

## Title: Development of a patient-reported outcome measure to assess health-related quality of life of children, adolescents, and young adults with cystinosis – QUALIFY-US

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### Background

Cystinosis is a lysosomal storage disease with an incidence of 1 in 100-200 thousand live births. Biallelic mutations of the *CTNS* gene cause it. *CTNS* encodes the lysosomal membrane protein cystinosin, which transports cystine out of lysosomes. When cystinosin is deficient, cystine accumulates in the lysosomes, resulting in crystal formation and cell death in several different tissues and organs.

The introduction of oral cysteamine therapy in the early 1980s has changed the course of cystinosis from a primarily lethal to a chronic multiorgan disease. Both the disease-specific symptoms and the therapy significantly impact the health-related quality of life (HrQoL). The few studies examining HrQoL in patients with cystinosis suggest a limited HrQoL for patients and their caregivers. Primarily generic instruments are used; no disease-specific patient-reported outcome measures (PROMs) exist so far.

### Objectives

This study aims to develop and validate a disease-specific PROM to measure HrQoL in children, adolescents, and young adults with cystinosis in Germany, France, Spain, and the United States of America.

### Methods

Questionnaire development includes the following steps: (1) a semi-structured interview guide based on a systematic literature review and feedback from physicians, patients, and representatives from patient organizations, (2) focus (group) interviews in all participating countries to identify relevant aspects of the disease-specific HrQoL, (3) data analysis and item formulation for the preliminary questionnaire, including a cognitive debriefing (CD), (4) a field and re-test for psychometric testing, including test-retest reliability, internal consistency, and convergence validity. Also, factors influencing the HrQoL are analyzed using clinical and sociodemographic data.

### Results

We will present the preliminary results of the qualitative interviews with patients and parents in Germany, describing essential aspects that influence HrQoL in patients with cystinosis that will be the basis for item development.

Outlook: The expected final result is a cross-cultural, psychometrically validated, practically feasible, and conceptually suitable instrument to assess HrQoL in children, adolescents, and young adults with cystinosis. It will be available in German, English, Spanish, and French for further (inter-)national studies. Further, the study will characterize the HrQoL of cystinosis patients.

### Conclusions

A challenge for further research is to unravel the contributions of disease-specific symptoms and psychosocial factors to HrQoL in patients with cystinosis. With a better understanding of the impact of cystinosis and its treatment on the well-being and functioning of affected patients and their families, psychological interventions tailored to patients' needs should be developed and applied to improve patients' HrQoL.

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