

# The Burden of Pharmacological Treatment on Health-Related Quality of Life in People with a Urea Cycle Disorder – A Qualitative Study

Gillian Yeowell (✉ [g.yeowell@mmu.ac.uk](mailto:g.yeowell@mmu.ac.uk))

Manchester Metropolitan University <https://orcid.org/0000-0003-3872-9799>

Danielle Burns

Manchester Metropolitan University <https://orcid.org/0000-0002-5522-5384>

Francis Fatoye

Manchester Metropolitan University

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## Research

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# Abstract

**Background** Urea cycle disorders (UCD) are a group of conditions of inborn errors of metabolism, typically presenting neonatally. Excess ammonia builds rapidly within the body, risking hyperammonemic episodes and potentially death. Long-term management of the condition includes restrictive protein consumption, pharmacological interventions and, in extreme cases, liver transplantation.

Pharmacological treatments such as sodium benzoate and sodium phenylbuturate have proven effective but not without a multitude of negative attributes including poor taste, elevated volume and associated gastrointestinal discomfort that impacts on health-related quality of life. Glycerol phenylbutyrate (GPB) has recently become a widely available pharmacological treatment with early reports of improved qualities. The following study aims to explore the burden of pharmacological treatment on health-related quality of life in people with a UCD.

**Results** 9 carers of people living with a UCD were interviewed regarding their experiences of pharmacological treatment in relation to their, and their dependent's health-related quality of life after transitioning to GPB. Three main themes were identified following data analysis: psychological health, physical health and social participation. Carers struggled with anxiety surrounding their dependent's condition and the relentless battle of administering medication. Medication administration was perceived to have improved since the transition to GPB, alleviating distress for both carer and dependent. Issues involving schooling were commonly described, ranging from difficulties integrating their dependent into mainstream schooling and the impact of treatment on participation in school and extracurricular activities. Based upon participant's experiences, it could be suggested that some burden had been relieved by the transition to GPB. However, it appeared that the burden would persist despite treatment, owing to the continuing nature of the condition.

**Conclusions** Adhering to a strict pharmacological regime caused immense stress for both carers and dependents, severely impacting on typical social activities such as eating at a restaurant or going on holiday. GPB was perceived to have alleviated some burden in terms of administration. Practitioners should consider these findings when making clinical decisions for UCD patients and the effect of pharmacological treatment on health-related quality of life. Emotional support resources should be made readily available to families to assist with daily living.

## Full Text

Due to technical limitations, full-text HTML conversion of this manuscript could not be completed. However, the latest manuscript can be downloaded and [accessed as a PDF](#).