# Natural History Studies – Hyperinsulinism Hyperammonemia Syndrome

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# Natural History of the Hyperinsulinism Hyperammonemia Syndrome – A Multi-center Observational Study Incorporating Patient-centered Data through the HI Global Registry

**Aim:** A comprehensive summary of the clinical course, natural history, and quality of life outcomes in HI/HA syndrome

**Approach:** Pair medical record review with patient-centered data from HIGR

Children's Hospital of Philadelphia℠
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CONGENITAL HYPERINSULINISM
HI GLOBAL REGISTRY

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# Approach Rationale

- **Electronic medical record is a rich source of clinical information but….**

  - Only patients followed by a given center are included

  - Incomplete information (>> information captured in medical record)

  - Variable follow-up at a given center

  - Information entered is clinician (and clinical problem) specific

# Approach Rationale

1) **Address potential selection bias** – especially among those with milder or improved HI

- Differential referral patterns (CHI CoE)
- Loss to follow-up

2) **Gain insight into patient-perspective data that is typically absent from medical records**

3) Sample size – strength in numbers!

# Preliminary findings

| Age at presentation/diagnosis | CHOP/Cook EHR (n=61) | HIGR (n=14) |
|---|---|---|
| **Presentation**, median (range) | 5mo (0d- 2.5y) | — |
| 0-6 days, % (n) | 31% (19) | 50% (7) |
| 1-4 weeks, % (n) | 7% (4) | 14% (2) |
| 5 weeks - 6 months, % (n) | 23% (14) | 14% (2) |
| 7-11 months, % (n) | 18% (11) | 7% (1) |
| 1-3 years, % (n) | 21% (13) | 7% (1) |
| >3 years, % (n) | 0% (0) | 7% (1) |
| **HI diagnosis**, median (range) | 6mo (0d- 17.1y) | — |
| **HI/HA diagnosis**, median (range) | 14mo (0d-28.9y) | — |

# Preliminary findings
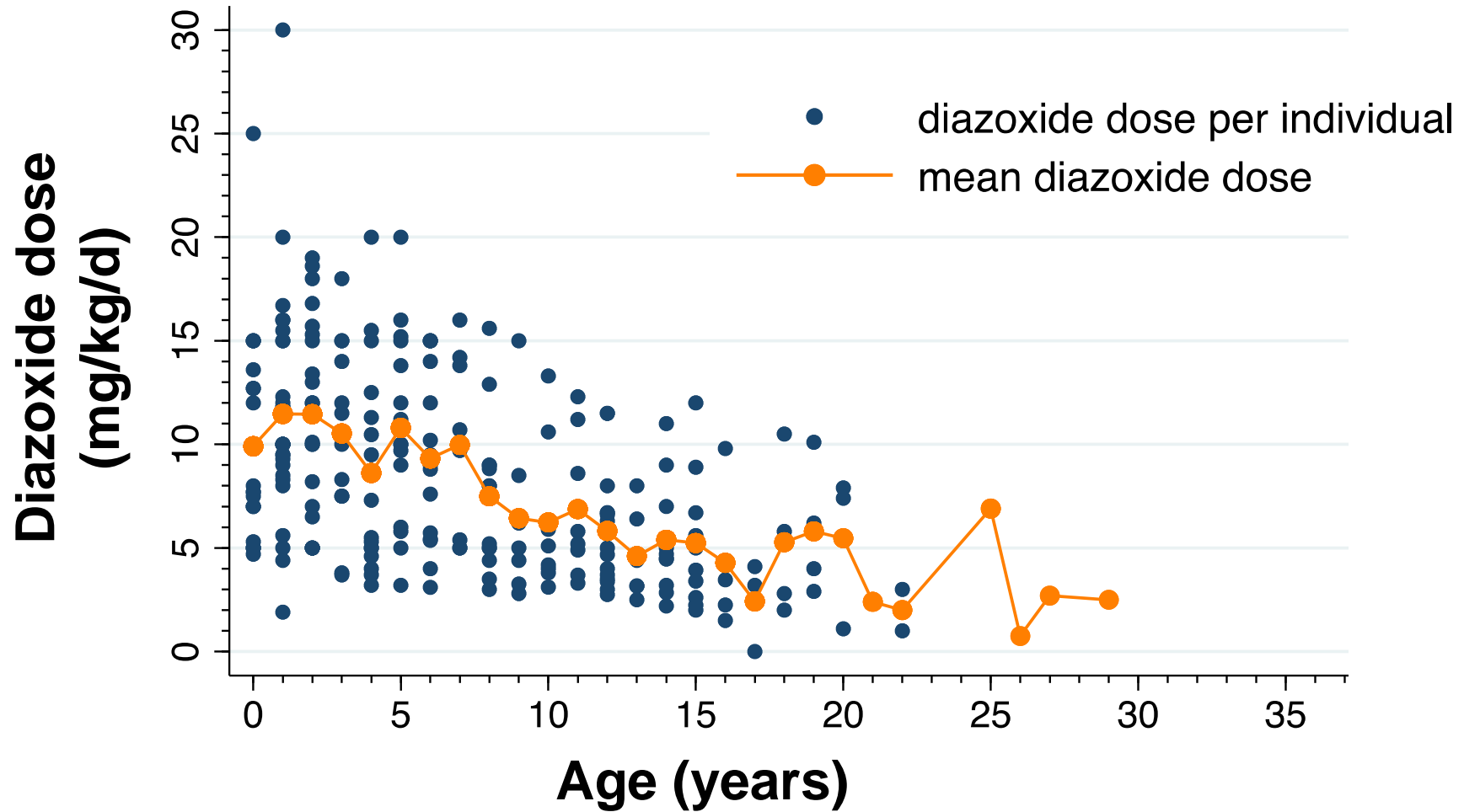


Symptoms at presentation

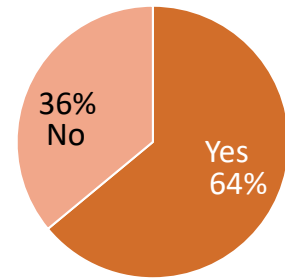# Preliminary findings



Initial HI Management
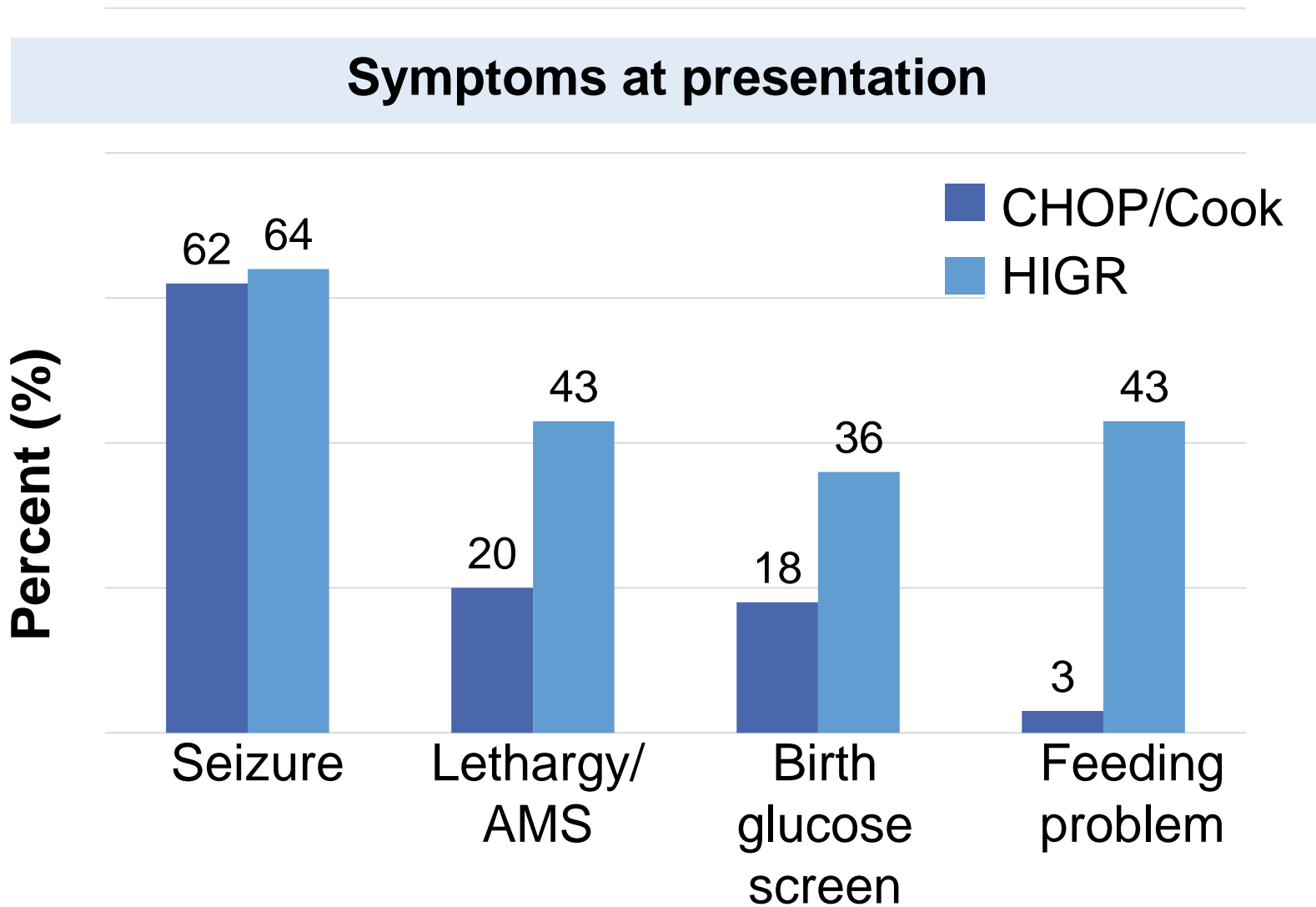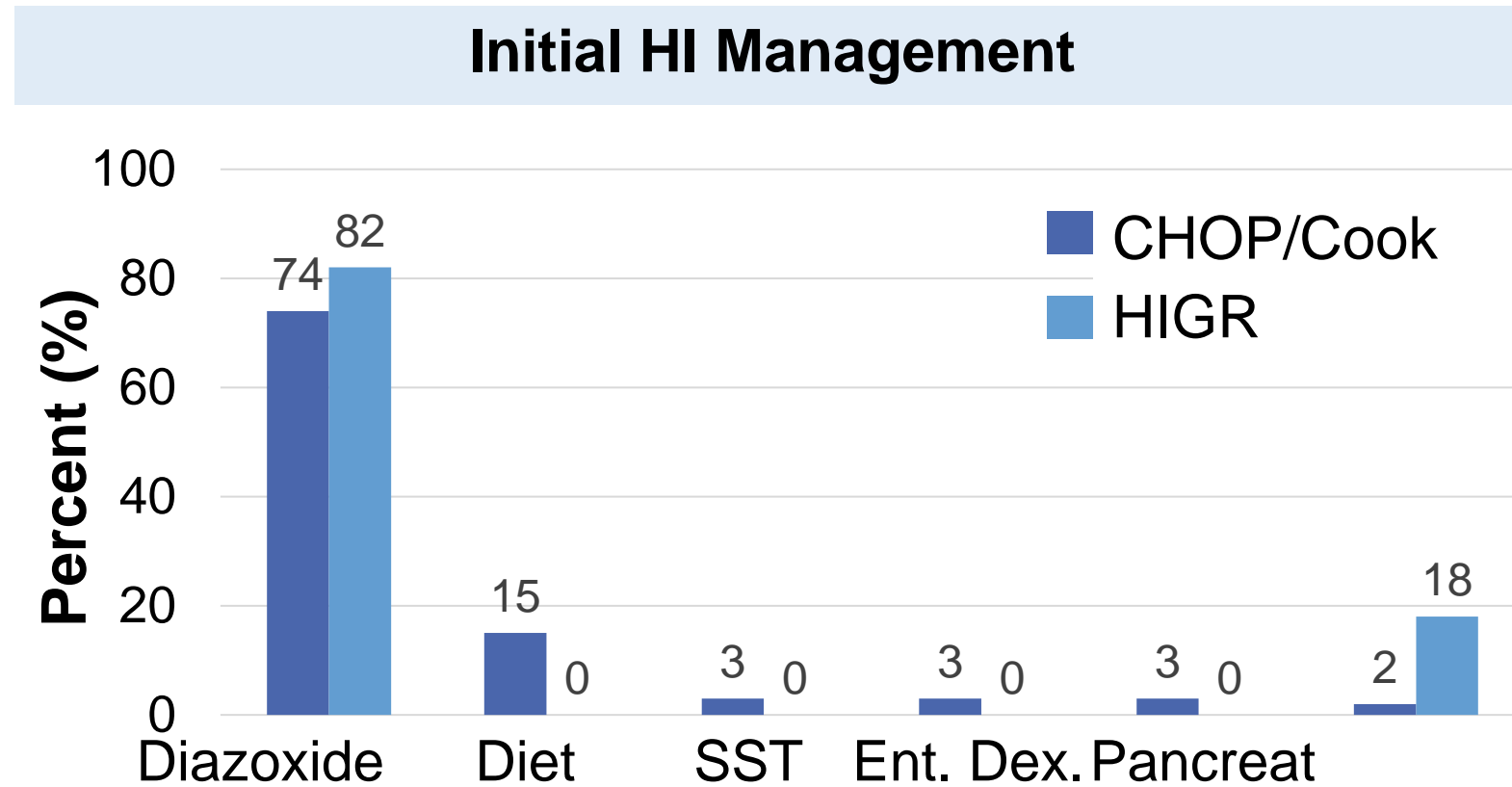
# Preliminary findings: EHR data only…

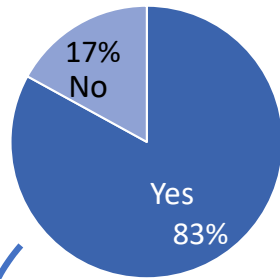# Preliminary findings: Seizure course

## History of any confirmed or suspected seizure

■ CHOP/Cook (n=66)　　■ HIGR (n=14)

**CHOP/Cook pie chart:**
- 17% No
- Yes 83%

**HIGR pie chart:**
- 36% No
- Yes 64%

Diagnosed with epilepsy _before_ HI
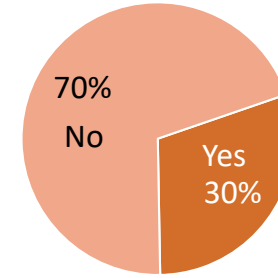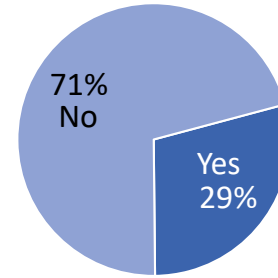
CHOP/Cook: 22%

HIGR: 44%

33% Fully resolved after HI treatment
13% Persisted despite HI treatment
38% Resolved w treatment, then recurred
16% Onset after HI treatment

Of these, 51% diagnosed with epilepsy

## Epilepsy history after HI treatment

■ CHOP/Cook (n=66)　　■ HIGR (n=10)

**CHOP/Cook pie chart:**
- 71% No
- Yes 29%

**HIGR pie chart:**
- 70% No
- Yes 30%

53% atypical absence type
58% remain on AED
Age last contact: 12.8 years
range: 2.2-31 years

Seizure type unrecorded
100% remain on AED
Age last contact: 14.0 years
range: 13.8-19.9 years

# QoL

- More to come!