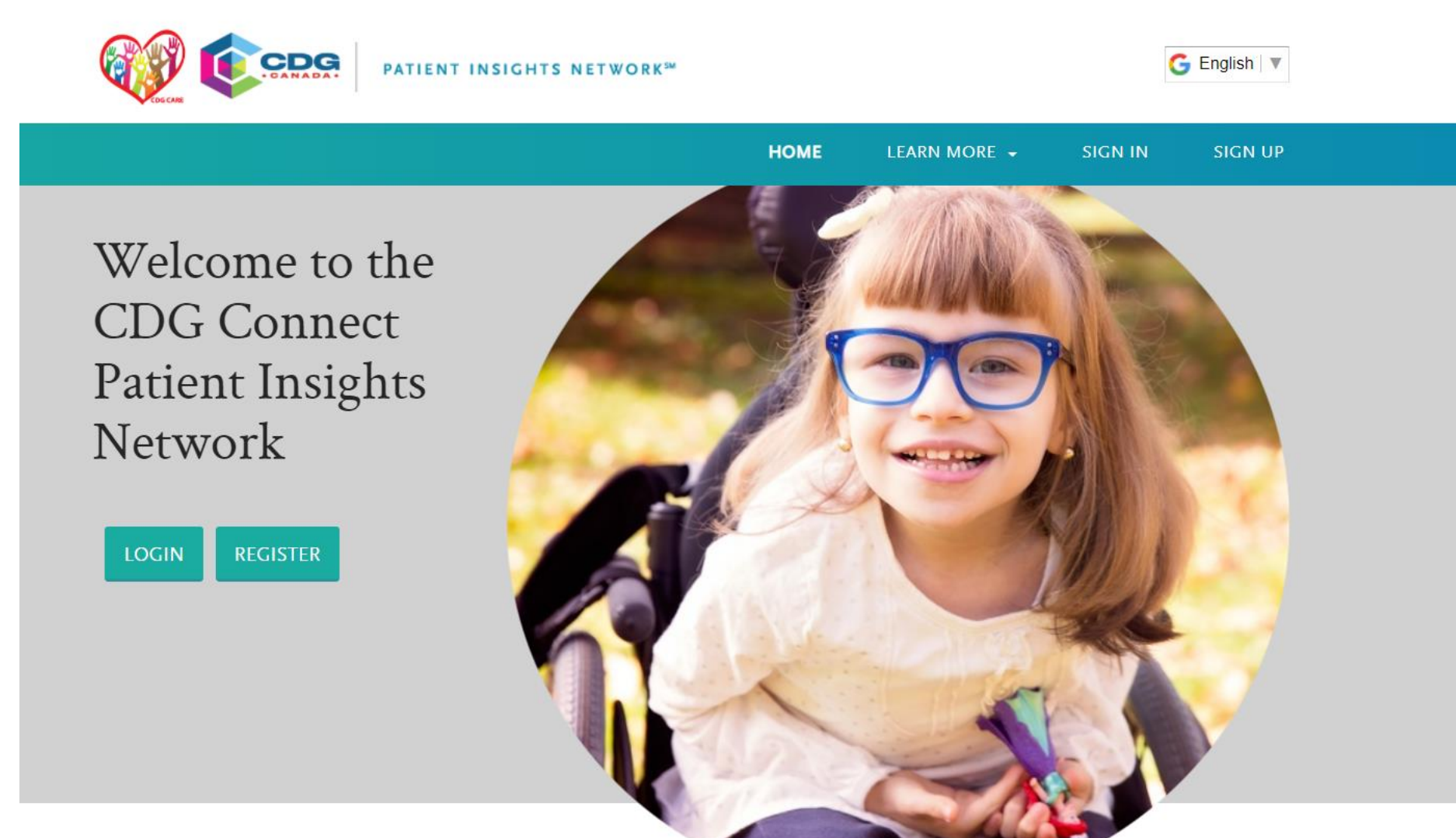


CDG Connect Patient Insights Network (PIN)

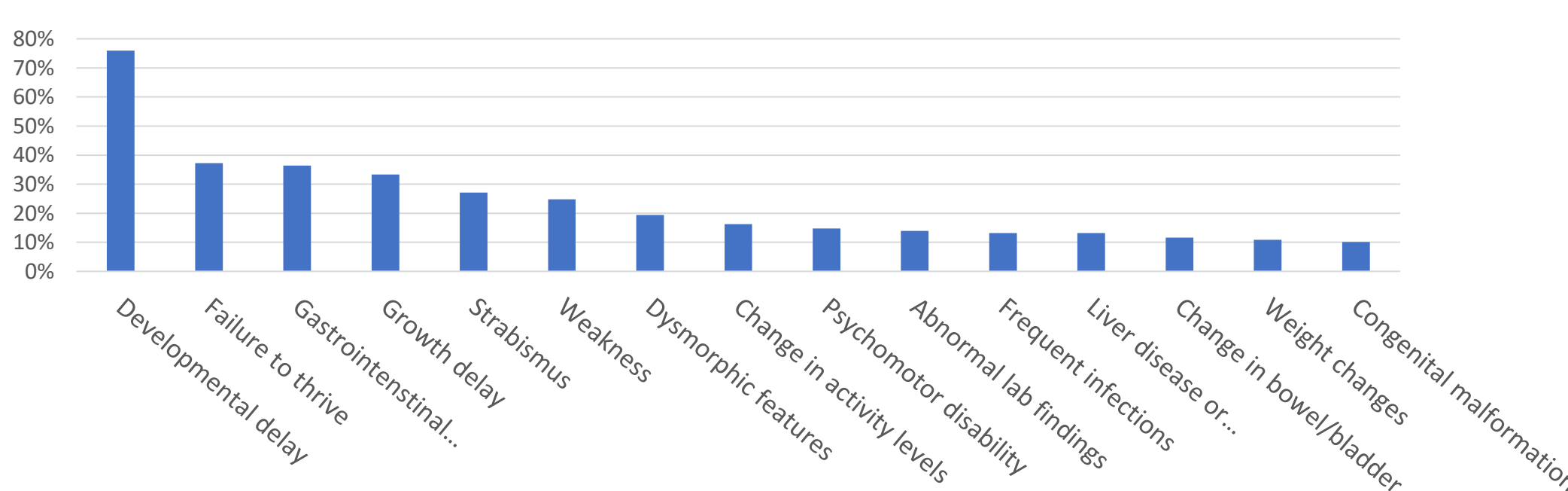
- Launched May of 2018 in partnership with Ichorion Therapeutics (now Cerecor, Inc.) through the Invitae Patient Registry Platform
- To serve as a baseline and custom survey data collection for CDG patients around the globe, to speed understanding around this ultra rare disease
- The CDG Connect PIN hosts one custom survey as well as 4 core surveys intended to capture high level data to make pan-disease study possible
- Genetic, biochemical, and developmental reports can be collected and curated
- 57% of participants have completed the CDG-specific embargoed IRB study survey



What are Congenital Disorders of Glycosylation (CDGs)?

- A large group of ultra-rare, inherited disorders that affect the complex process in the body called glycosylation
- There are currently over 150 types of CDG that have been identified. CDGs are severely under-diagnosed and misdiagnosed with only approximately 1,200 cases diagnosed with CDG globally, and only about 220 cases currently reported in the USA.
- Most children present with neurological issues and symptoms, developmental problems, growth delays, and problems with organs not working like they should.

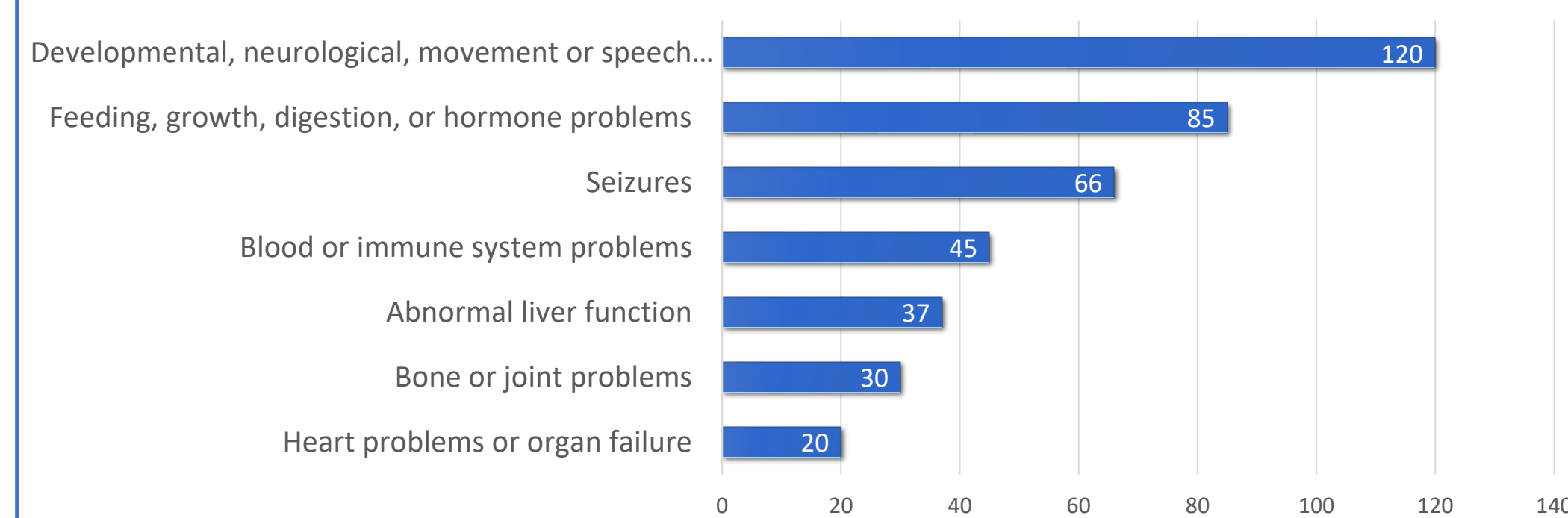
Which of the following symptoms led to the patient's initial medical evaluation?



What is Glycosylation?

- Glycans are sometimes called “sugar trees,” “antennas” or “sugar chains” by health care providers.
- When the sugar building blocks attach to proteins, they are called “glycoproteins.” When the sugar building blocks attach to lipids, they are called “glycolipids.”
- When someone has CDG, his or her body cannot properly add or attach the sugar building blocks to proteins or lipids. Every single system in the body needs the process of glycosylation to work right so the body can function normally.

Which of the following problems has the patient ever had due to CDG?



CDG Connect – Survey Population

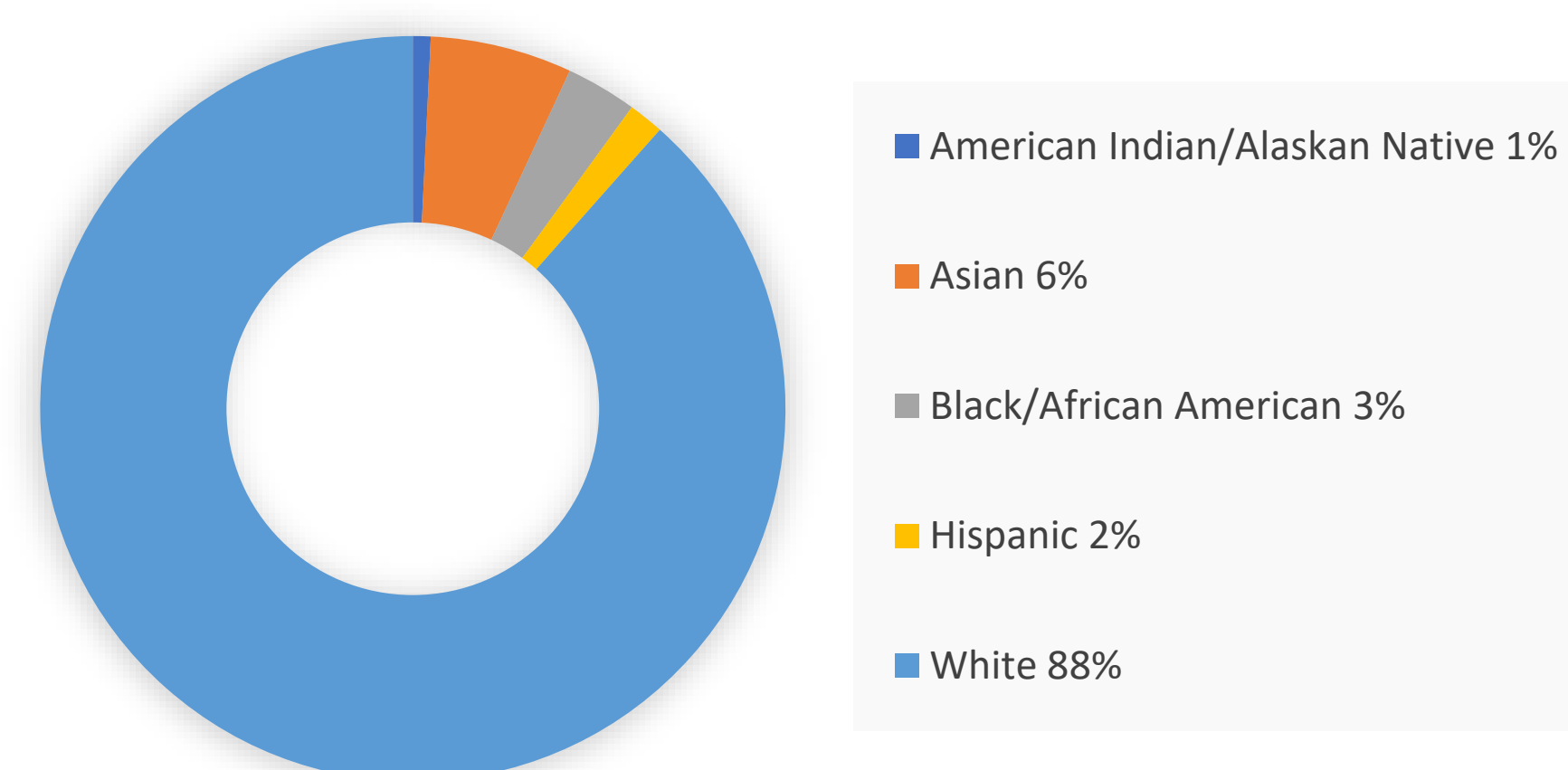
- Currently 225 registered participants
- Geographic representation from 24 countries
- 60% of survey patients reside in the United States
- 33 different CDG types reported
- For purposes of survey analysis, CDG types have been classified into the following groupings:

CDG Patients by Classification

GPI Anchor Disorders	19
N-glycosylation Type I CDG	33
N-glycosylation Type II CDG	20
PMM2-CDG	44
Other & Unknown CDG type*	13

*Other includes disorders of O-linked glycosylation & unknown CDG type

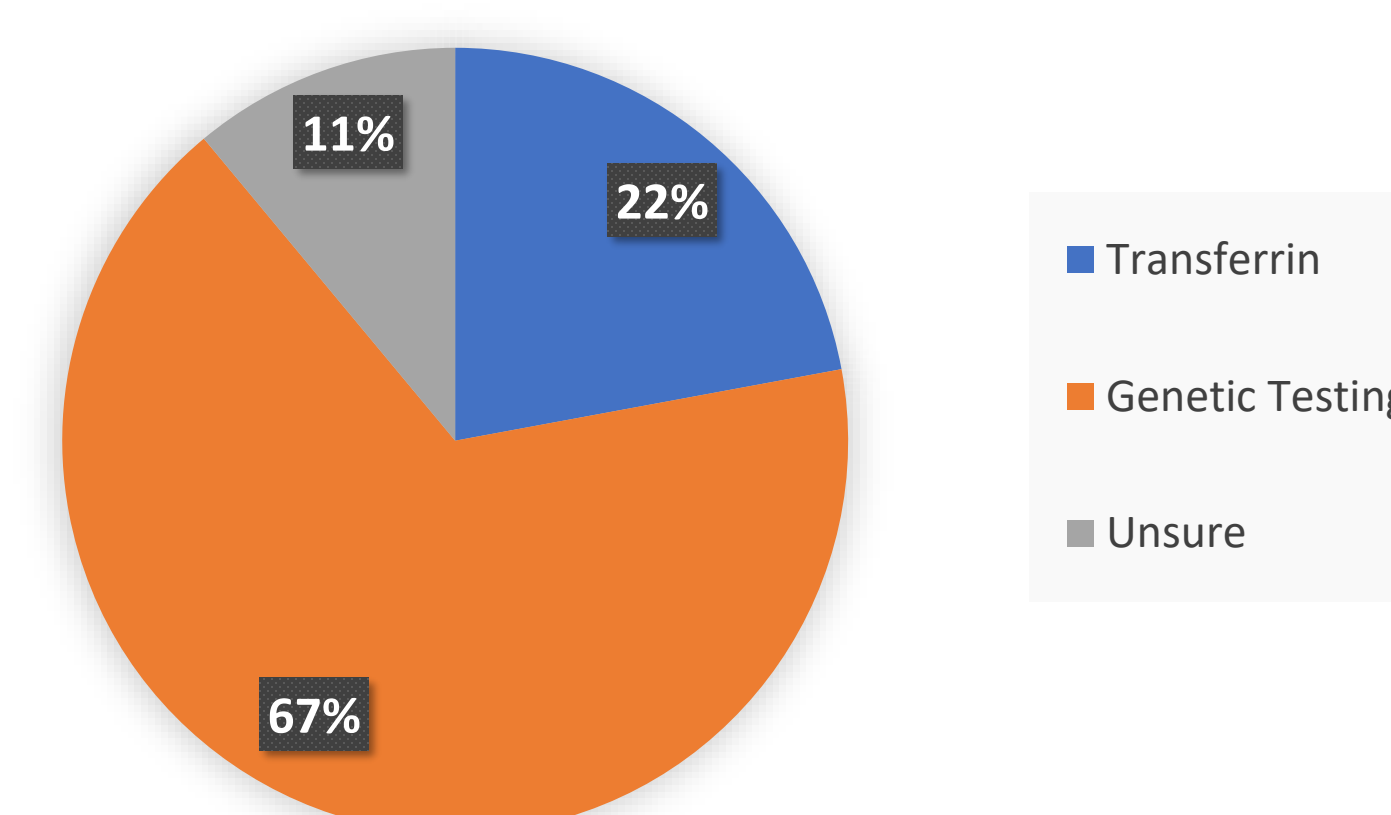
Survey Participation by Patient's Race



How and When is CDG Diagnosed?

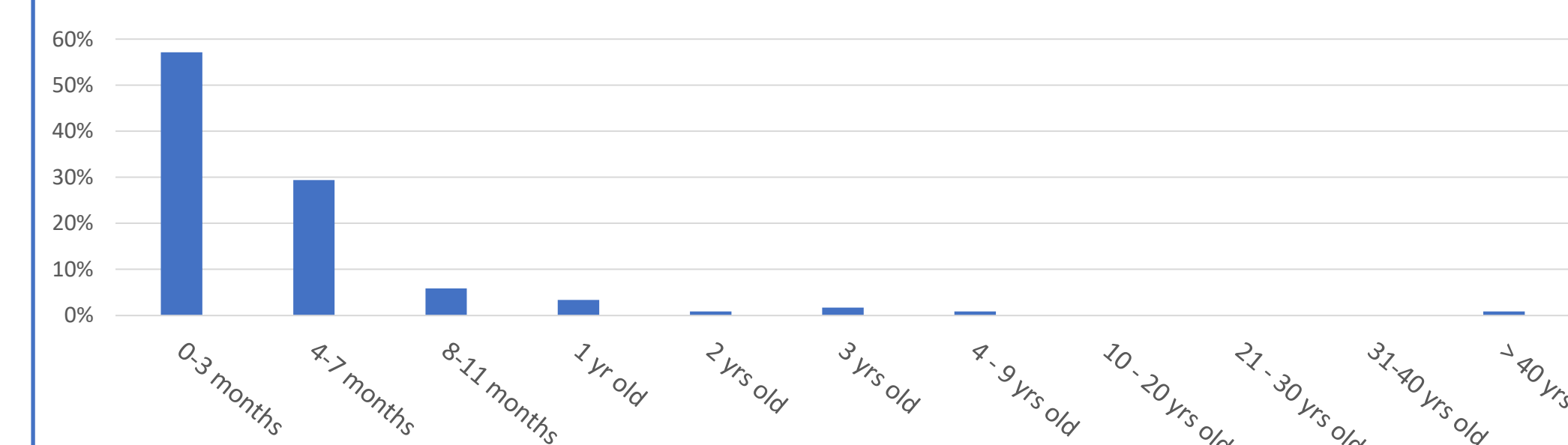
- Genetic testing is the most common and reliable way to diagnose CDG and can also determine the CDG type
- Blood tests to check for many things, including missing sugar building blocks on hormone proteins, coagulation factors, transport proteins, and elevated liver enzymes (ALT and AST)
- A transferrin glycosylation test - checks for missing or incomplete sugar chains

How was the patient diagnosed?

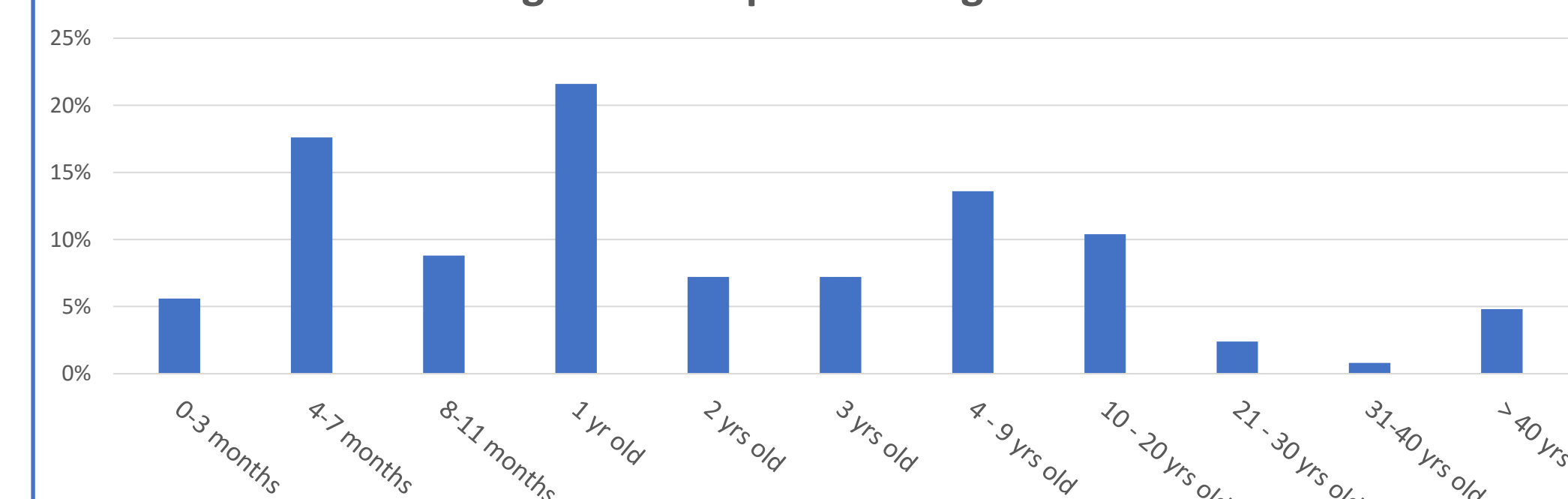


While the average age of diagnosis is 8 – 24 months old, 86% of respondents report that the patient first began experiencing symptoms of CDG between 0 months – 6 months old

At what age did the patient first begin experiencing symptoms of CDG?



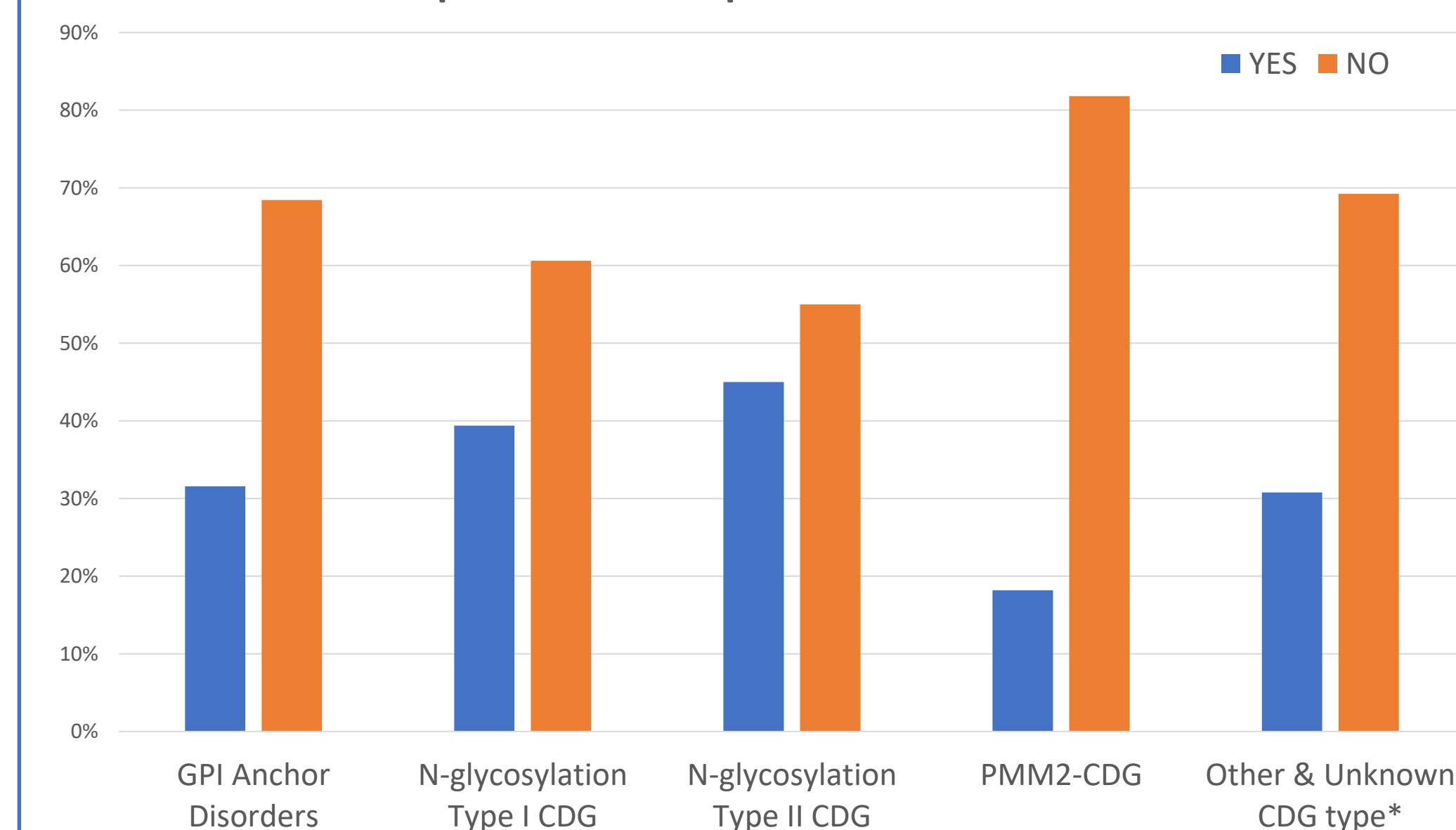
At what age was the patient diagnosed with CDG?



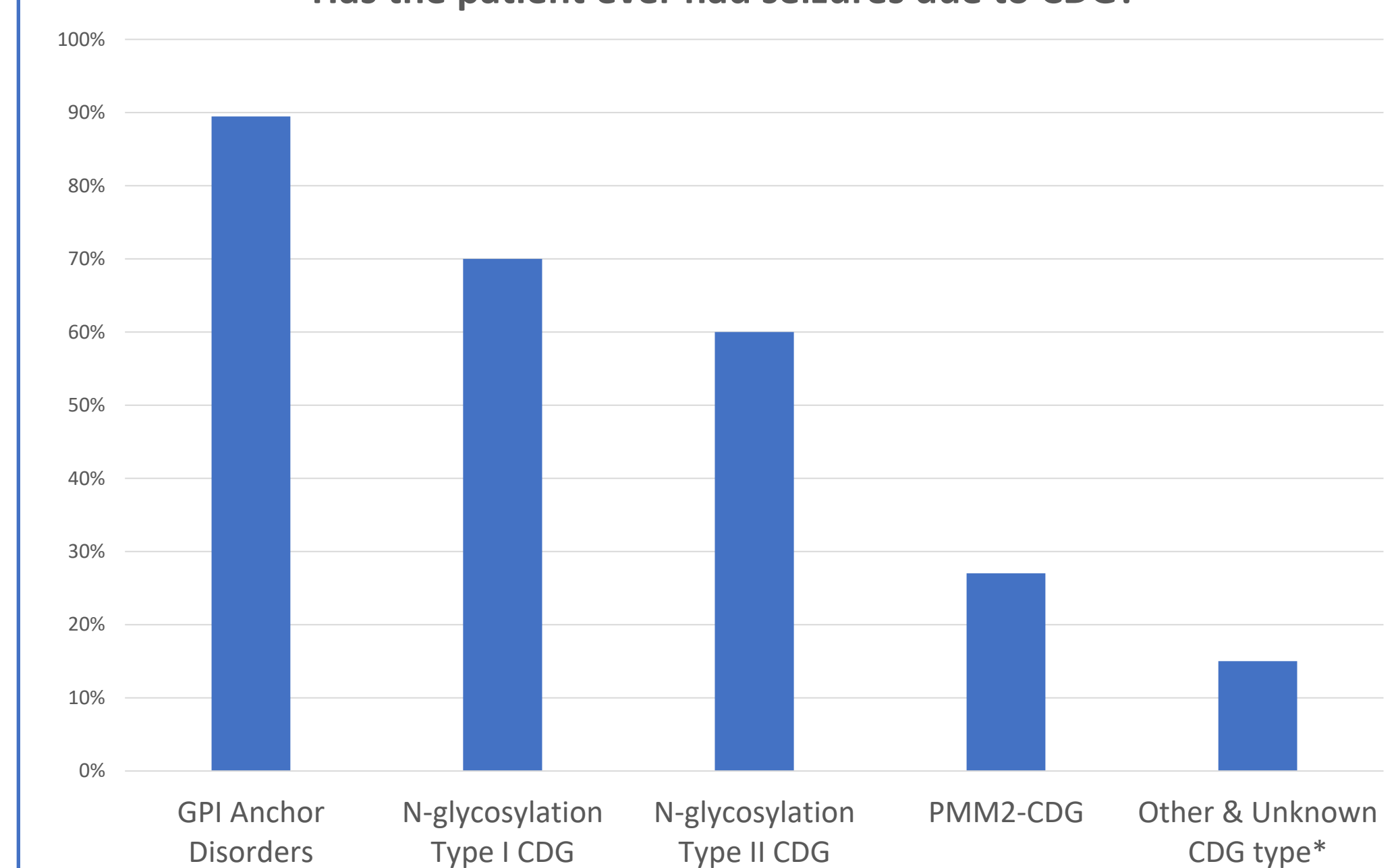
- Diagnosis varies with access to specialist care, as well of severity of symptoms & misdiagnosis
- CDGs should be suspected in any child with multiple unexplained health concerns including those who present with: hypotonia, failure to thrive, developmental delay, liver disease, elevated liver enzymes, coagulopathy, esotropia and seizures

Clinical Findings & Research Opportunities

Has the patient ever required a G-tube due to CDG?

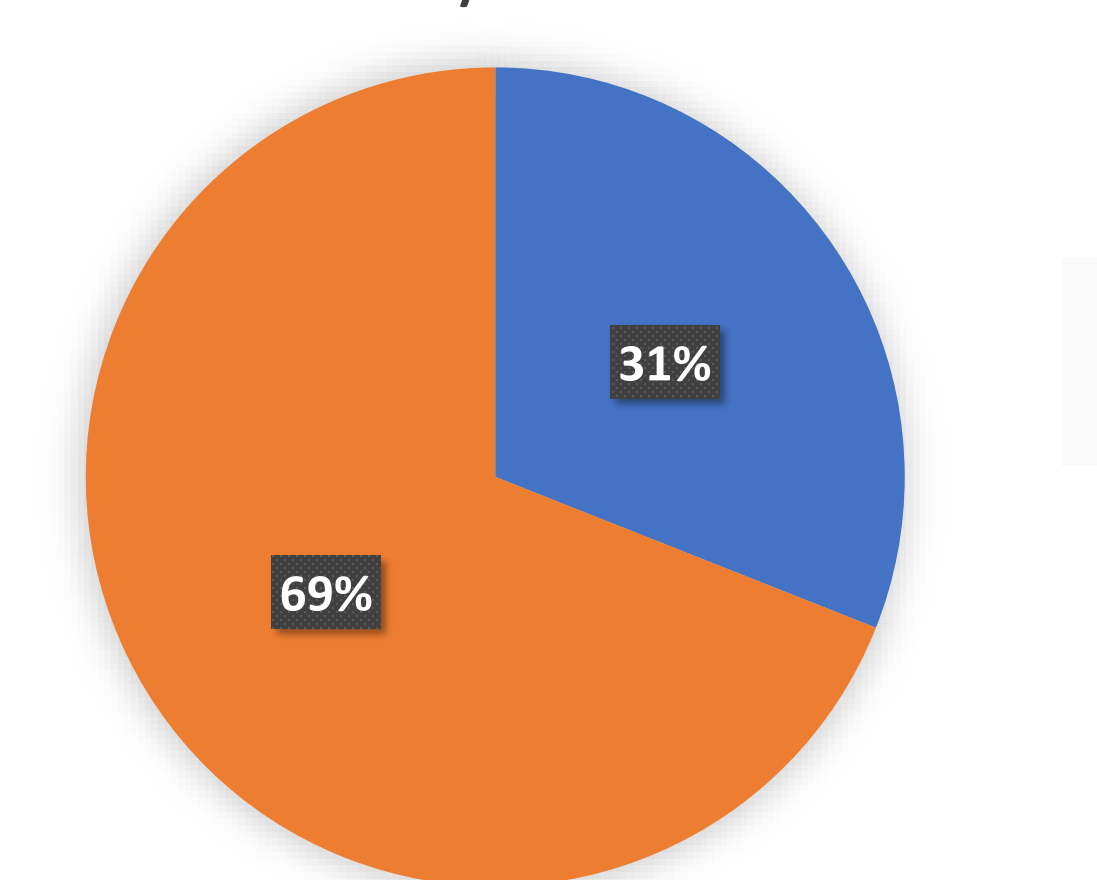


Has the patient ever had seizures due to CDG?



Current Research Involvement & Interest

Has the patient ever participated in a research study and/or a clinical trial?



Is the patient willing to participate in a research study and/or a clinical trial?

