Classical Galactosemia (CG) is a rare inborn metabolic disease caused by an autosomal recessive mutation that severely depletes galactose-1-phosphate uridyltransferase (GALT), leading to accumulation of galactose and galactitol. 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. Understanding the patient perspective of the condition is a key part of any clinical trial and is set out in the FDA PEGO guidance.

Background

- Classic Galactosemia (CG) is a rare inborn metabolic disease caused by an autosomal recessive mutation that severely depletes galactose-1-phosphate uridyltransferase (GALT), leading to accumulation of galactose and galactitol. 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.
- Understanding the patient perspective of the condition is a key part of any clinical trial and is set out in the FDA PEGO guidance.

Objectives

- To explore the patient experience of CG, including symptoms (onsset 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.

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% female, 25.0% 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.

Figure 1. Patient Led Conceptual Model of CG

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 caregiver support. 67% of patients lived with their caregiver.
- Findings suggest all patients will require caregiver support. 67% of patients lived with their caregiver.
- 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 caregiver support. 67% of patients lived with their caregiver.
- 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 caregiver support. 67% of patients lived with their caregiver.
- 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 caregiver support. 67% of patients lived with their caregiver.