

DERIVING VIGNETTES FOR A RARE DISEASE USING PARENT, CAREGIVER AND CLINICIAN INTERVIEWS TO EVALUATE THE IMPACT ON HEALTH-RELATED QUALITY OF LIFE

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Background: Capturing health-related quality of life (HRQoL) through health utilities is critical to evaluating treatments, including their cost-effectiveness. HRQoL, however, is often difficult to measure in ultra-rare diseases, especially in pediatric populations such as AADC deficiency (AADC-d) since utility elicitation through proxy caregiver/parent ratings is challenging. HRQoL data may instead be derived through vignette studies and discrete choice experiments (DCE) using the general public. It is crucial to include input into the design from caregivers/parents of AADC deficiency patients, and clinicians treating these patients, to ensure face and content validity. The objective of this study was to develop vignettes and DCE attributes using caregiver and clinician input. These will be used in a subsequent study to estimate patient and caregiver HRQoL.

Methods: Initial vignettes were developed based on a literature review, discussions with 3 clinicians, as well as from a caregiver/parent advisory board. In a second step 3 caregivers/parents were asked to provide input into the descriptions via an anonymised survey to ensure the vignettes were capturing aspects pertinent to the disease. Additional input was obtained from 4 clinicians currently caring for children with AADC-d during an ad-board. To capture improvements on therapy, input into key DCE attributes were also obtained via a ranking of attributes from a clinical, patient and caregiver perspective.

Results: Based on the clinical trials, vignettes describing 5 health states were developed using all input collected: bedridden, head control, sitting unsupported, standing with assistance and walking with assistance. For the DCE, six attributes were identified: mobility, muscle weakness, oculogyric crises, feeding, cognitive impairment, and crying.

Conclusion: Vignettes and attributes of AADC-d were developed through consultation with carers and clinicians. These will be used in a subsequent utility elicitation and DCE study with the general public to derive utilities for a cost-effectiveness model of an AADC-d treatment.