Cystinosis Research Foundation

*Lay Abstract Template for Awardees*

Spring 2013 Grants

Please complete this lay-oriented grant abstract form which will be published on the CRF web site and in the CRF Star Facts with announcement of your award. Please do not exceed 350 words total. Please submit this form to us as a Word file.

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**Principal Investigator (s)**: Stephanie Cherqui

**Project Title**:Pharmacology/Toxicology studies for gene-modified stem cell transplantation for cystinosis

**Objective/Rationale**: Please write a lay-oriented statement of the scientific rationale for this project. Approximately 75-85 words.

Cystinosis is an inherited disease involving the defect of the gene *CTNS*. It causes cystine, a naturally occurring protein degradation products in the cells, to accumulate to toxic levels in the body. This leads to cell death and tissue damage to the kidneys, liver, eyes, muscle and brain. There is a treatment available that delays, the onset of these health problems, but it does not cure the disease. Thus, there is a pressing need for a better treatment for cystinosis.

**Project Description**: Please write a brief, lay-oriented description of how you will carry out the project. Approximately 125-130 words.

The long-term goal of this project is to develop a new treatment for cystinosis using gene therapy. It requires that the patient’s own bone marrow stem cells be isolated and modified in the laboratory through the introduction of a normal version of the *CTNS* gene. These modified cells are then transplanted back into the patients to create a reservoir of healthy stem cells in the bone marrow that can rescue the organs throughout the patient’s lifetime.

Preliminary studies have been done in the mouse model of cystinosis, the Ctns-/- mice, which develop similar symptoms as the patients. We transplanted mice stem cells that had been modified in the laboratory through the introduction of a functional *CTNS* gene into the Ctns-/- mice. This treatment significantly improved the disease in this model.

**Relevance to the Understanding and/or Treatment of Cystinosis**: Please explain how the project will impact cystinosis treatment or increase our understanding of cystinosis. Approximately 75 words.

This proposal represents the safety and efficacy studies required by the Food and Drug Administration (FDA) that will establish whether it is warranted to test this therapy in humans who have cystinosis. This includes testing the impact of cysteamine on the stem cell transplantation in order to determine if cysteamine will be able to be used by patients who got transplanted with stem cells. This also includes the collection of information related to the natural history of cystinosis for the design of the clinical trial.

**Anticipated Outcome**: Please write a lay-oriented description of what you expect to learn/discover. Approximately 75 words.

At the end of these safety studies, we should be able to initiate a clinical trial for gene-modified stem cell transplantation for cystinsosis.