CONGENITAL HYPERINSULINISM BEYOND CHILDHOOD: CLINICAL CHARACTERISTICS AND UNMET NEEDS IN ADULTS

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INTRODUCTION

- **Congenital hyperinsulinism (HI) is a rare group of conditions where increased insulin secretion causes severe, recurrent hypoglycemia.**
- In HI, pancreatic beta-cell dysfunction leads to excessive insulin production and severe hypoglycemia.
- While hypoglycemia resolves over time for some individuals, HI persists into adulthood for many, presenting long-term management challenges.
- In recent studies, adults with HI emphasized significant difficulties transitioning from pediatric to adult care, often encountering clinicians unfamiliar with the condition.
- Objective: We characterized the adult population enrolled in the HI Global Registry (HIGR) to demonstrate ongoing clinical burden and the need for improved preparedness among adult endocrinologists.

METHODS

The HI Global Registry (HIGR) is the only international patient-powered registry for people with HI.

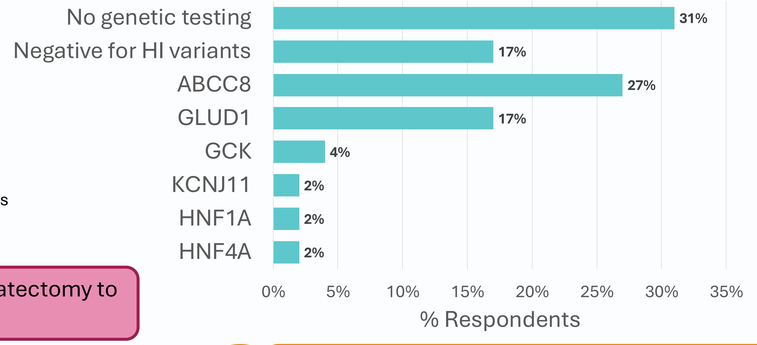
- Adults (≥18 yrs old) who completed at least one of the following surveys in HIGR were included in the study: *Diagnosis, Medication Management, Surgical Management, Glucose Monitoring, Diet and Feeding,* or *Other Medical Conditions*.
- Qualitative interviews from previous studies were analyzed for themes relating to clinical management in adulthood and transitioning care from pediatric to adult practitioners.

RESULTS

85
Participants were included in this study* (ages 18-80 years)

*Not all participants completed all surveys

Genetic Testing Results in Adults with HI (n=52)



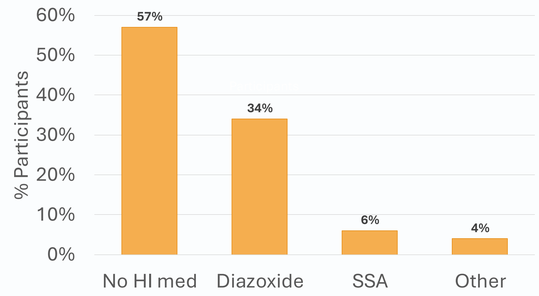
25% Reported having a pancreatectomy to treat HI (n=69)

Of these who had a pancreatectomy, **11 reported having a diabetes diagnosis** at the time of survey.

"I don't know how intense management would have been without that surgery, because I feel we could have put her at greater risk of other complications, more seizures or more permanent brain damage, more developmental delays. Anything like that would have been worse."-*Caregiver to child who had a subtotal pancreatectomy for diffuse HI*

63% Reported having used a CGM to monitor blood glucose levels (n=65)

43% Of participants who did not report diabetes 43% were taking at least one HI medication at the time of survey (n=70)



Other includes sirolimus (n=1), steroids (n=1), and routine glucagon (n=1).

	Participants who do not report having diabetes		Participants who report having diabetes	
Self-reported frequency of glucose event	**Hypoglycemia**	**Hyperglycemia**	**Hypoglycemia**	**Hyperglycemia**
More than once per day	9 (21%)	2 (5%)	1 (7%)	7 (47%)
Once a day or several times per week	14 (33%)	4 (10%)	5 (33%)	2 (13%)
Once per week or several times per month	8 (19%)	2 (5%)	4 (27%)	2 (13%)
Once per month or less frequently	9 (21%)	13 (31%)	5 (33%)	2 (13%)
Unknown	2 (5%)	5 (12%)	0 (0%)	1 (7%)
Never	0 (0%)	16 (38%)	0 (0%)	1 (7%)
Total	**42**		**15**	

Hypoglycemia: Blood glucose level below 70 mg/dL (3.9 mmol/L, 0.7 g/L)
Hyperglycemia: Blood glucose level above 180 mg/dL (10 mmol/L, 1.8 g/L)

The genetic landscape of HI continues to expand as new genes and mechanisms are identified. Therefore, adults with previously unknown genetics may consider re-testing, if appropriate.

43% Reported a history of feeding issues (n=65), yet of those, only 50% reported resolution.

- Feeding challenges may be the result of treatment side effects, diet management, or as a feature of the condition.
- Some participants reported **feeding therapy** to be helpful.

"Even the action of chewing, swallowing, I had to learn all of that. It wasn't instinctive. At first, it created a lot of anxiety to swallow and not get food stuck because it was hard… After I got the help, it got better at 9, 10 years old." –*Adult with HI*

During qualitative interviews, participants frequently expressed:

- **Challenges in transitioning care** from pediatric to adult endocrinologists
- Difficulties with **finding adult practitioners** having HI knowledge and/or experience
- **Decreased satisfaction in their clinical management** as an adult, compared to as a child.

CONCLUSIONS

- HI is often misunderstood as a childhood condition. However, many individuals experience lifelong challenges.
- Adults with HI represent a clinically complex and underserved population within adult endocrinology.
- There is an urgent need for improved education and awareness about HI to support the care of all individuals with the condition.

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Learn more about HIGR!

If you have any questions, please email krohli@congenitalhi.org