

Your Voice Has Power

How Research Helps to Tell the HI Story

Kristen Rohli, PhD

Associate Director of Research

Congenital Hyperinsulinism International

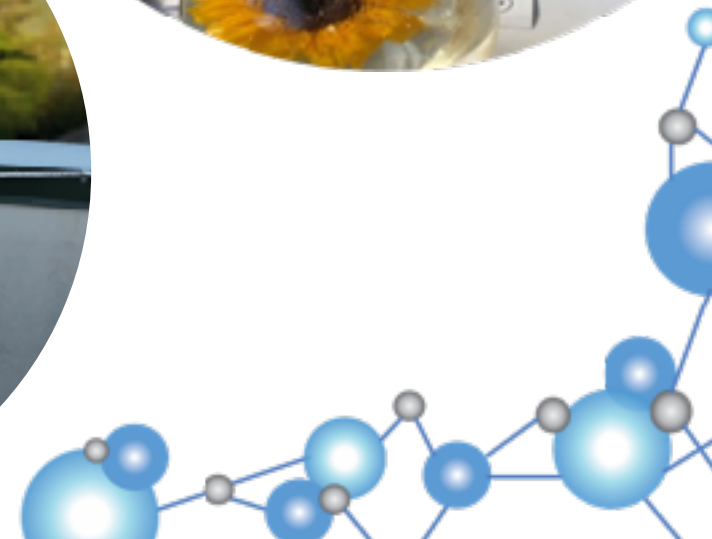
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What Does “Quality of Life” Mean?

Quality of Life = QoL

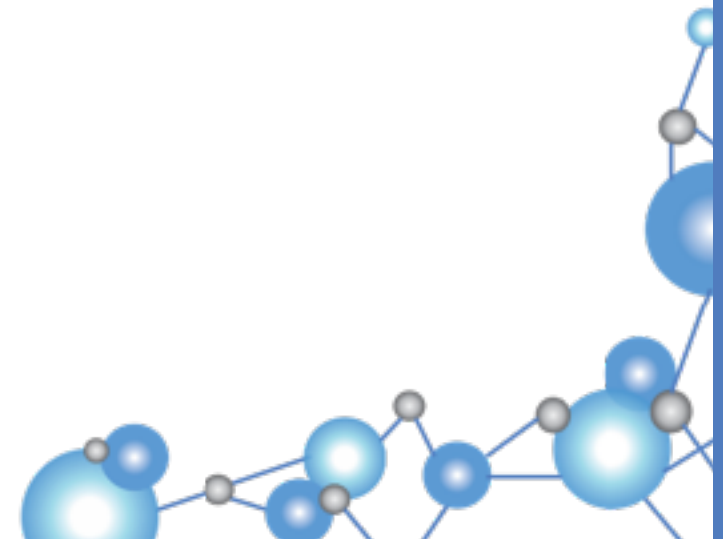
World Health Organization: An individual's **perception** of their position in life in the context of the culture and value systems in which they live and in relation to their **goals, expectations, standards, and concerns**



Why Do We Care About QoL?

- Quality of Life (QoL) differs for every family
 - Sleeping through the night
 - Treatments that do not cause pain
 - Transitioning to school – academically, socially, developmentally
 - Considering impact of puberty, child-bearing
 - Financial cost of healthcare

What does a **good quality of life** look like to you?
To your family?



How Do People with HI & their Parents/Caregivers Feel?

What matters to you?

What causes your quality of life to **decrease**?

What makes your quality of life **improve**?



What is QOL data used for?

Developing new treatments

*Change in QOL tells us if treatment is **effective**, beyond biomarkers and glycemic control*

Coverage & Access to new treatments

Improvements in QOL compared to existing treatments can support getting new drugs covered by insurance

Support & Advocacy Documents

By letting us know what the most pressing issues are to your family, we can develop resources to support your family

KONJENİTAL HİPERİNSÜLİNİZM NEDİR?

Konjenital hiperinsülinizm (KH) yenidoğan ve çocukluk dönemindeki yideli hastalığı hipoglisemiyinin en sık karşılaşılan nedenlerinden biridir. Uzun süreli hipoglisemi nörolojik beyin hasarına sebep olmaktadır.

Hiperinsülinizm birçok farklı sebebi bulunmaktadır. Bazı türleri geçirdiği diğer bazı türleri ise genetik bozukluklardan kaynaklanmakta ve hayat boyu sürdürebilmektedir. Beyin hasarı riski her iki durumda da yüksek olduğudur.

Hipoglisemi, HHI bebeklerin yaklaşık %60'ında, hayatlarının ilk ayında ortaya çıkar. Diğerlerin neredeyse tamamı 1 yaşında önce hipoglisemi yaşar.

HHI bebekler sağlıklı çocuklardan 5 kat daha fazla sekece (glukoz) ihtiyaç duyar.

Hiperinsülinizmde insülin salgılayan endokrin olan pankreas, sende glukoz düzeyini düşürmek etmekte ve glukoz miktarı düşük olsa bile insülin salgılamaktadır. Bunun sonucunda yideli ve uzun süreli hipoglisemi ortaya çıkar.

Çoğu ülkede HHI'nin görülme sıklığı 25.000-50.000 doğumda 1'dir.

**#bmysugar
#stopthelows**

Erken teşhis ve tedavi sonucunda hipoglisemiyi önlenmesiyle, beyin hasarı ve diğer engelleme riski azalmaktadır.

CHI Congenital Hyperinsulinism International
www.congenitalhi.org

f Congenital Hyperinsulinism
t @congenitalhi
m @chi_hypoglycemia

HI Global Registry (HIGR)

2 surveys
focused on
Quality of Life
(QoL)

Participant
Quality of Life
People with HI
age 14+ years

Caregiver
Quality of Life
Parents/Caregivers
of people with HI

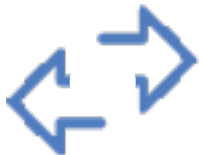
Aspects of Quality of Life also captured in many other surveys

The HI Global Registry surveys allow us to assess:

Where needs are being met



Where gaps exist



What changes may or may not make an impact on the lives of those living with HI



HI Global Registry (HIGR)



HIGR QOL Surveys cover a range of topics, including:

- Health
- Social support
- School / work
- Medical care
- And more!

Most questions rates on a **subjective scale** – no right or wrong answers!

Every question has a **“Prefer not to answer”** option. You do not have to share anything you are not comfortable sharing.

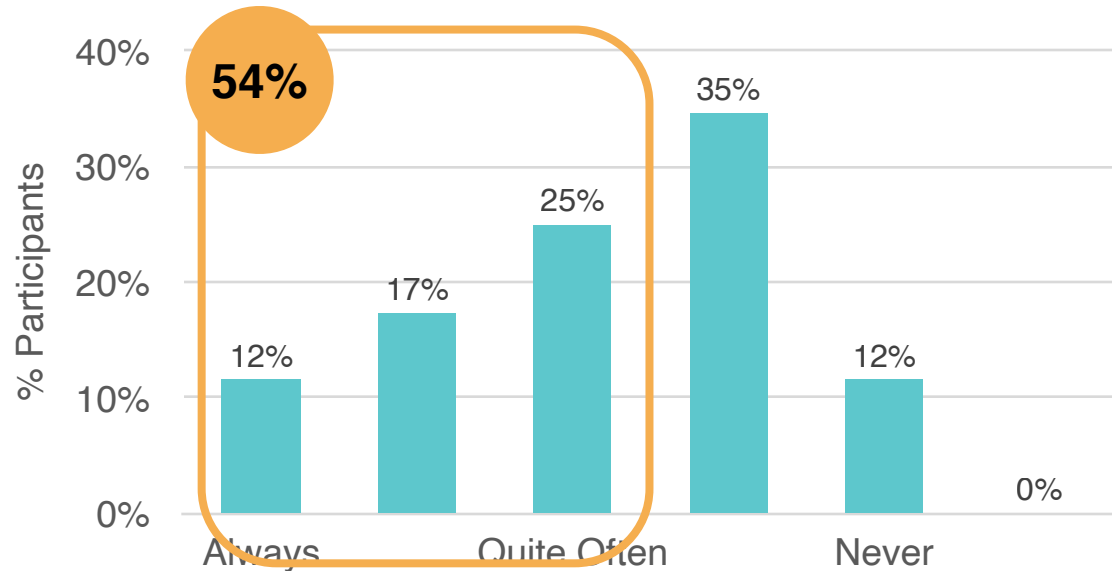
In general, how would you rate the support you receive from healthcare professionals? *

- Excellent
- Very Good
- Good
- Fair
- Poor
- Prefer not to answer

Participant Perspectives



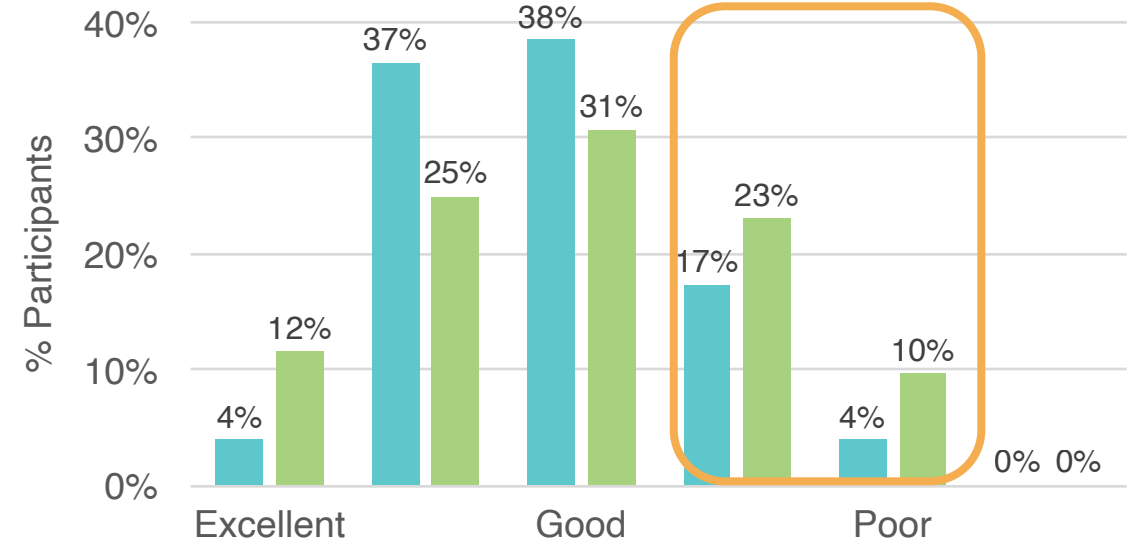
Do you feel that your life is ruled by your HI? (n = 52)



Over half (54%) of participants with HI feel that their life is ruled by their HI

How would you rate your:

- Physical health? (n = 52)
- Mental health? (n = 52)



- **1 in 4** participants rated their **physical** health as fair or poor
- **1 in 3** participants rated their **mental** health as fair or poor

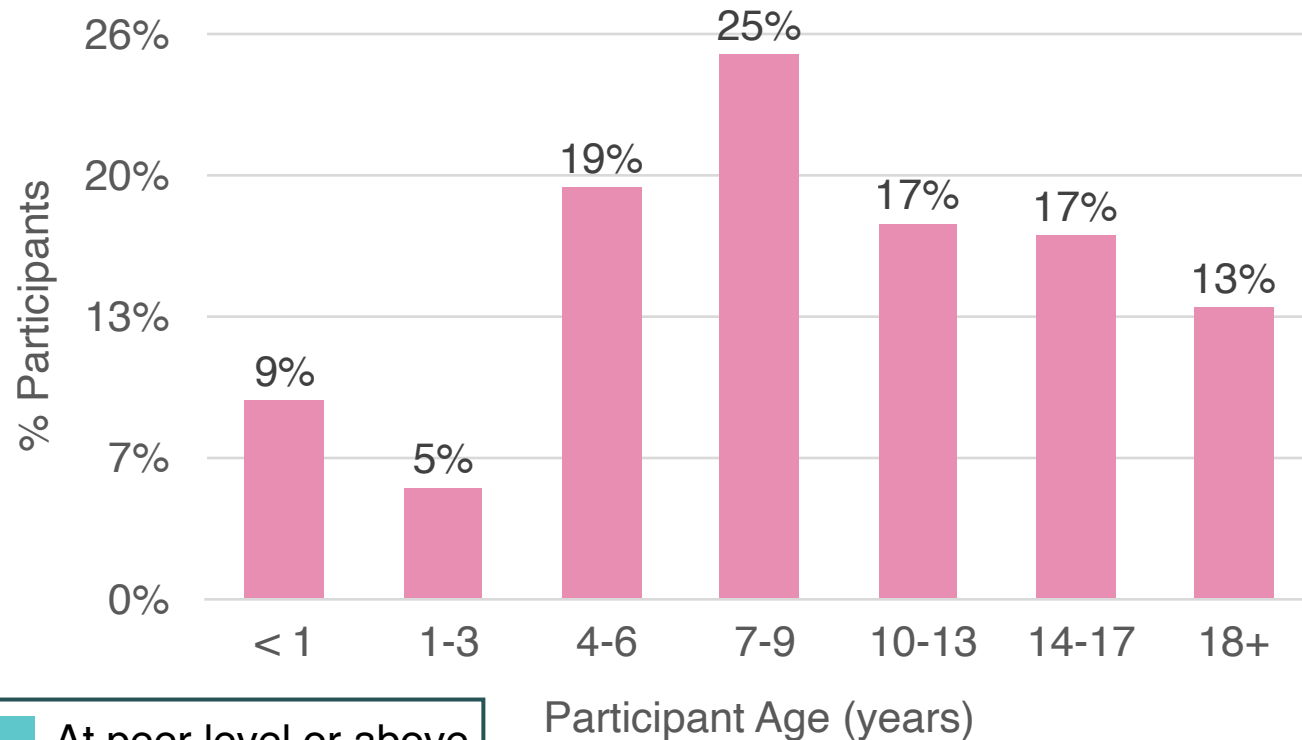
“Participant” = person with HI

Participant QOL is assigned to all participants > 14 years old

Relationship Behaviors



Is the participant capable of performing most **relationship behaviors** as peers of the same age? (n = 262)



- At peer level or above
- Variable
- Below peer level
- Unsure / Unknown

“Participant” = person with HI

Difficulties performing relationship behaviors are evident in some younger children and become **more common in older children and teens**

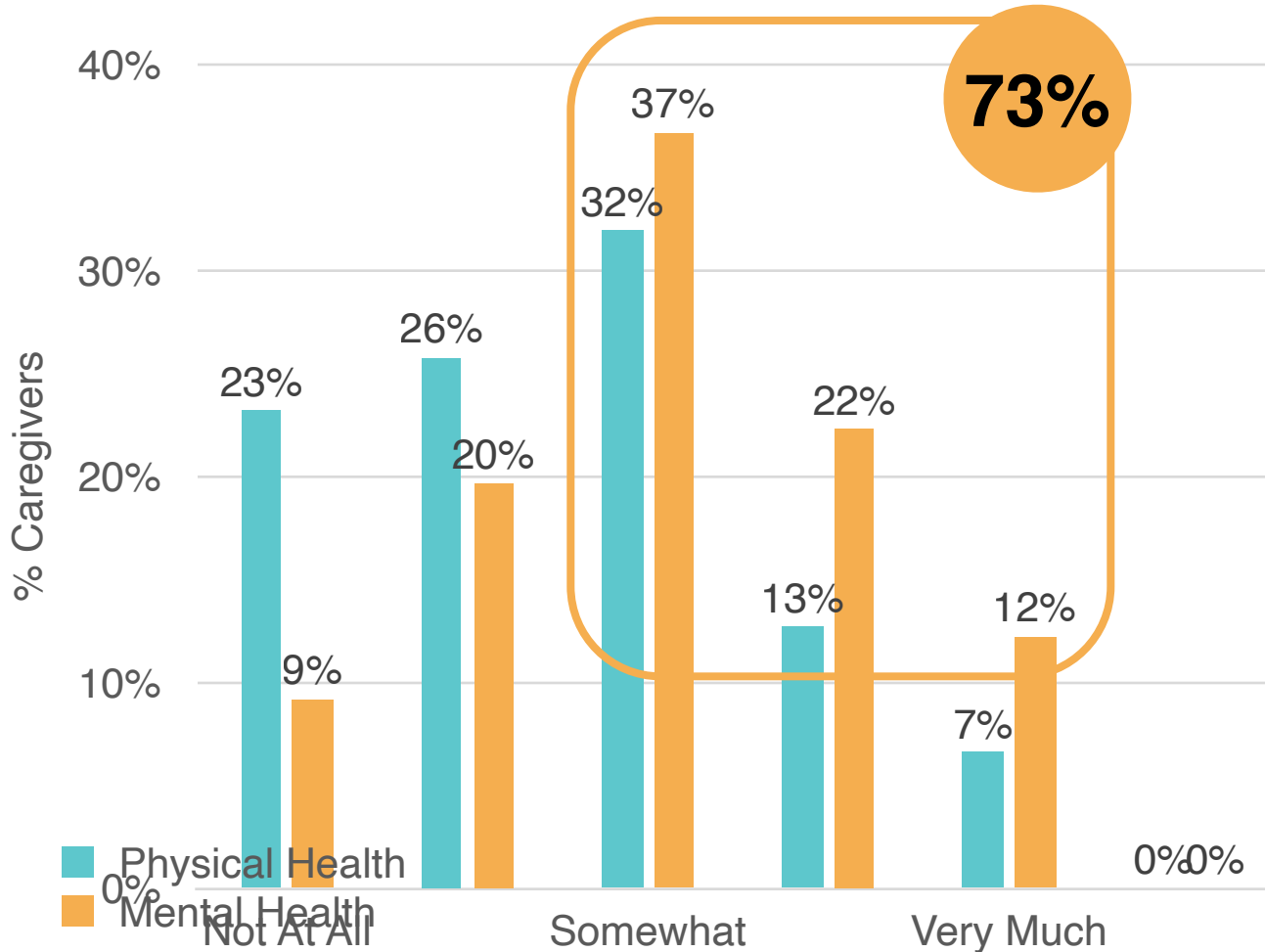


Parent Mental Health

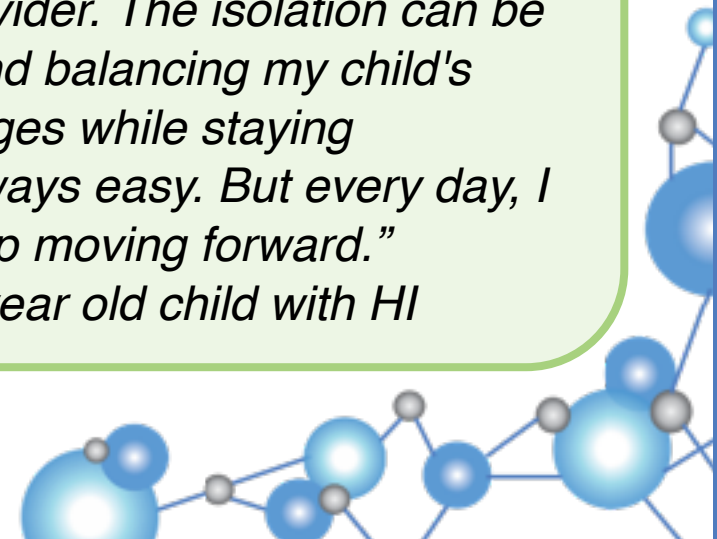


Do you (as the caregiver of the participant with HI) feel your **physical / mental health** has suffered from caring for the participant with HI? (n = 229)

Many parents report that their mental health has suffered



“This journey has also taken a personal toll, straining my marriage to the point of divorce and leaving me as the sole caregiver and provider. The isolation can be heavy at times, and balancing my child's emotional challenges while staying motivated isn't always easy. But every day, I do my best to keep moving forward.”
- Parent of an 8-year old child with HI

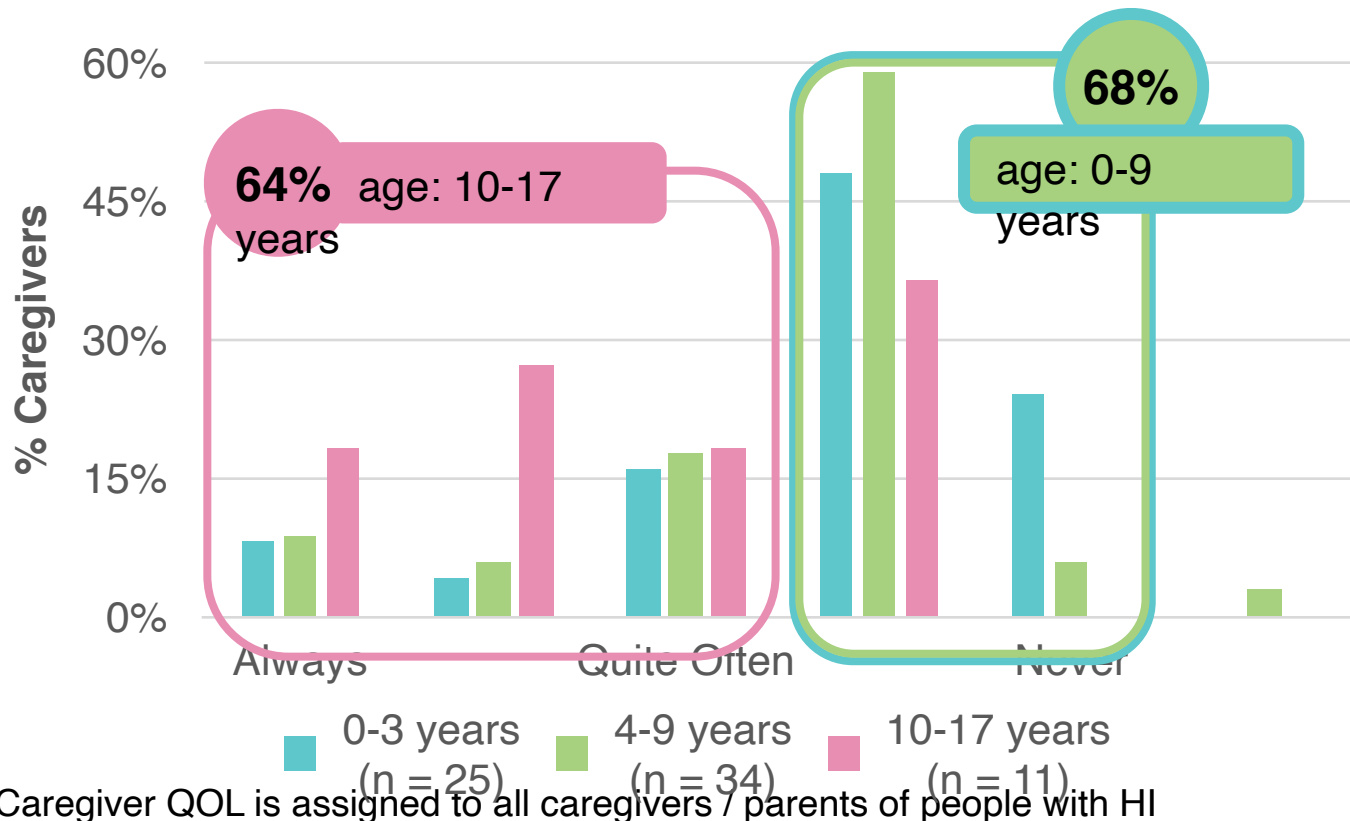


Ability to Trust Others with Responsibility of Care



Do you feel comfortable allowing others to care for the participant without you being present (i.e. babysit)? (n = 70)

Most parents of young children do not feel comfortable leaving participant with others, but parents of older children are more comfortable with this



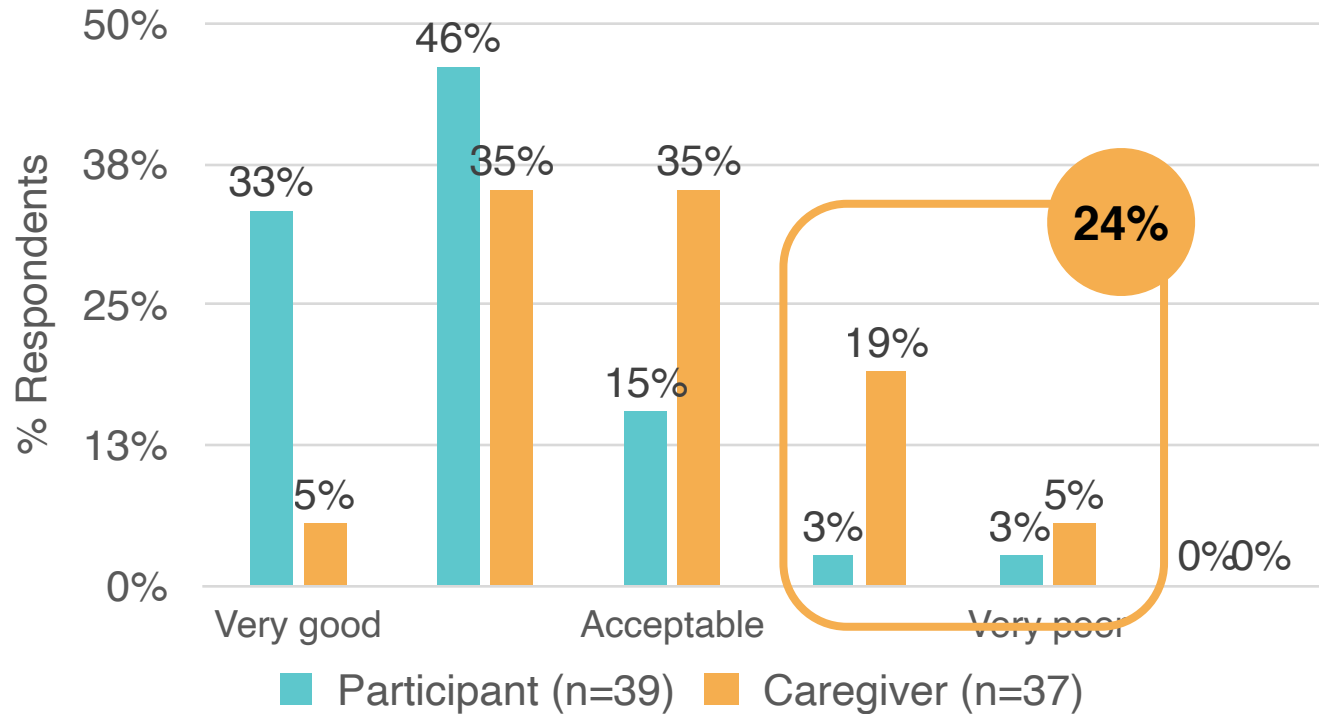
"I take him with me most places, some people are afraid to babysit him and would even say that they prefer to babysit my 2-year-old daughter rather than him".

- Parent of a 4-year-old child with HI

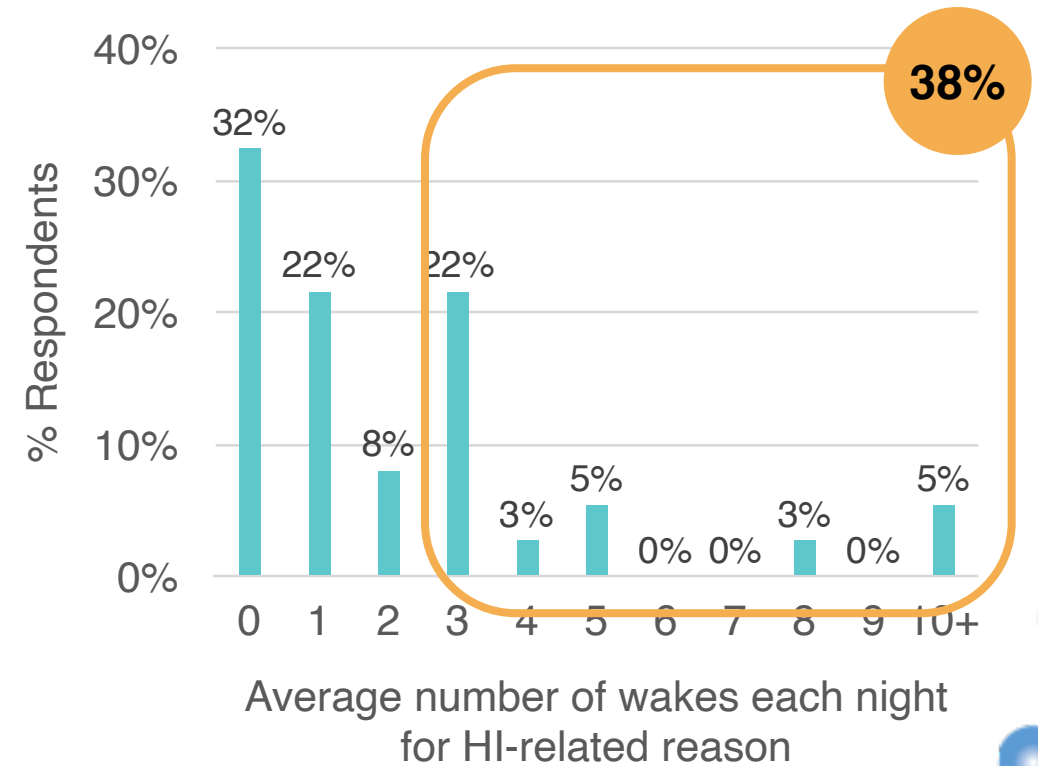
Sleep Quality



On average over the past 6 months, how would you rate the quality of the **participant's / caregiver's** sleep?



On average over the past 6 months, how many times do you wake up in the night for HI-related reasons? (n = 37)



Current Research Projects

European Society for Pediatric Endocrinology (ESPE)

Feeding Issues: An Under-Recognized Complication in People with Hyperinsulinism

Lauren N. Lopez, Tai L. S. Pasquini, Julie Faskin
Congenital Hyperinsulinism International, Glen Ridge, New Jersey, USA

INTRODUCTION & METHODS

- Congenital Hyperinsulinism (HI) is the most common cause of persistent hypoglycemia in newborns and children.
- In HI, dysregulated pancreatic beta-cells produce too much insulin, leading to severe hypoglycemia and, in many cases, irreversible neurological damage.
- An under-studied and under-recognized feature of HI is feeding issues.
- This study's aim was to characterize the pressures affecting issues in individuals with HI and the impact on their families.

RESULTS

Of 148 people with HI, 85% reported experiencing feeding issues.

85% reported feeding issues had not fully resolved, and 41% of these individuals were 7 years or older.

Feeding issues are a very long and stressful painful process. Around 7 years later, could we still: I struggle my social life and well-being is a major issue. - Adult with HI

DISCUSSION

Feeding issues are poorly understood within the context of HI. It is unknown exactly what leads to feeding issues, although factors such as direct neurophysiology of HI, the effects of medications, and the psychological impacts of the frequent interventions of feeding and over-feeding prevent hypoglycemia may play a role. Many individuals with HI and their caregivers feel feeding issues represent a major obstacle in the clinical management of HI. More research into the causes of feeding issues and the most effective strategies to manage them are urgently needed.

Pediatric Endocrine Society (PES)

The Impact of Congenital Hyperinsulinism on Family Finances and Time: A Report from the HI Global Registry

Lauren N. Lopez, Kristen E. Rohli, Julie Faskin, Tai L. S. Pasquini
Congenital Hyperinsulinism International, Glen Ridge, New Jersey, USA

INTRODUCTION

- Congenital hyperinsulinism (HI) is a rare, life-threatening, neuroendocrine condition that results in severe hypoglycemia and, in many cases, irreversible neurological damage.
- It is the dysfunction of pancreatic beta-cells that leads to excessive insulin production and severe hypoglycemia, which often causes neurodevelopmental damage.
- The management of HI is a complex, long-term process, often involving multiple medications, frequent blood sugar monitoring, and frequent hospitalizations.
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RESULTS

More than 80% of participants reported that HI significantly impacted their family's finances and time.

Participants reported spending an average of \$1,000 per month on HI management, with some reporting over \$2,000 per month.

Participants reported spending an average of 10 hours per week on HI management, with some reporting over 20 hours per week.

DISCUSSION

HI is a complex, long-term condition that significantly impacts family finances and time. The management of HI is a complex, long-term process, often involving multiple medications, frequent blood sugar monitoring, and frequent hospitalizations.

NEW RESEARCH PUBLICATION

Real-World Experience with the Use of Diazoxide Among People Living with Congenital Hyperinsulinism and their Caregivers

Tai L. S. Pasquini^{1*}, Kristen E. Rohli¹, Fiona J. Almeida², Indraneel Banerjee³, Antonia Dastamani⁴, Diva D. De Leon⁵, Lauren N. Lopez¹, Paul Thornton⁶, Julie Raskin¹

Research Publications Coming Soon!

The Impact of Subtotal Pancreatectomy on People with Congenital Hyperinsulinism and Their Caregivers

Real-World Insights into Neurodevelopmental Outcomes Amongst People with Congenital Hyperinsulinism

Change over Time

- Things affecting Quality of Life change over time
 - Change in HI management
 - Change in life stage
 - Change in family priorities



Gabriel, 6 months

How has your Quality of Life
changed over the **past year**?
The **past 5 years**?

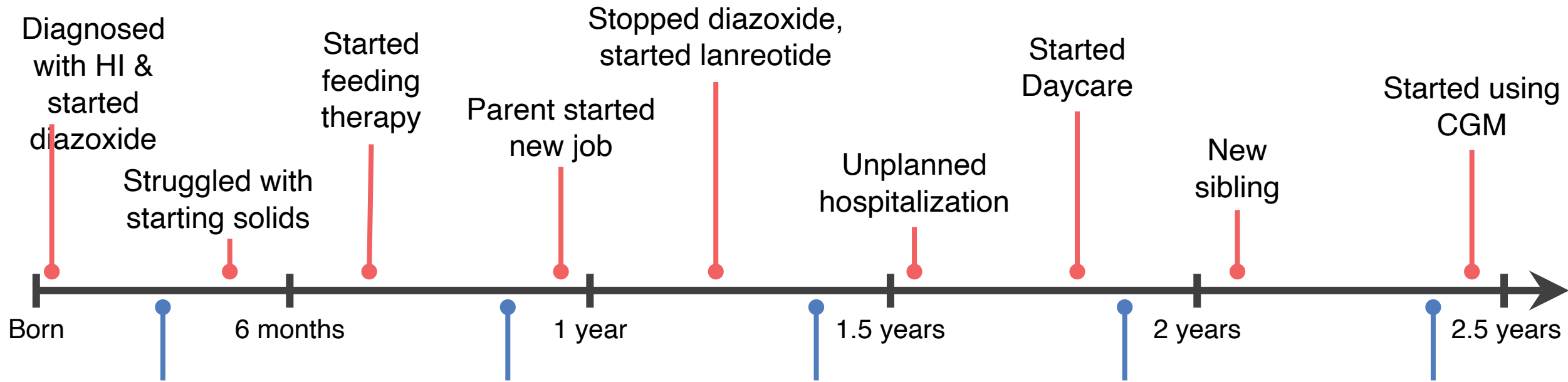
The **HI Global Registry (HIGR) Quality of Life surveys** are assigned every 6 months

- Allows us to track changes in QoL over time
- Allows us to match changes in key HI management events to changes in QoL



Gabriel, 6 years

Change over Time

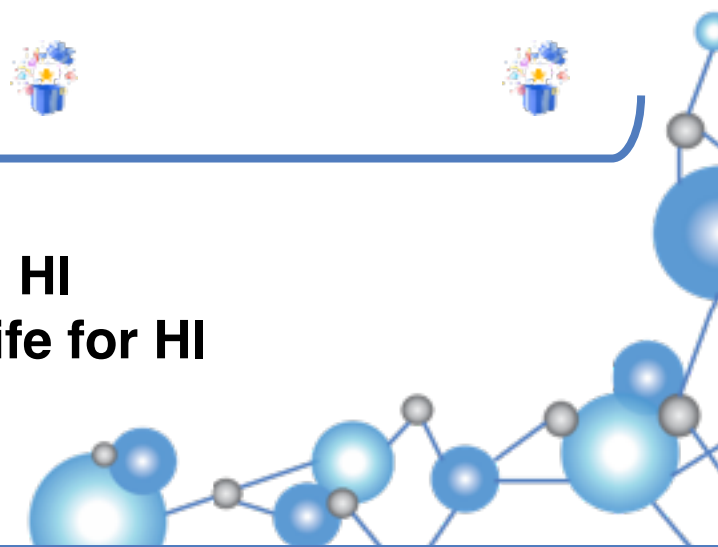


Rich insight into how changes in HI management affect overall quality of life for HI families

- ✓ Time to Say HI!
- ✓ Quality of Life



Participant Appreciation Program
Survey completion gift *(if all surveys completed)*



Final Message

- Quality of Life is different for every individual and every family
- Information about Quality of Life is critical to inform drug development and advocacy initiatives
- HIGR provides a way for you to share what is most impactful to **you and your family**
- Quality of Life changes over time → important to keep completing surveys over time

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



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

higlobalregistry.org



Share your story
Join the HI Global Registry!
www.higlobalregistry.org