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Qualitative Interviews of Adults with Classic Galactosemia (CG) and their Caregivers: Disease Burden and Challenges with Daily Living

Evan Bailey[1], Stella Wang[1], Jason Randall[2], Carolyn Sutter[3], Lydia Raither[2], Riccardo Perfetti[1], Claire Burbridge[2], & Shoshana Shendelman[1]

[1]Applied Therapeutics, 545 Fifth Avenue, Suite 1400, New York, NY 10017; [2]Clinical Outcomes Solutions, Unit 68 Basepoint, Shearway Business Park, Shearway Road, Folkestone, Kent, CT19 4RH, UK; [3]Clinical Outcomes Solutions, 53 W Jackson Blvd, Suite 1150, Chicago, IL 60604,

Background

- Classic Galactosemia (CG) is a rare inborn metabolic disease caused by an autosomal recessive mutation that severely depletes galactose-1-phosphate uridylyltransferase (GALT), leading to accumulation of galactose and galactitol.[1,2] The condition is fatal in infancy if galactose is not eliminated from the diet.
- Galactitol is an aberrant toxic metabolite which affects the central nervous system (CNS) causing progressive deterioration in multiple functions.[3]
- Understanding the patient perspective of the condition is a key part of any clinical trial and is set out in the FDA PFDD guidance. [4]

Objectives

- To explore the patient experience of CG, including symptoms (onset and progression), and impact on the individual, caregiver, and family.
- To explore which symptoms and impacts were the most critical problems in CG, and how they impact daily life and health-related quality of life (HRQoL).

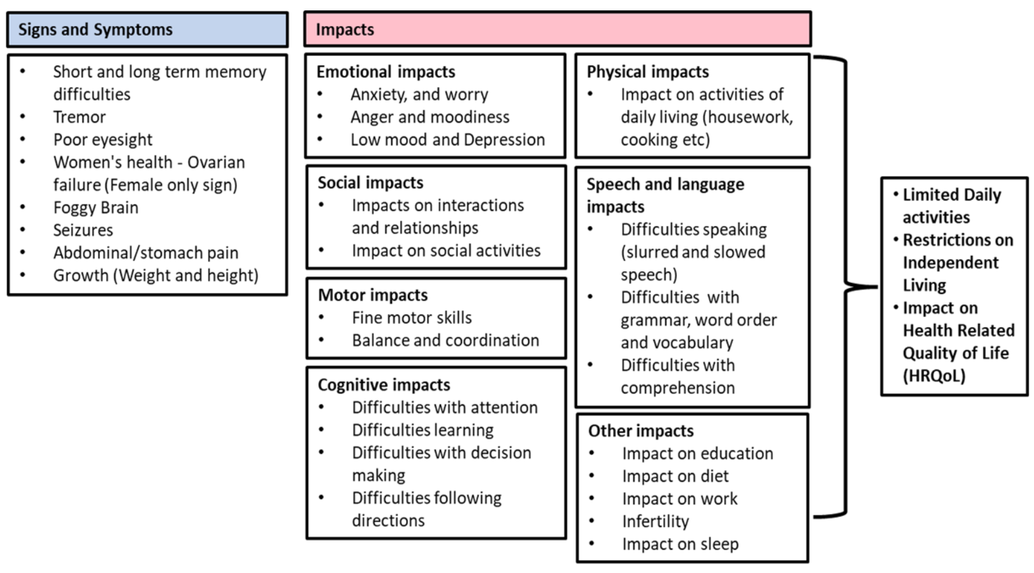
Methods

- Adult patients with CG, and their caregivers, who were taking part in Applied Therapeutics' clinical trial for Galactosemia were approached for participation in this study. (Study AT007.1001 Part D Extension)
- 60-minute semi-structured qualitative interviews, were conducted over the telephone, and analyzed using Thematic analysis.[5]

Results

- N=20 interviews were conducted with 12 adult patients and 8 caregivers (caregivers were caregivers to 9 of the 12 adult patients taking part; 1 was caregiver to 2 patients in the study).
- Of the 12 adult participants, 42% were female, 58% were male. Age range was 19-46 years (Median age 24). Caregivers were 75.0% were female, 25% male and age range was 45-68 years (Median age 55.5). All participants and caregivers were white, non-Hispanic, which is in line with the genetic manifestation of CG.
- 100% of patients required caregiver support. 67% of patients lived with their caregiver and required constant support, 33% lived semi-independently, as they still required support from family due to the day-to-day challenges.
- The patient led conceptual model of CG is presented in Figure 1. with the frequency of reported signs and symptoms in Figure 2 and the frequency of reported impacts in Figure 3. Figure 4 contains quotes from patients/caregivers.

Figure 1. Patient Led Conceptual Model of CG



Note: Some caregivers also discussed how their child experienced jaundice as a newborn. As this is a sign that only occurs as a new-born, it has not been included in the conceptual model.

Figure 2: Frequency of patient reported signs and symptoms

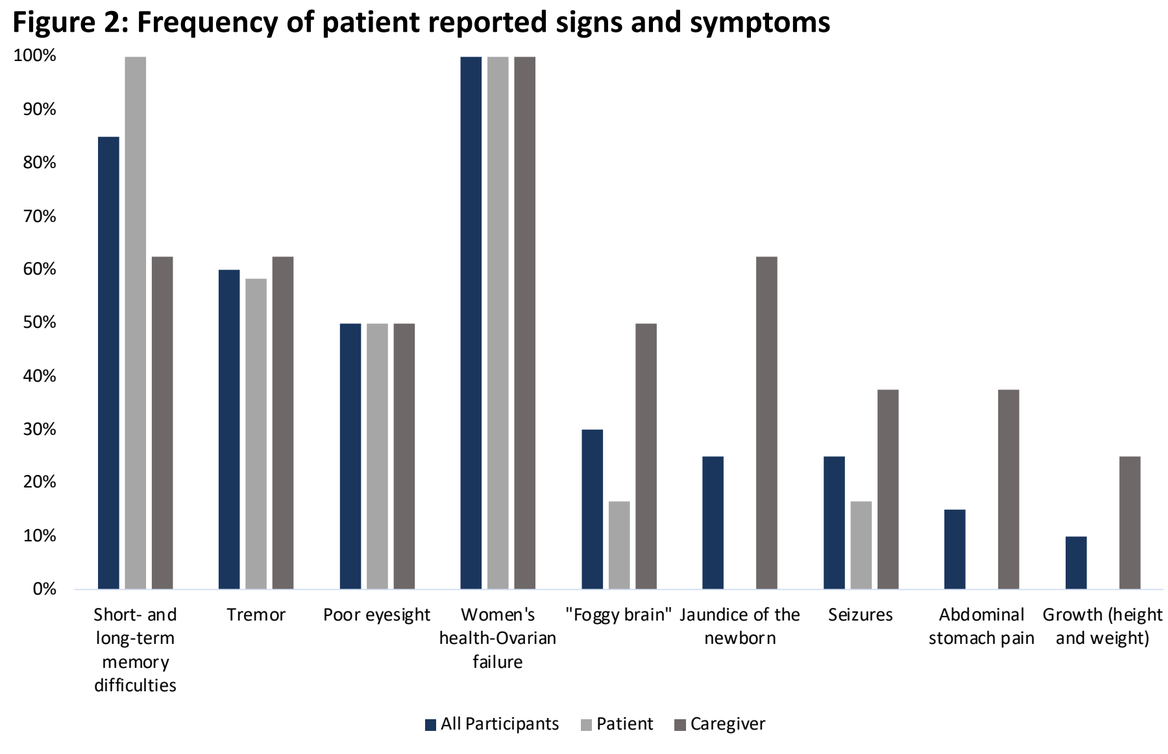
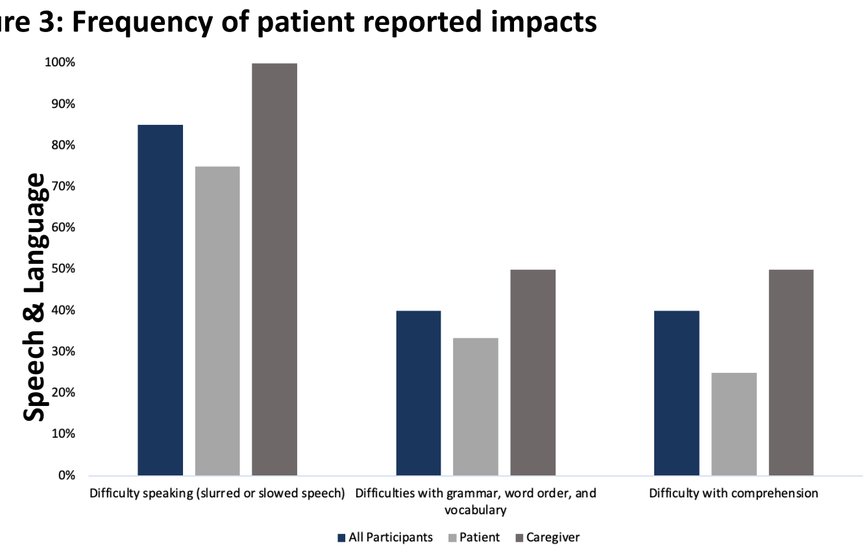
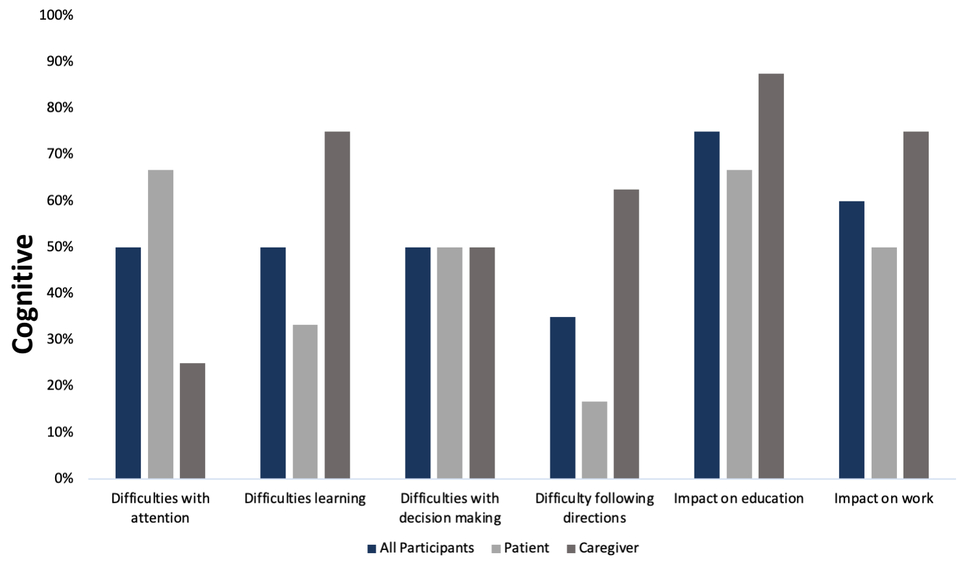
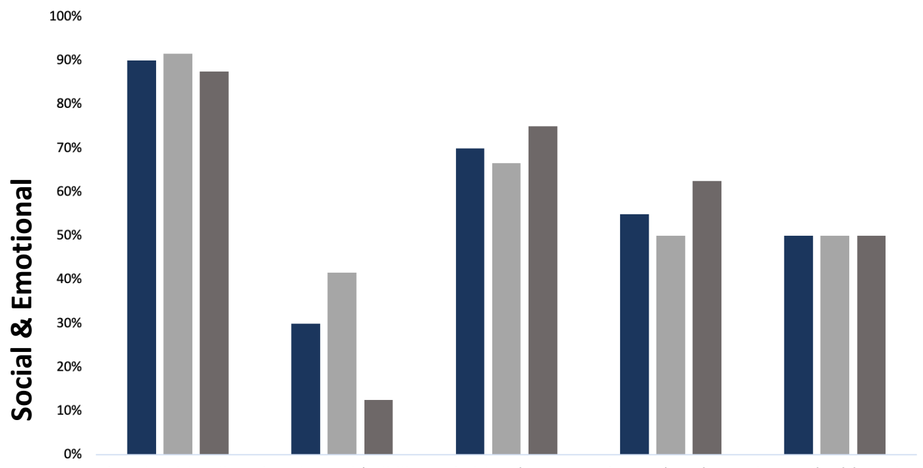


Figure 3: Frequency of patient reported impacts







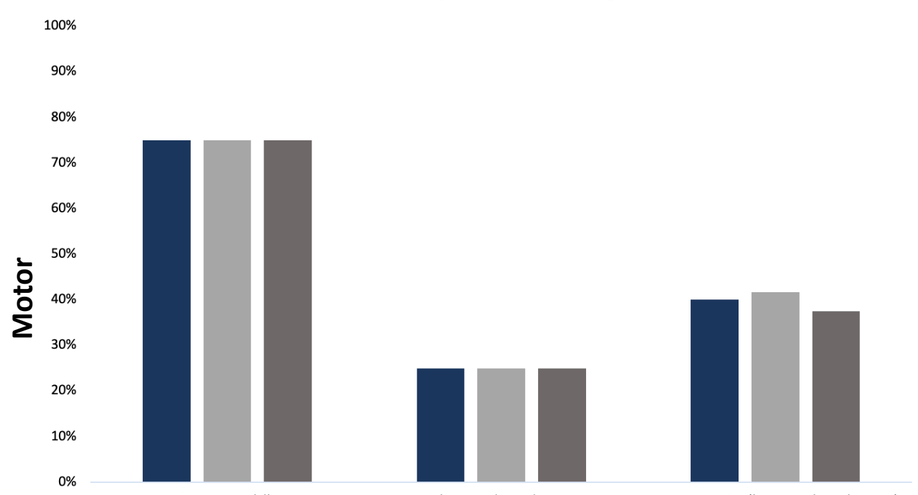


Figure 4. Patient/ Caregiver Comments (in their own words)

The shakiness [tremor] is apparent…it can go all the way from the fingertips up to the elbows" **Patient**

"I ended up being diagnosed with depression [due to CG] and… I would [sighs]… It hurts me to say that I basically cried myself to sleep, because I was sad" Patient

"It was difficult communicating with him…he'd get very frustrated because you'd try to ask him something and he couldn't find the words for it" **Parent**

"it's [galactosemia] impacted him quite a bit. He is just is going to always have a different path than another person that is his same age…he's not going to be able to drive or live independently, or go to college, or have a job." **Parent**

"I can just tell [the difference]…I have to use different words when I speak to him than I did with my older son when he was a senior…to make sure he is comprehending" **Parent**

Discussion

- These interviews provided in-depth qualitative data giving real insight into the lived experience of Galactosemia direct from patients and caregivers.
- Galactosemia has a substantial impact across many areas of life affecting both caregivers and patients.
- Findings suggest all patients will require **some degree of support for the rest of their lives**. The level of support needed increases as the condition worsens over time, creating greater impact on the families.
- This study therefore highlights the significant unmet medical need in Galactosemia because of the burden and progression associated with this condition for both patients and caregivers and the severely limited treatment options currently available.

References

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DISCLOSURES:
EB, SW, RP, and SS are employees and stakeholders of Applied Therapeutics Inc.
JR, CS, LR and CB are employees of Clinical Outcomes Solutions