Living with Cystinosis as an Adult: Community Insights Study

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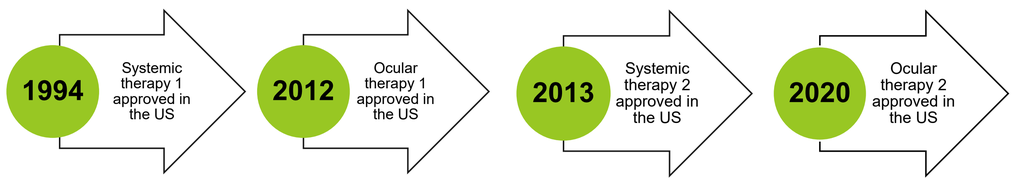
Background

- Cystinosis is an ultrarare, autosomal recessive lysosomal storage disorder caused by pathogenic variants in the CTNS gene, leading to toxic intracellular cystine accumulation and progressive tissue and organ damage [1,2].
- Cystinosis has an estimated prevalence of 2,500 individuals world-wide and 500–600 cases in the United States, although true prevalence is likely underestimated due to diagnostic delays[2].
- Historically, cystinosis was considered a fatal childhood disease; however, kidney transplantation, earlier diagnosis and cysteamine therapy have transformed its course, with many patients living into adulthood[2].
- As the disease advances, virtually all body systems may be impacted, including the endocrine, ocular, neuromuscular, gastrointestinal, and central nervous systems [1,2,5].

Treatment Landscape

- Treatments of cystinosis are finite, and the only systemic disease-modifying therapy is oral cysteamine along with the usage of cysteamine eye drops[2].
- Early treatment is associated with delayed kidney failure, reduced incidence of diabetes, hypothyroidism, and neuromuscular complications[1,5].
- Advances in treatments and transplantations have transformed cystinosis from a fatal childhood disorder into a chronic multisystemic condition where extrarenal complications become increasingly prevalent[2,5].
- Transition to adult care is a critical stage complicated by reduced adherence, fragmented specialty support, and significant mental health impacts for both patients and caregivers[3].
- There is currently a strong emphasis from the cystinosis community regarding how treatments cripple the quality of life for patients and their loved ones.



- Although there is a progression of treatment options for management of cystine levels and advancing treatments are life-enabling, available therapies for cystinosis have led to an evolution of burden that creates a desperate need of new options.

Objectives

Given the evolution of burden to cystinosis as a lifelong condition, the authors have designed and launched the *Living with Cystinosis as an Adult: Community Insights Study*.

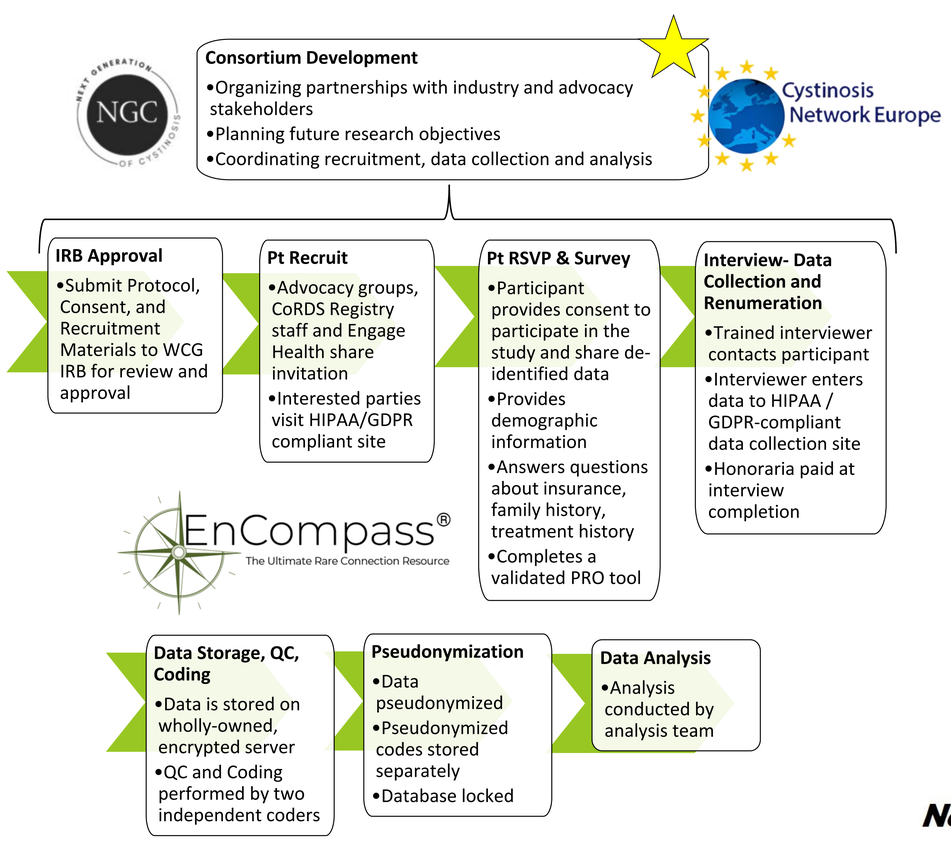
- Primary objective: to improve the understanding of the lived experience of adults diagnosed with cystinosis, including, but not limited to physical symptoms, emotional, psychosocial and quality of life experiences.
- Secondary objectives: to publish findings in order to enrich the medical literature and bring awareness to the scientific, medical and patient community in order to optimize understanding and care.
- Lastly, to inform strategies for long-term support and management of cystinosis in the adult population.

Study Methods

- Participants will be recruited via patient organizations (*Next Generation of Cystinosis, Cystinosis Network Europe*, and others), *the CoRDS Registry*, and *Engage Health's EnCompass® database*. Convenience and snowball sampling will be used, with purposive recruitment to ensure diversity across geography, age, and treatment-related characteristics.
- The team anticipates recruitment of up to 100 international participants who are age 18 years or older and who have been diagnosed with cystinosis, or the parent/legal guardian/primary caregiver of such an individual.
- This is an estimated 6-month study that will require approximately 90 minutes of each participant.

Study Design

- A consortium has been developed to conduct this is a qualitative, observational, non-interventional study.



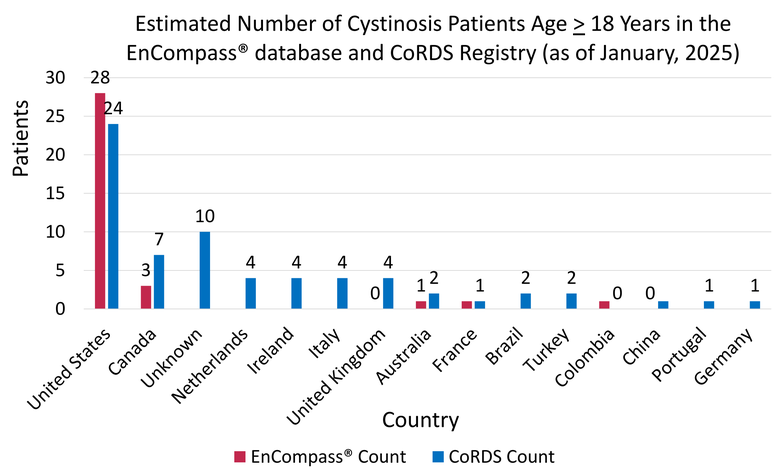
Study Design (continued)

- Interested individuals will complete online recruitment materials and provide informed consent via a secure, web-based platform prior to study participation.
- Consented participants will submit de-identified demographic and disease-related information, including insurance status, family and treatment history, and complete validated patient-reported outcome (PRO) measures that assess quality of life.
- Participants engage in one-on-one interviews conducted through Engage Health's proprietary, HIPAA- and GDPR-compliant system using an IRB-approved interview guide to capture patient perspectives.
- Participant data is pseudonymized, aggregated into a locked analytical dataset, and securely stored on wholly owned servers with controlled access for monitoring, auditing, and analysis.
- Quantitative and qualitative data are analyzed to identify key patterns, trends, and drivers shaping the lived experience of individuals with cystinosis and to contextualize factors most meaningful to patients.

Feasibility Results

Assessment of Adult Cystinosis Patients

- To determine if sufficient numbers of adults diagnosed with cystinosis exist to conduct the study, Engage Health reviewed their EnCompass® database; there are currently 97 patients in the EnCompass® database who have opted-in to be contacted for future research (34 - Age 18+, 59 - Age <18, and 2 - Age Unknown). As of January 2025, the CORDS Registry contained 216 patients with cystinosis (61 Age 18+, 107- Age <18, 47 – Age Unknown).
- Based on this assessment it is likely that the study will be able to recruit up to 100 adults impacted by cystinosis.
- To assess the languages in which to conduct the study, the research team reviewed territories where cystinosis advocacy efforts are in place, where patients have been reported, and assessing the percent of the population in each of these territories who speak English.
  - Based on this assessment, and our recruitment plan, the study team plans to conduct the study in English, German, and French.



Country	% of Population who speak English[6]
United States of America	95.3%
Canada	83.1%
Netherlands	90.9%
Ireland	98.4%
Italy	13.7%
United Kingdom	98.3%
Australia	92.8%
Brazil	5.00%
Turkey	17.0%
China	0.90%
Portugal	27.0%
France	57.3%
Germany	56.3%
Colombia	4.20%

Collaborative Efforts

- The *Living with Cystinosis as an Adult: Community Insights* study team was assembled by Next Generation of Cystinosis and their Medical Advisor, Dr. Jeanine Jarnes (University of Minnesota) and Engage Health.
  - Dr. Maya Doyle serves as the study's medical advisor, whereas Dr. Julia Quitmann and Dr. Steffi Witt (authors of the QUALIFY and LITERACY studies) were consulted in the survey and interview guide design.
- Our team seeks additional corporate sponsors for support of recruitment, fielding, analysis and publications efforts.
  - Any interested parties should reach out to Austin Letcher at aletcher@engagehealth.com.

Discussion & Next Steps

- Advances in cysteamine therapies have significantly improved survival for individuals living with cystinosis, resulting in a growing adult population with an emerging disease phenotype.
- Adults living with cystinosis now experience a range of long-term and delayed complications that were once underrecognized and for which care guidelines are limited.
- This study is anticipated to begin recruitment in early March 2026 and will contribute to addressing a critical gap in cystinosis research by capturing patient-reported experiences and outcomes among adults with cystinosis. By encouraging patients to share data about their lived experiences, this work establishes a foundation to support broader cystinosis research efforts.
- Importantly, insights generated from this study can inform clinical care, guide cystinosis therapy efforts, and strengthen advocacy efforts for improved adult cystinosis management and support.
- Our future efforts prioritize establishing long-term, multi-stakeholder partnerships to sponsor continued research focused on adult cystinosis. These collaborations will be critical for expanding participant cohorts, enabling long-term follow-up/patient relationships?, and ensuring that adult cystinosis patient voices are acknowledged and translated into meaningful action that addresses improvements in care, patient needs, and support.

Acknowledgements

We sincerely thank our medical advisor, Dr. Maya Doyle (Quinnipiac University) as well as Dr. Julia Quitmann and Dr. Steffi Witt (University Medical Center, Hamburg) for their substantial contributions to the study survey and interview guide design. We also acknowledge *Cystinosis Network Europe* for their support in recruiting members of the cystinosis community outside the United States. Their participation will make this project possible.

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Next Generation of Cystinosis
Run By & For Adult Patients