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UNDERSTANDING THE LIVED EXPERIENCES OF ADULT PATIENTS WITH CITRIN DEFICIENCY: RESULTS FROM AN IN-DEPTH SURVEY

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Background: Citrin deficiency (CD) is an inherited metabolic disorder and a urea cycle disorder (UCD) caused by pathogenic variants in *SLC25A13*, which encodes the mitochondrial aspartate–glutamate carrier 2 (AGC2, "citrin") that transports glutamate into—and aspartate out of—the mitochondrion to support the urea cycle. How this biochemical defect translates into day-to-day symptoms and quality-of-life burden, especially among adults patients not with the most severe onset, remains poorly characterised. To address this gap and to support the development of patient-reported outcomes (PROs), we examined lived experiences in adults with genetically confirmed CD.

Methods: We conducted a mixed-methods study with seven adults aged 20 to 55 from our registered patient cohort, each with genetically confirmed CD. We first carried out six in-depth interviews; their findings were used to develop a self-administered, 64-question questionnaire, with opportunities for real-time feedback and clarification.

Results: Quality-of-life impacts were prominent. Fatigue was universally reported in all patients that participated in the survey and substantially limited daily functioning. Anxiety was common with 5 patients out of 7 experiencing it and affecting school, social, and family life, and often led to social withdrawal. Frequently reported clinical symptoms included elevated cholesterol and hypoglycaemia-like episodes in five patients underscoring their burden in this group.

Conclusions: Findings highlight two priority domains, quality of life and clinical symptoms, that should be addressed in CD care and research. This study provides useful information to aid the development of validated PRO measures for future clinical trials and supports a more patient-centered approach to CD research and management.