Dear Friends,

As 2020 draws to a close, we are looking back on a remarkable year. Despite the pandemic and the obstacles thrown in our path, there are so many reasons for hope and optimism. This has been one of the busiest and most productive years on record for CHI.

Thank you for all you do to support the work of CHI, advancing research for better treatments and cures, raising awareness of HI to prevent brain damage and death, and being there for HI families every step of the way.

We hope you enjoy our year in review!

We started the year off on a high note when we learned that CHI was the recipient of a Rare As One Project grant to establish the CHI Collaborative Research Network, funded and organized by the Chan Zuckerberg Initiative (CZI). “No one is more committed to finding cures for rare diseases than the patients and families of those affected by these disorders,” said Priscilla Chan, Co-Founder & Co-CEO of CZI. Today, there is an explosion of knowledge and ways to share and collaborate. The Rare as One Network provides resources and structure to maximize the amazing opportunities brought to us by science, technology, and brilliant, passionate minds. We are so fortunate to have such a fantastic community of supporters, CHI families, HI researchers and clinicians, and Board of Directors that led us to this moment.
CHI gathered at the New Jersey State Museum in Trenton for the 10th Rare Disease Day Event in Trenton, NJ. People living with rare diseases, their family members, rare disease advocates, legislators, members of the biotech, pharma, and other industry supporters all came together to share information, network, and learn from each other. We spent time together at the New Jersey State Museum and visited the NJ State House as a group to hear the proclamation read stating Feb 24, 2020 as Rare Disease Day in NJ! We are so grateful to the NORD Rare Action Network for providing us with the support to put on such an event. A big shout out to BioNJ, HINJ, NORD, and Soligenix who also sponsored this event once again.
On Saturday, February 29, Thrivent and HI Dad Paul Castellano and his family hosted an Ice Cream Social and fundraiser to raise awareness about HI. HI advocates Hannah and Ben Raskin-Gross, spoke at the event about life with HI.

The CHI staff also hosted CHI Board members, doctors and parents, for Rare Disease Day on Saturday, February 29, to discuss exciting developments for HI research, awareness and support in the near future. And we introduced our newest team member Tai Pasquini, PhD, Research and Policy Director to the CHI community!
Though the event was virtual this year, we were able to rally our support around Team CHIbra (and our new mascot!) to participate in the Million Dollar Bike Ride, from afar, for the 7th year! Through matching funds from Penn Medicine we were able to offer a $72,000 hyperinsulinism research grant for 2020! 

Every year, funding from the Million Dollar Bike Ride gives an opportunity for new HI research to take place. We announced the winner of the 2019 Million Dollar Bike Ride research grant, Thomas Smith, PhD, of the University of Texas.

**RESEARCH TOPIC:**

Towards new therapeutic treatments for the hyperinsulinism/hyperammonemia syndrome (HI/HA)

Dr. Thomas Smith and his research team are using a wide array of techniques to develop drugs to treat the hyperinsulinism/hyperammonemia (HI/HA) syndrome. HI/HA is one of the more common genetic hyperinsulinism subtypes. Patients suffering from HI/HA have a higher basal level of insulin in their blood and over secrete insulin upon consumption of protein. This disease not only affects the pancreas but is also linked to seizures and developmental problems. These pathologies are due to mutations in glutamate dehydrogenase (GDH) that affect how the enzyme is regulated. While diazoxide is effective at controlling insulin secretion, it comes with side effects and does not target GDH and therefore does not treat the other affected organs.

Our goal is to target GDH directly to treat all symptoms associated with HI/HA throughout the body. To this end, we have been creating the various HI/HA mutants to better understand their subtle differences. We are collaborating with a pharmaceutical company and have performed more than a half million screens looking for active compounds. The company is helping us to analyze and optimize activity. As soon as possible, we will test the best compounds in whole cell and animal assays.

**AWARD RECIPIENT:**

Thomas Smith, PhD

**HOME INSTITUTION:**

University of Texas Medical Branch

**AWARD AMOUNT:**

$72,014
The HI Global Registry 2020 Annual Report was released in July and provides insight into the HI experience as reported by participants of the HI Global Registry (HIGR). HIGR is the first global patient-powered congenital hyperinsulinism patient registry and consists of a series of thirteen surveys made up of questions related to a patient’s HI experience over their lifetime. The analysis and descriptive statistics shared in the report are based on the responses that participants provided from HIGR’s launch in October 2018 through February 2020. This includes individuals living in 45 countries and participants ranging from just a few weeks of age to 58 years old. You can learn more here.

This fall, the HI Global Registry celebrated its second birthday. HIGR tracks the experiences of those who live with HI, making it possible to quantify and characterize life with HI in a rigorous, scientific fashion to support new research leading to treatments and cures. In our second year of operation, 389 people with HI have registered from 49 countries and every inhabited continent joined HIGR.
FIRST VIRTUAL CONFERENCE

CHI hosted our First Virtual Congenital Hyperinsulinism Family Conference on July 11-12, 2020 from 10:30 AM to 3:00 PM (EDT) each day. 420 people from 33 countries, a true worldwide HI community, participated in the Conference. 38 speakers contributed to the live presentations, interactive Q & A, and small group discussions in breakout rooms. Participants learned all about HI through presentations from specialists, panel discussions, small group interactive sessions, and the sharing of family stories. You can learn more and see presentations from the conference here.

SUGAR SOIRÉE

In support of people with HYPERINSULINISM
The 2020 Sugar Soirée, which took place virtually on the evening of October 24, was a wonderful celebration of the HI Community. It was a spectacular event bringing together many of CHI’s most beloved supporters from around the world. We were once again charmed by host Tony Dokoupil, the co-host of CBS This Morning. We were honored to be joined by Dr. David Fajgenbaum, author of *Chasing My Cure, Turning Hope into Action* during our VIP Cocktail Reception for a compelling conversation he had with Tony, about his life and work with his own rare disease, Castleman’s Disease.

We were thrilled that we could also honor the following people during the Soirée:

- **The Be My Sugar Awareness Award** was given to Drs. Tatjana Milenkovic and Rade Vukovic of the Department of Pediatric Endocrinology of Mother and Child Healthcare Institute of Serbia
- **The Be My Sugar Extraordinary Volunteer Award** was given to Sunny Chapel of Ann Arbor Pharmaometrics, Inc

We want to thank our honorary chairs, Dr. Diva D. De León-Crutchlow and her husband Michael Crutchlow, and Dr. Paul Thornton and his wife, Ricci Thornton, board member Turaya Bryant-Kamau who lent her professional expertise and helped produce the virtual event, our benefit committee and our junior committee who all went above and beyond to support our event this year.

Please click [here](#) to read more about the 2020 Sugar Soirée.
We are thrilled to announce the launch of the Congenital Hyperinsulinism International (CHI) Hyperinsulinism Centers of Excellence (COE) Program. The CHI COE Program will designate specialist facilities providing the highest quality of care for hyperinsulinism (HI) patients and their families around the world.

Every year, around the globe, thousands of babies are born with HI. They need appropriate and expert care to reduce the risk of brain damage and death and to ensure proper management of their disease for the best possible quality of life, as do all children and adults living with HI.

There are some superb institutions known for providing excellent HI care and participating in groundbreaking research, yet until now there has not been a review process or certificate awarded to institutions for the care of those with HI.

We plan on announcing the recipients of this designation on Rare Disease Day 2021. You can read more about the program [here](#).

This program was made possible through a grant from [Global Genes](#).
During the CHI Collaborative Research Network Virtual Convening, on December 5-6, we began the work of creating a patient-led sustainable, lasting structure to prioritize and execute an international research agenda to better understand congenital hyperinsulinism (HI), identify new ways to improve the lives of those living with HI, reduce the incidence of irreversible brain damage, detect the genetic causes of HI types not currently known; and to find cures for each and every type of HI.

Led by our CHI CRN Team: Patient Leader Julie Raskin, Lead Researcher Diva D. De León-Crutchlow, MD, Lead Clinician Paul Thornton, MD, and Team Members Tai Pasquini, PhD, Davelyn Hood, MD, MBA and Jennifer Schmitt, we were joined by more than 100 people from 32 countries.

You can read more about the 2020 CHI CRN Virtual Convening [here](#).

**EXETER GENETIC TESTING PROGRAM**

We continued our support for the genetic testing partnership project with the University of Exeter in the UK. Those suspected of HI from anywhere in the world, who otherwise would not be able to afford it, received genetic testing for HI to determine the type of treatment each baby or child with HI needs. Joining forces with Exeter has enabled 432 individuals from 51 countries to access genetic testing. Getting the right diagnosis and treatment can help avoid brain damage and other severe complication.
CHI posters, “What is Hyperinsulinism” and the “Signs and Symptoms of Hypoglycemia” are now available in 21 languages - the most recent addition is Czech! The posters are a fantastic way to raise awareness of HI all over the world. We’re always looking to expand the collection, so please drop us a line if you know someone with the expertise to translate the posters into more languages. See all languages here. https://congenitalhi.org/chi-posters/

THANK YOU TO OUR 2020 SPONSORS

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Thank you to the CHI Board of Directors for their continuous dedication to the CHI cause and hard work this year.

- Davelyn Hood, President
- Sheila Bose, Vice President
- Mason Smith, Treasurer
- Margaret Maher, Secretary
- Erin Greaves
- Matthew M. Hopkins, PhD
- Turaya Bryant Kamau
- Julie Sheldon
- Dina Tallis
- Pam Williams

We cherish each and every member of our community: HI families and our sweetest volunteers and most generous donors.

Wishing you all very happy holidays and a wonderful new year. We’re gearing up and ready for an exciting 2021!

Give a Gift to CHI Today!

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