

## **6-Month Progress Report**

**Funding Source: Cystinosis Research Foundation**

**Funding Period: September 1, 2013 – August 31, 2014**

### **Patient-Reported Outcome and Health-Related Quality of Life in Adults with Cystinosis: A Study Utilizing the NIH “PROMIS”**

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#### Project Overview and Specific Aims

The advent of renal transplantation and cysteamine therapy have served to extend the lifespan of individuals with cystinosis into adulthood, but they do not prevent the ultimate progression of the disease. For this reason, it is important to learn about the longer-term effects of cystinosis on quality of life in adults with the disease, as well as the final illnesses and causes of death in this population. This knowledge can then be used in the planning of and/or justification for emerging therapies and treatments for adults living with cystinosis.

**Aim #1:** The first aim of the proposed study is to provide vital information on patient-reported outcome and quality of life in the expanding population of adults living with cystinosis.

**Aim #2:** Although treatment is serving to extend the lifespan of patients with cystinosis, there is currently no available cure and the disease and/or its complications are fatal. A second aim of this study is to gather information on the final illnesses and specific causes of death in adults who had cystinosis and passed away during the past 10 years.

**Aim #3:** This study represents a 1-year extension of the original 1-year study, so that information could be obtained from a larger and more representative sample of the cystinosis population.

#### Progress in Meeting Aims of Study

##### Recruitment

We continue to identify and recruit participants for this study. We have been utilizing both our group and individual contacts, including the following:

We have established alliances with numerous cystinosis and related groups or organizations, both nationally and internationally. These include the Cystinosis Research Foundation (CRF); Cystinosis Research Network (CRN); Cystinosis Foundation; Cure Cystinosis International Registry (CCIR); NYU Emerging Adulthood Study; AIRG France; Cystinosis Australia (support group & Facebook page); German, Brazilian, Italian, South African, Mexican, and Canadian support groups/associations. Our approved recruitment advertisement is periodically sent to these groups and organizations for posting/distribution, thereby reaching large audiences within the cystinosis population.

In terms of recruitment of individuals we know to be eligible, we have identified potential participants through cystinosis meetings and cystinosis media, including publicly available websites and magazines/newsletters. We have also solicited participation from individuals who indicated in advance that they would be interested in participating in this new study, and from individuals in our research records from previous studies.

Lastly, we have been following-up with individuals who indicated their intent to complete the PROMIS questionnaire but who have not yet done so. Some individuals have found it difficult to complete due to personal/medical demands. We have also been following up with parents/nearest living relatives, several of whom have found it difficult to participate, and we are attempting further outreach to enroll additional parents/nearest living relatives of deceased individuals with cystinosis.

The outcome to date of these recruitment efforts is shown below:

Broad Advertising w/ Recruitment Ad	<b>14</b> groups/organizations
<i>Recruitment of Adults with Cystinosis</i>	
Number of cystinosis individuals who have completed PROMIS	<b>42</b> adults with cystinosis
Number of cystinosis individuals who have been sent the link but who have not yet completed the PROMIS	<b>20</b> adults with cystinosis
<i>Recruitment of Parents/Relatives</i>	
Number of parents/relatives we have personally contacted	<b>7</b> parents/relatives
Number of parents/relatives who have completed PROMIS	<b>2</b> parents/relatives
Number of parents/relatives who indicated intention of participating but who have not yet done so	<b>5</b> parents/relatives

### Participant Enrollment

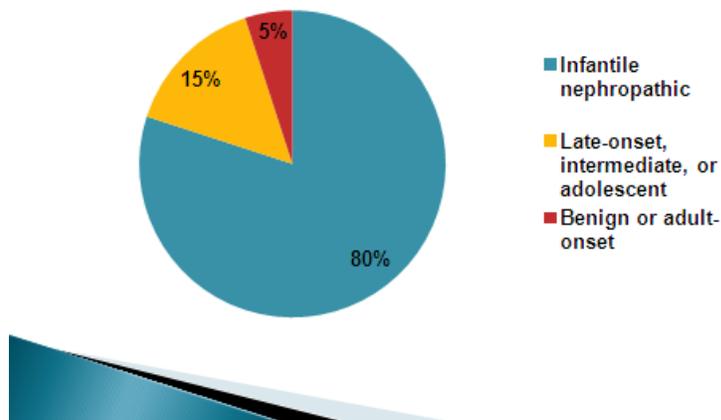
As of 3-1-14, 42 adult participants with cystinosis completed the study (20 males, 22 females), and 2 parents have completed the study. Preliminary data for the adult group follows.

### Preliminary Data for Adult Sample

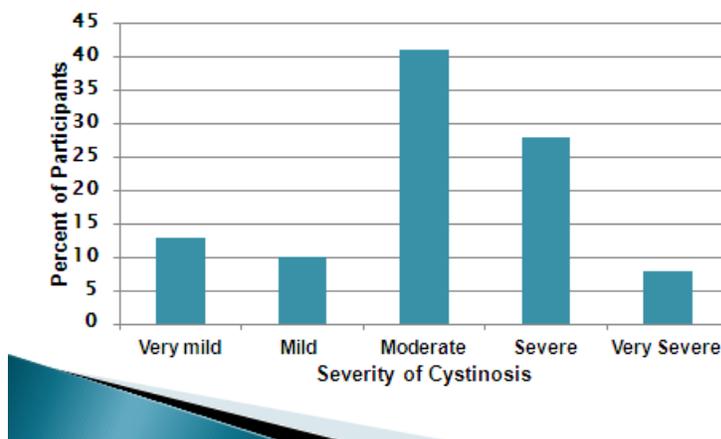
The age range of the sample is 18 to 59 years old, with a mean age of 31 years. The data presented below represent all participants, and summarize a sampling of the general indices within the study. Once data collection is complete, more detailed data analyses will be performed.

The tables and figures that follow show demographic information, compliance, other medical diagnoses, and select quality of life indices.

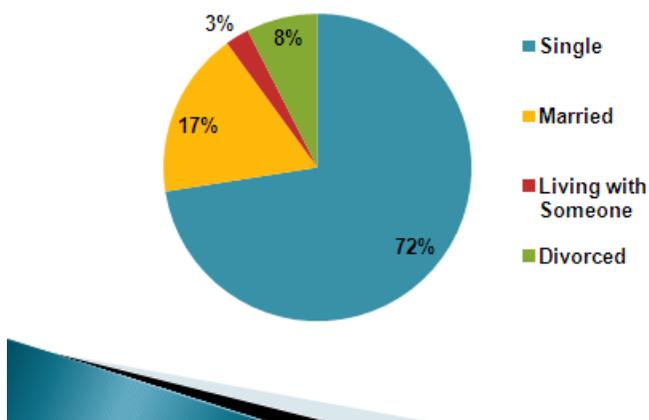
## Type of Cystinosis



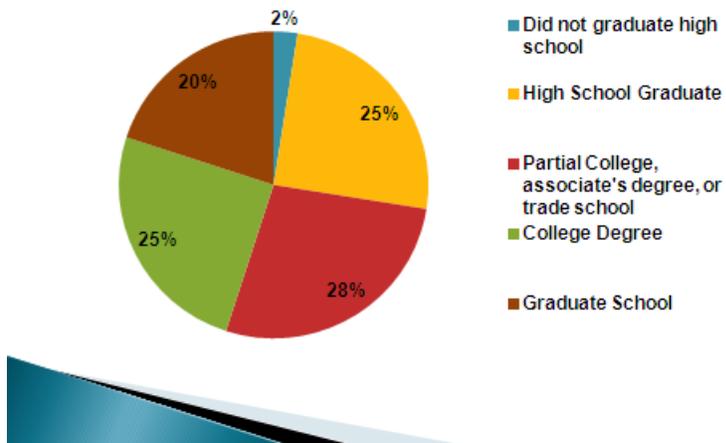
## Severity of Cystinosis



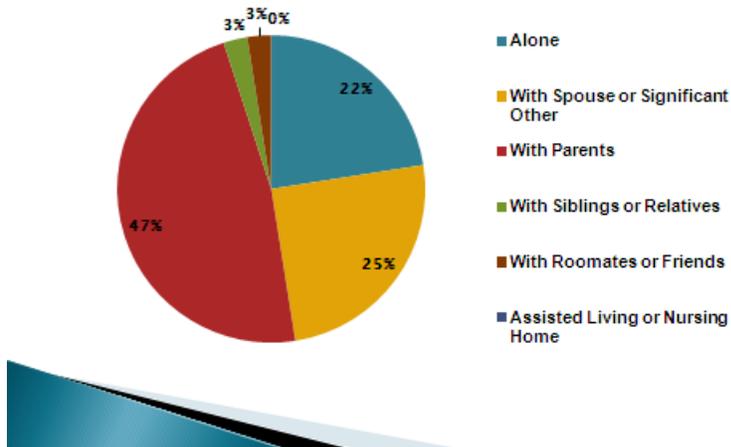
## Marital Status



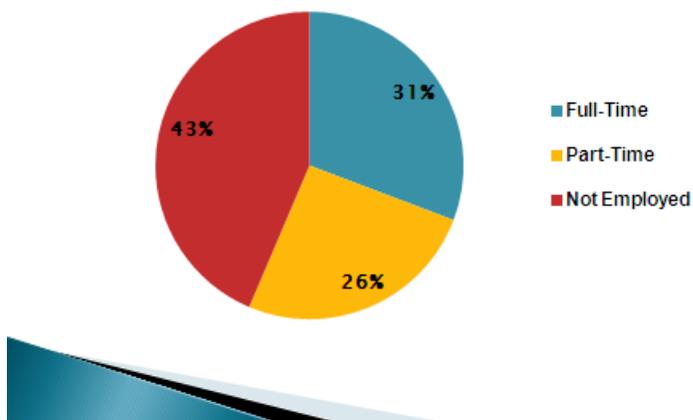
## Educational Status



## Living Situation



## Employment Status



## Compliance:

- ▶ “I always take my pills and use my eye drops at the prescribed time”:

Percent “yes” = 26%

...What about the other 74%?



## Top Reasons for Non-compliance

I just did not remember	69%
I did not have any of my pills/eye drops with me when it was time for me to take them	51%
I was too busy	33%
Skipping an occasional dose doesn't really matter	23%
I take so many medications that sometimes I don't remember to take one of them	21%



## Other Reasons for Non-compliance

I don't want to take the medicine at work/school	18%
I am tired of taking medicine	15%
I am tired of living with a medical condition	13%
I do not like taking medication, so sometimes I just skip it	13%
I believe the medicine has too many side effects	13%
“Other”	36%



## Medical Diagnoses

Medical Condition with Representative Examples	Percent
<b>Endocrine, Lymphatic, or Metabolic Conditions</b> Examples: Cystinosis, diabetes, thyroid gland conditions, nutritional deficiency, other endocrine conditions	100%
<b>Kidney/Urinary, Genital, and Reproductive Conditions</b> Examples: Kidney infection, kidney or bladder stones, renal failure, other nephrotic or urologic conditions	87%
<b>Eyes, Ears, Nose, Throat</b> Examples: Difficulty swallowing, eye or retina disorders, photophobia, sinusitis, hearing loss	79%

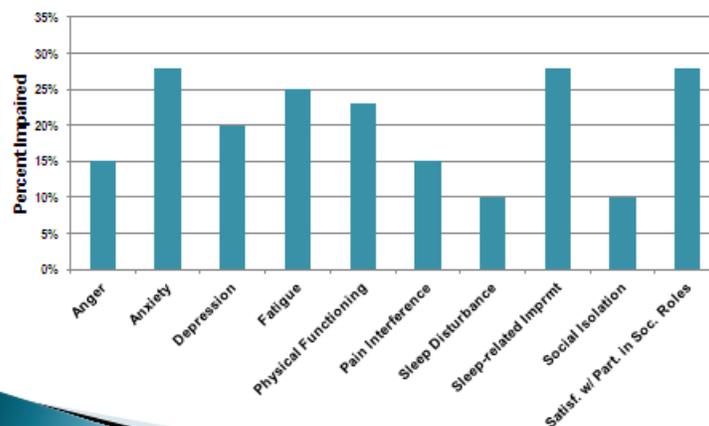
Medical Condition with Representative Examples	Percent
<b>Muscle, Skeletal, or Skin Conditions</b> Examples: Arthritis, muscle weakness, swallowing difficulty, orthopedic deformity, short stature, osteoporosis, broken bones, fractures, tendon ruptures	74%
<b>Circulatory, Heart, or Blood Conditions</b> Examples: Anemia, cardiomyopathy, blood transfusion, high blood pressure, high cholesterol, blood clots	54%
<b>Pain, Body Sensations</b> Examples: Stomach pain, back pain, pain in arms/legs/joints, numbness/tingling in feet or legs, headaches, dizziness, fainting, feeling heart pound or race, shortness of breath, chills/sweats, swelling/edema	59%
<b>Digestive/Gastrointestinal Conditions</b> Examples: Gastroesophageal reflux, inflammation of esophagus, gastritis, hernia, inflammation/infection of intestines or abdomen, irritable bowel syndrome, pancreatitis, gall stones	47%

Medical Condition with Representative Examples	Percent
<b>Respiratory/Lung Conditions</b> Examples: Allergies, asthma, pneumonia, sleep apnea, other inflammatory lung diseases and respiratory conditions	42%
<b>Neurological Conditions</b> Examples: Learning disabilities, speech difficulties, sleep disturbances, migraine headache, neuropathy, seizures, cognitive decline, stroke, paralysis	51%
<b>Mental Health Conditions</b> Examples: Depression, anxiety disorder, bipolar disorder, substance abuse, eating disorder, seasonal affective disorder	28%
<b>Cancer</b> Examples: Cancer, benign tumors, leukemia/lymphoma	10%
<b>Other:</b> Examples: Immune deficiencies/disorders, toxic effects of drugs or poisoning, septicemia/septic shock	30%

## Oral-Motor Functioning

- ▶ **Approximately 33%:**
  - coughing / choking / gagging during meals
- ▶ **Approximately 40%:**
  - have a breathy voice or difficulty talking loudly / articulation problems (difficulty pronouncing words clearly)
- ▶ **Approximately 66%:**
  - eat very little / have a poor appetite / take a long time to eat

## Quality of Life–Standard Scores



## Psychological Impact of Illness

- ▶ Overall, areas of relatively greater difficulty include:
  - Feeling inferior to others
  - Fear/worry about the future
  - Having trouble asking others for help
  - Hard to get close to people
  - Having lost close relationships
  - Lacking confidence
  - Feeling like a burden to family
  - Hard to accept the way things work
  - Difficulty dealing with uncertainty

### Future Plans

We plan to keep the study open for enrollment for an additional 4 months, during which time we will continue to advertise and actively solicit participation from adults and parents/nearest living relatives. The final 2 months will be a period for analyzing data, interpreting results, and preparing a manuscript for publication.