education. Our strategic approach makes the PHRF not only the leader in Pitt Hopkins research, but a leader in rare disease research. We are proud and humbled that many fledgling rare disease foundations now come to us for advice on how to get the job done.

In the last six years we have raised nearly $4 million for groundbreaking research; research the NIH deemed promising enough to augment with more than $5 million in matching funds. Beyond helping our children, the NIH realizes that Pitt Hopkins research could hold answers for many other neurological diseases, like Alzheimer’s, Autism, Schizophrenia and more.

In 2019, others stood up and took notice as well. For the first time, pharmaceutical companies
such as Healx and Neuren were coming to us, initiating pipeline projects for Pitt Hopkins. And finally, after just six years of pre-clinical research, we are the process of launching our first ever clinical trial for microbiota transfer therapy at Arizona State University — a huge milestone for our small foundation.

We feel a great responsibility to our donors and our families to make the most of every dollar donated. As president, this weighs heavily on me, and my commitment to you is to make smart, focused decisions where every dollar is maximized, where research does not languish in the lab, but instead gets translated as quickly as possible to potential treatments.

Thanks to you, we have so much to be grateful for. However, there is still much to be done. First and foremost, I am the mother of a child with Pitt Hopkins syndrome. I want so much more for all of our kids than the life they have been dealt. For my son and all the other courageous children and families dealing with this debilitating syndrome, our fight continues. We can't stop. And we won't stop.

Thank you, as always, for joining us in this fight.

Audrey Davidow Lapidus
President
Pitt Hopkins Research Foundation

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

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. Our Foundation is powered entirely by volunteers, so that we can focus every penny on what matters most: helping our children and adults as quickly as possible.
What would you do if your child could not speak? Or walk? Or suffered with debilitating anxiety, seizures, gut issues, or motor 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.

Pitt Hopkins Syndrome (PTHS) is a rare genetic disorder affecting a specific gene on chromosome 18, called TCF4. It is characterized by significant developmental delays, problems with motor coordination and balance, breathing abnormalities, and seizures. Most individuals with Pitt Hopkins do not develop functional speech. The gene that causes Pitt Hopkins was only discovered in 2007. Currently there are approximately 1000+ people in the world diagnosed with this syndrome. We believe there are many more.
1,000+ known* diagnosed cases

— *Worldwide. It is estimated that there are many more undiagnosed or misdiagnosed. Other scientific estimates project that between 1 in 34,000 and 1 in 41,000 are living with PTHS.

2007 year TCF4 gene is linked

— First described by D. Pitt and I. Hopkins in 1978. The connection to TCF4 was not established until 2007 when it was found to play an essential role in the development of the nervous system and the brain.

Since Inception

7 years as a 501(c)3

$4M in research grants funded

40+ grants funded

1st clinical trial in 2020

$5M in NIH grants awarded for PTHS research
$500,000
raised by donors this year

$500,000
research grants funded

— Exact figure is $493,775

$500,000
raised by donors this year

— Exact figure is $493,775

In 2019

$0
spent on staff salaries

$1.68M
in NIH grants awarded for PTHS research

9
funded research labs

60+
# of family fundraisers
Our Commitment
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.

Our Focus
We do more than just hold fundraisers and write checks. We are defining the landscape of Pitt Hopkins Syndrome 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 through the drug development pipeline. 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 Progress
Since our beginning in 2012, we have funded nearly $4,000,000 and over 40 research grants to scientists. In just seven years, scientists discovered not one, but two therapeutics that have reversed the symptoms of PTHS in the mice. We are working hard to bring these drugs to trial. Since our efforts have shown dramatic symptom reversal in mice, we have attracted attention from university and pharmaceutical scientists and the National Institute of Mental Health (NIMH). The NIMH has supported our seed grants with more than $5,000,000 in additional research dollars.

We are the only organization in the world devoted to funding research for PTHS.
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
In 2019, we announced our first-ever human clinical trial to attempt to eradicate the severe gastrointestinal effects of Pitt Hopkins Syndrome. In partnership with Arizona State University’s Autism & Asperger’s Research Program, a clinical trial is scheduled to begin in 2020 focused on Microbiota Transfer Therapy (MTT), also known as a fecal microbiota transplant.

“This clinical trial is a huge milestone for our young foundation,” said Audrey Davidow Lapidus, president of the Pitt Hopkins Research Foundation and mother to Calvin, diagnosed at one-years-old with PTHS. “Not only do we hope the trial will bring some much-needed relief for our families living with PTHS, it signals the continued growth of the Pitt Hopkins Research Foundation as a serious player in the medical research field," she added.

“Our research team and I are honored to be working with the Pitt Hopkins Research Foundation, and we hope that our microbiota transplant therapy is able to improve the chronic constipation and other gut problems in children with Pitt Hopkins,” said Prof. James Adams.
When the Pitt Hopkins Research Foundation funds pilot grants to our world’s leading researchers, we hope that our researchers can secure additional funding based on their successes.

In 2019, the National Institute of Neurological Disorders and Stroke (NINDS) announced that they have funded the lab of Ben Philpot, PhD, Kenan Distinguished Professor of Cell Biology and Physiology at the UNC School of Medicine for $1.68 million over five years to create a therapy to treat children with Pitt Hopkins Syndrome.

“We are extremely excited to receive this funding, which will allow us to move one step closer to a transformative therapeutic agent or a gene therapy approach to treat Pitt Hopkins Syndrome,” said Philpot, who is the associate director of the UNC Neuroscience Center and member of the Carolina Institute for Developmental Disabilities. “Unfortunately, there are no treatments for the core symptoms of this condition. But we are working hard to change that.
2019 Funded Research

TCF4 gene activation therapy for Pitt Hopkins Syndrome

Dan Wang, Ph.D. (Assistant Professor, Horae Gene Therapy Center, University of Massachusetts Medical School); Guangping Gao, Ph.D. (Professor, Horae Gene Therapy Center, University of Massachusetts Medical School)
Total amount requested: $74,721

A human Pitt-Hopkins Syndrome model for therapeutic proof of principle II

Alysson R. Muotri, PhD
UC San Diego School of Medicine
| 2019-2020
Total amount requested: $150,000
Generation, validation, storage, and dissemination of an induce pluripotent stem cells (iPSC) collection derived from Pitt Hopkins Syndrome patients

Brady J. Maher Ph.D.; Lead Investigator, Lieber Institute for Brain Development, Asst. Professor, Psychiatry and Behavioral Sciences, Asst. Professor, Dept. of Neuroscience, Asst. Professor Dept. of Biochemistry, Cell and Molecular Biology, Johns Hopkins School of Medicine, 2018-2019
Total Amount Requested: $87,140

Characterization of the expression of TCF4 mRNA and protein isoforms during the development of rodent and human brain

Tonis Timmusk, PhD
Tallinn University of Technology, Estonia | 2019-2020
Grant Awarded: $50,000

None of this research would be possible without all our donors that believe in a better future for our children.
2019 Funded Research, continued

Identification of small molecules upregulating TC4F in PTHS

Ben Philpot, PhD
University of North Carolina at Chapel Hill | 2019
Awarded from the 2018 MDBR for 2019: UPenn Million Dollar Bike Ride Grant $50,000

Tissue-specific and temporal reinstatement of Tcf4 function to treat Pitt Hopkins Syndrome

Andrew Kennedy, PhD
Bates College | 2019
Awarded from the 2018 MDBR for 2019: UPenn Million Dollar Bike Ride Grant $50,000

PTHS Late Onset Gene Therapy Grant

Ben Philpot, PhD, University of North Carolina at Chapel Hill, and Steve Gray, PhD, University of Texas Southwestern
UT Southwestern: 2018 - 2020
Total Amount Requested: $123,970
Assessment of re-introduction of TCF4 gene on Pitt Hopkins Syndrome mouse model in vivo

Patricia Cogram, Ph.D – IEB, University of Chile Team; Dr. Robert Deacon: intracranial and intravenous injections and behavior evaluation; Dr. Ariel Avila: in utero electroporation and electrophysiological studies; Dr. Chris Rex, AFRAXIS, Neuronal spine morphology and density

Total Amount Requested: $76,600 | 2019 – 2020

Differential analysis of genome-wide TCF4 binding in neural progenitor cells derived from Pitt Hopkins Syndrome patients and controls

Joseph L. McClay, PhD
Virginia Commonwealth University | 2019-2020
Grant Awarded: $50,000

Full details can be found at: pitthopkins.org/research
### Financial Snapshot

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<tr>
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<th>Amount</th>
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<tbody>
<tr>
<td><strong>Total Assets</strong></td>
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<tr>
<td><strong>Total Liabilities</strong></td>
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<td><strong>Net Assets</strong></td>
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### Revenue

<table>
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<tr>
<th>Revenue Source</th>
<th>Amount</th>
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<tr>
<td>Contributions</td>
<td>$485,117</td>
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<tr>
<td>Investment Income</td>
<td>$317</td>
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<tr>
<td>Other Revenue</td>
<td>$8,341</td>
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**Total Revenue** $493,775

### Expenses

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<tr>
<th>Expense Type</th>
<th>Amount</th>
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<tr>
<td>Grants Paid</td>
<td>$504,411</td>
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<tr>
<td>Board Salaries</td>
<td>$0</td>
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<tr>
<td>Administrative</td>
<td>$27,691</td>
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</table>

**Total Expenses** $532,102

An average of 94 percent of every dollar donated goes directly to research initiatives.

Revenue Distribution:
- Contributions: 98%
- Investment Income: <1%
- Other Revenue: 2%

Expenses Distribution:
- Grants Paid: 95%
- Administrative: 5%
- Board Salaries: 0%

All figures are from 2019 Audited Financial Statement and IRS Form 990.
Fundraisers Hall of Fame

- Rafa's 4th Annual Fundraiser, in honor of Iris Eyres
- Cecily's Smile Squad Campaign, in honor of Cecily DiNuzzo
- Flatbread Social for PTHS Awareness Day, in honor of Cecily DiNuzzo
- 5th Annual Love for Lakely Golf Tournament, sponsored by Aegis Technologies Group, in honor of Lakely Stapler
- 3rd Annual PHRF Open, in honor of Calvin Lapidus
- Charity Buzz Fundraiser, in honor of Calvin Lapidus
- Awareness Day Arizona Family Fundraiser, in honor of all Arizona kids
- RareiTees for a Cure Apparel, funding research in honor of all PTHS individuals
- Alexandra's Pitt Crew Campaign, in honor of Alexandra Anderson
- 6th Annual Texas Trekkers, in honor of Ashton Green and all Texas kids
- Oktoberfest: Pints for Pitt Hopkins, in honor of Rylie Maginn
- 5th Annual Pauca-Plemmons Letter Campaign, in honor of Victor Pauca
• Anthony’s 7th Annual Awareness Day, in honor of Anthony Mirigliani
• CH Robinson 14th Annual Northeast Regional Golf Classic, in honor of Anthony Mirigliani
• Paint for a Cause, in honor of Sofia Izzi
• CycleBar Charity event, in honor of Sofia Izzi
• Sofia’s Story Campaign, in honor of Sofia Izzi
• Childhood Cancer Society Fundraiser with split proceeds, in honor of Logan Bomar
• Ella’s Savers Fundraiser, in honor of Ella Schilling
• Cornhole for a Cause, in honor of Jonathan Ackerman
• Play date for Pitt Hopkins, in honor of JohnWesley Holley
• Team John Wesley Campaign, in honor of JohnWesley Holley
• Luton Park Time Trial Bike Race, in honor of Emma Schmucker
• Edith’s Lost Boys Campaign, in honor of Edith Sherman
• Mighty Macy Campaign, in honor of Macy McDonell
Million Dollar Bike Ride Teams
2019 Penn Medicine Orphan Disease Center

- Team Alexandra Anderson
- Team Noah Anderson
- Team Emma Boyce
- Team Iris Eyres
- Team Conner Fitzgerald
- Team Elliot Hill
- Team April Loveday-Brown
- Team Rylie Maginn
- Team Brendan Marlin
- Team Anthony Mirigliani
- Team Landon Patterson

Facebook Fundraisers over $1000

- In honor of Alexandra, by Nicole & Matt Anderson
- In honor of Alivia, by Amie Ramos
- In honor of Blake, by Jessica Willett Gipson
- In honor of Calvin, by Eric & Audrey Lapidus
- In honor of Christoffer, by Agnes Tyldum
- In honor of Ian, by Jessica & Trevor Fletcher
- In honor of Isa, by Nazia Khan
- In honor of Logan, by Brooke & David Bomar
- In honor of Mia, by Crystyl Mallozzi
- In honor of Nicolas, by Mauricio Loria
- In honor of Oron, by Elie Appleson Hale
- In honor of Sarah, by Nicole Dyehouse
- In honor of Sofia, by Stephanie & Tyler Izzi
Facebook Fundraisers between $500 and $999

- In honor of Adaline, by Heather St Pierre Johnson
- In honor of Alex, by Laura Ownby Barido
- In honor of Ben, by Andrea Hoyle
- In honor of Blake, by Vandana Arcot
- In honor of Brae, by Leyna Scarberry Vaughter
- In honor of Harper, by Amanda & Eric Strasburg
- In honor of Hunter, by Tammy Gross
- In honor of Isabel, by Martin Cuevas
- In honor of Luke, by Julie Spealler
- In honor of Maddie Zarleno, by Matt DePersis
- In honor of child with PTHS, by Ingrid Muri

All other fundraisers:
Thank you to the many families who have held fundraising campaigns and events. There are so many that we could not list them all! You have contributed a great deal to research this year!
The Pitt Hopkins Research Foundation is served by two boards; the **Board of Directors** and the **Scientific Advisory Board**. Together, we are working hard to promote awareness of Pitt Hopkins Syndrome and bring practical treatment into current medical practice as quickly as possible.

The Board of Directors is comprised of individuals dedicated to advancing research and supporting families of children and adults with Pitt Hopkins Syndrome. The Board is actively engaged in fundraising efforts toward research, in providing parental support to families worldwide, and in offering communication and media representation for the Foundation. Our board and officers are parents and professionals who volunteer their time, without compensation for their service, so that an average of 94% of all funds donated go to finding a cure.

### Leadership

**Officers and Executive Committee**

- **Audrey Davidow Lapidus**, President
- **Theresa Pauca**, Vice President
- **Jessica Fletcher**, Vice President
- **Heather Maginn**, Secretary
- **Diane Sellew**, Treasurer

### Board Members

- **Audrey Davidow Lapidus**
- **Eric Lapidus**
- **Theresa Pauca**
- **Paul Pauca**
- **Heather Maginn**
- **Jack Maginn**
- **Jeff Davidow**
- **Jessica Fletcher**
- **Diane Krell**
- **Traci Green**
- **Claudette Mirigliani**
- **Nicole Anderson**
Scientific Advisory Board

The Science Advisory Board for the Pitt Hopkins Research Foundation is comprised of top Scientists around the country who are able to offer their expertise in advising the PHRF Board of Directors. Duties include reviewing grant proposals and budgets and offering expert opinions about the direction of the Foundation in research into Pitt Hopkins syndrome.

Chief Science Officer | Sarah Harris, Ph.D., Drug Development and Translational Medicine Consultant

- **Dr. Marcelo Wood**, Ph.D., University of California, Irvine, Associate Professor; Director, Interdepartmental Neuroscience Program; Department of Neurobiology and Behavior; Center for the Neurobiology of Learning and Memory

- **Dr. Craig M. Powell**, M.D., Ph.D., Associate Professor; Ed and Sue Rose Distinguished Professorship in Neurology; Departments of Neurology & Neurotherapeutics, Psychiatry; University of Texas Southwestern Medical Center

- **Dr. Stanley Nelson**, M.D., Professor in Residence, Human Genetics; Director, DNA Microarray Technology; Co-Director, Center for Duchenne Muscular Dystrophy; University of California Los Angeles

- **Dr. Eric Klann**, Ph.D., Professor, Center for Neural Science, New York University

- **Dr. Joel Gottesfeld**, Ph.D., Professor of Molecular Biology, Scripps Research Institute, Associate Editor, Journal of Biological Chemistry

- **Dr. Mary Jacena Leigh**, M.D., Developmental Behavioral Pediatrics, University of California Davis, MIND Institute
Scientific Advisory Board, con’t

- **Dr. Pat Levitt**, Ph.D., Provost Professor, University of Southern California, Dept. Pediatrics; WM Keck Chair in Neurogenetics, Keck School of Medicine; Director-Program in Developmental Neurogenetics; Institute for the Developing Mind, Children’s Hospital Los Angeles; Director-Neuroscience Graduate Program

- **Dr. Joachim Herz**, M.D., Professor, Thomas O. and Cinda Hicks Family Distinguished Chair in Alzheimer’s Disease Research; Departments of Molecular Genetics, Neurology & Neurotherapeutics, Neuroscience; Graduate Program in Integrative Biology, University of Texas Southwestern Medical Center

- **Dr. Shafali Jeste**, M.D., Principal Investigator/Assistant Professor in Psychiatry and Neurology, UCLA Center for Autism Research and Treatment (CART)

- **Dr. Michael Tranfaglia**, M.D., Medical Director, Chief Scientific Officer, FRAXA Research Foundation

- **Dr. Sean Ekins**, Ph.D, DSc, CSO, Hereditary Neuropathy Foundation, Adjunct Professor, Division of Chemical Biology and Medicinal Chemistry, UNC Eshelman School of Pharmacy, University of North Carolina at Chapel Hill; Adjunct Professor, Robert Wood Johnson Medical School, University of Medicine and Dentistry, New Jersey (Now Rutgers University); Adjunct Associate Professor, School of Pharmacy Department of Pharmaceutical Sciences, University of Maryland

- **Dr. Robert H. Ring**, Ph.D, Chief Executive Officer, Kaerus Bioscience Ltd

- **Dr. Aaron C. Gerlach**, Ph.D, Director, Icagen, Inc.

- **Peter Lloyd-Thomas**, M.Eng, MBA, Management Consultant, Independent Researcher in the Translation of Autism Science into Therapy
Support future research initiatives at: pitthopkins.org/donate
“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