



Dare to Dream:

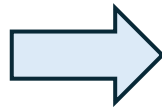
Patient-Powered Research and Advocacy Milestones



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PhD**
*Parent/CHI Board/Nurse & Research Manager
Congenital Hyperinsulinism International*

What is patient-powered research?

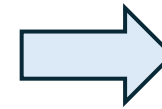
Traditional research model



Researchers
make a discovery
in the lab

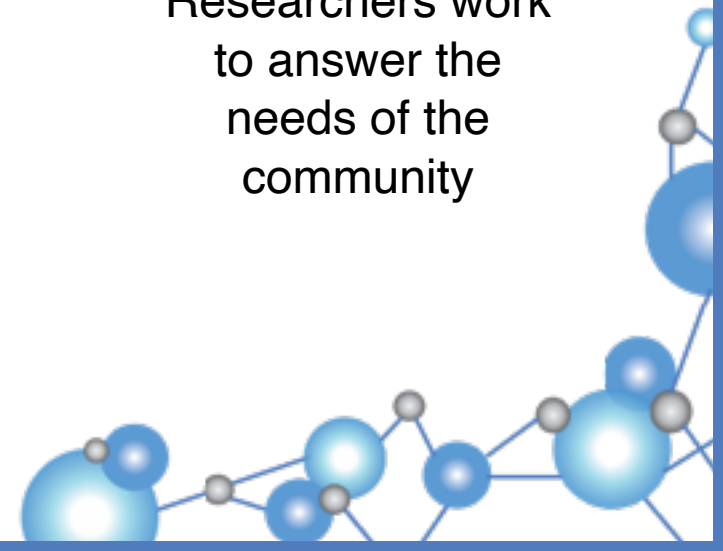
How could this
apply to a group
of people?

Patient-powered research model



Individuals from the
community identify
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needs

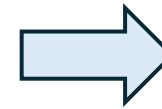
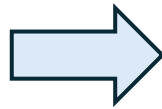
Researchers work
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Traditional research model

Patient-powered research
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How has the patient-powered approach influenced HI research and advocacy?

2005 – Congenital Hyperinsulinism International (CHI) is founded

- CHI was founded by a small parent group sharing their challenges with HI and has grown to a global community of people with HI and their families.
- This is the **35th family conference** hosted by CHI!

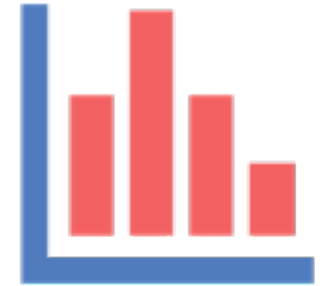


Group photo from 2025 Family Conference in Copenhagen, Denmark



2018—HI Global Registry is

The only patient-powered international registry for hyperinsulinism research



Qualified researchers can request **deidentified** data for studies to learn more about HI, develop new treatments, and improve care



Surveys

Completed online by people with HI or their parent / caregiver



Glucose Monitoring Data

- ✓ Glucose meters (Glucometers)
- ✓ Dexcom Continuous Glucose Monitors (CGMs)



Physician Form

Completed by the participant's physician (with participant/caregiver permission only)

Funded by the European Union
LightCure - 101080327



2018 – The Open Hyperinsulinism Genes Project Begins

- CHI partners with the University of Exeter to provide genetic testing to those in need.
 - Participants who receive genetic testing are invited to enroll in research to improve knowledge of HI genetics.
- Since 2018, 1,088 participants from 64 countries have been tested.
- 52% have received a cause for their HI. The samples provided through this program contributed to the confirmation that variants in the gene *HK1* cause HI, which others had suggested in the past.

Non-coding variants disrupting a tissue-specific regulatory element in *HK1* cause congenital hyperinsulinism

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2020 — CHI Establishes the Collaborative Research Network

- CHI established a Collaborative Research Network (CRN) for HI in 2020 thanks to a grant from the Rare As One program from the Chan-Zuckerberg Initiative.
- The CHI CRN is a diverse group of over 60 academic and industry researchers, clinicians, and expert patient advocates from over 19 countries.
- To accelerate tomorrow's breakthroughs, the CRN has extended research collaboration far beyond the traditional boundaries.



Gap Analysis for Research Progress

CRN members worked virtually over 2 years to identify gaps in knowledge & resources

Identified >360 individual deficits across **7 key areas**:



Genetics



Diagnosis



Glucose Monitoring



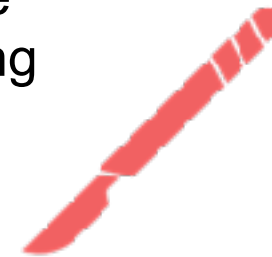
Care Guidelines & COEs



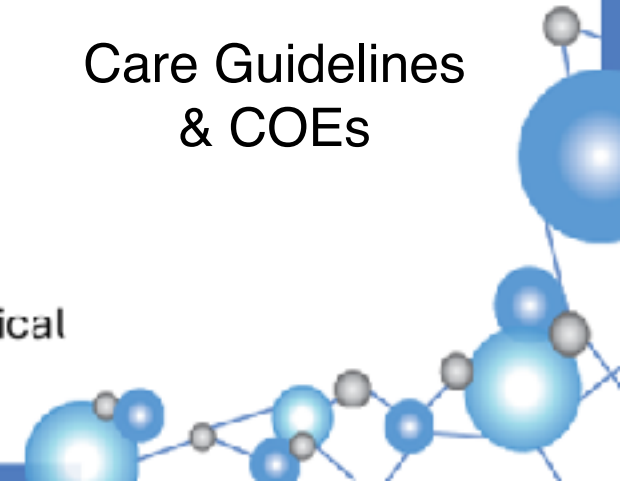
Clinical Trials & Industry Engagement



Nomenclature

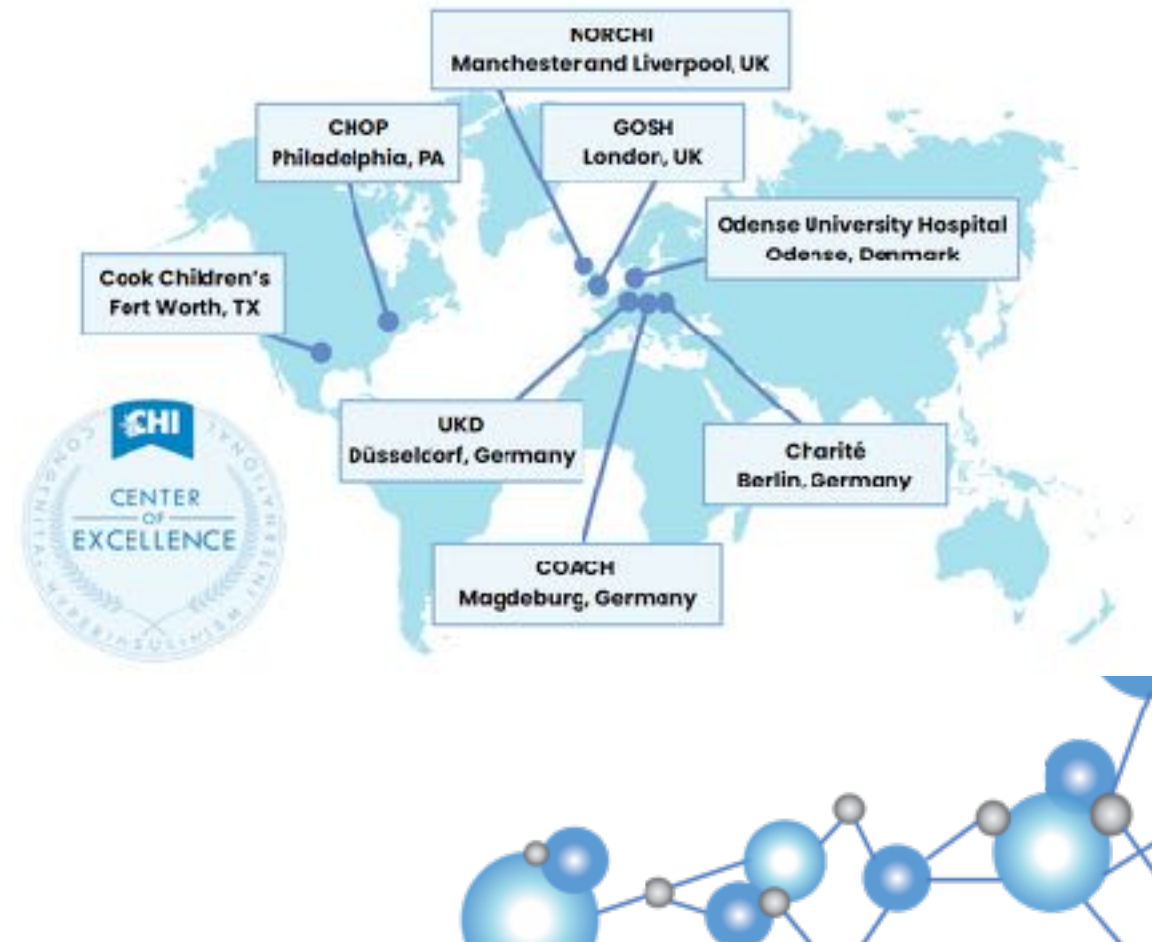


Medication & Surgical Management



2020—CHI Launches the Centers of Excellence Program

- HI is a complex condition requiring proper management to ensure the best possible standard of care.
- The CHI COE program designates specialist facilities providing the highest quality of care for HI families around the world.
- Recognizing COEs helps people with HI know where to go for multidisciplinary care and treatment.
- The first COEs were named in 2021.
 - In 2024, 8 centers were designated as COEs.
- Before the CHI COE program, there was no review process or certificate awarded to institutions for the care of those with HI.



2023—International Guidelines for the Diagnosis and Management of HI

- The guidelines were developed so that people with HI can receive evidence-based care rooted in agreement by the world's leading HI experts.
- CHI was consulted during the process of developing the guidelines to include the HI community voice.
 - CHI's Chief Research Officer, Tai Pasquini, is an author on the guidelines.

GUIDELINES | JULY 14 2023

International Guidelines for the Diagnosis and Management of Hyperinsulinism

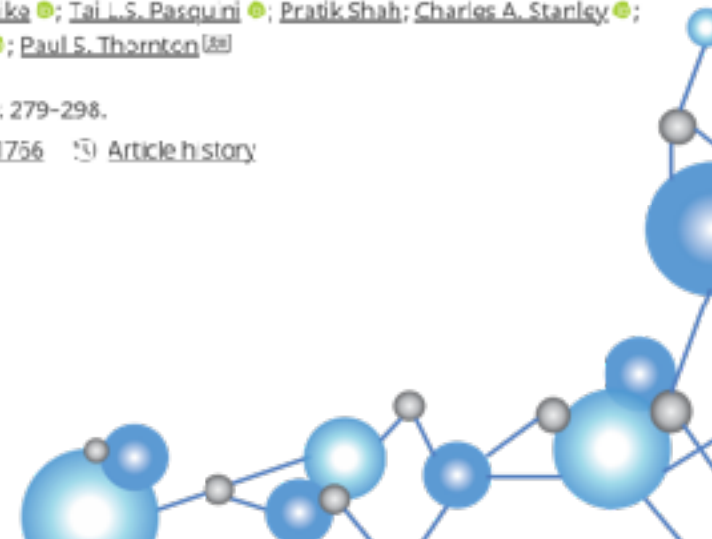
Subject Area:  Endocrinology,  Women's and Children's Health

[Diva D. De Leon](#) ; [Jean Baptiste Arneux](#); [Indraneel Banerjee](#) ; [Ignacio Bergada](#) ; [Tricia Bhatti](#); [Louise S. Corwell](#) ; [Junfen Fu](#); [Sarah E. Flanagan](#) ; [David Gillis](#); [Thomas Meissner](#); [Klaus Mohnike](#) ; [Tai L.S. Pasquini](#) ; [Pratik Shah](#); [Charles A. Stanley](#) ; [Adrian Vella](#) ; [Tehru Yorifuji](#) ; [Paul S. Thornton](#) 

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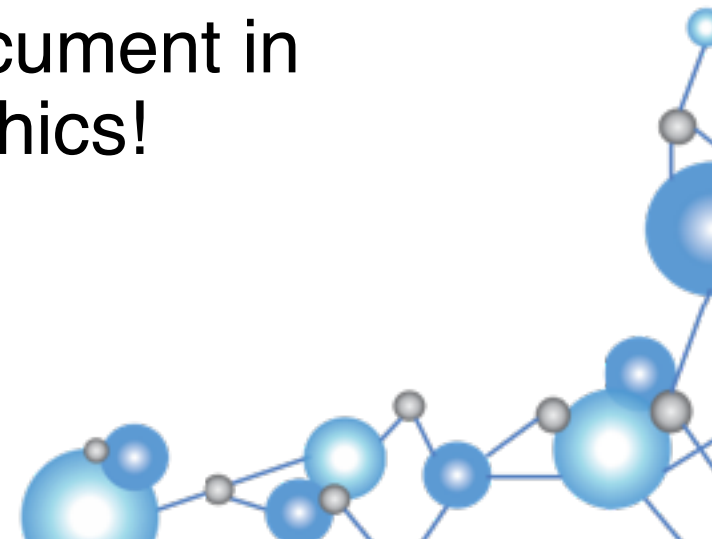
2025 – The Simplified Guidelines are Published on CHI's Website

- The CRN created a complementary version of the international clinician guidelines that is tailored for a broader audience.
 - This project was led by Janet Lee, a nurse and HI parent.
 - Authors of the clinician guidelines document provided feedback to ensure the simplified version remains accurate and true to the original meaning.
- Now, there's a direct translation of the formal document in simplified language, additional context, and graphics!

Complex clinical guidelines



Directly mapped to lay summary



2024-2025 — Creation of advocacy and awareness documents

- The CRN collaborated on two documents raising awareness of HI and advocating for the needs of people who live with the condition
 - The **Advocacy Statement** was prepared to improve the lives of people with HI by insisting on the use of best care practices.
 - The **Essential Medical Care, Medicine, Supplies and Services for People with HI** is a list of needs that should be required to be delivered by national and local health care and social systems.

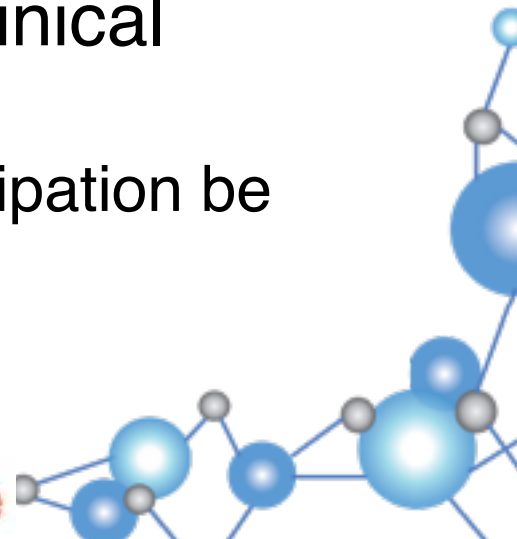
Both are available on our website in 5 languages!



Ongoing — Patient-powered research is at the forefront of therapeutic development



- HI experiences help the pharmaceutical industry and academic researchers understand what issues matter most.
 - Examples: unmet needs, undesirable side effects, complications of the condition, etc.
 - Several HI pharmaceutical groups consult CHI for understanding what is important to the community and the natural history of HI.
- People with HI have shaped study protocols for how clinical trials should be done.
 - Examples: what are the important outcomes, how can participation be inclusive and practical, etc.



How can YOU support patient-powered research endeavors?

- Person with HI or an HI family member? Join HIGR! Share your experiences, your questions, your challenges, and your wins.
- Researcher? Connect with CHI to support your research projects. Continue pursuing studies and grants in the field of HI.
- Biotech leaders? Use HIGR to inform you of the needs in the community and guide development of a therapy to advance the standard of care.



Your family's experience with hyperinsulinism could help find a cure.

Sign up for the HI Global Registry today at higlobalregistry.org

higlobalregistry.org



Dare to dream of the next milestones in HI history. Be a part of shaping the future of HI.