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**Leading Cystinosis Researchers Meet In Irvine, Calif., For Second  
CRF International Symposium On Rare, Deadly Disease**

More than 60 scientists and researchers from seven countries met at the Cystinosis Research Foundation's second International Cystinosis Research Symposium to exchange ideas about potential treatments, which include gene therapy stem cell investigations – currently the leading hope for a cure for the rare and fatal metabolic disorder.

It was announced at the symposium that a group of leading researchers have formed the CRF Cystinosis Gene Therapy Consortium to advance progress on the most promising current findings, including moving novel therapeutic modalities into human patients as quickly as possible. Work is now under way to develop the necessary preclinical animal model data and translate these results into an FDA-approved clinical trial.

A cystinosis patient's body is unable to rid itself of the amino acid cystine, which builds up and develops crystals that cause early cell death. There is a drug that can prolong the patient's life, but there is no cure. There are about 2,000 persons, mostly children, with cystinosis worldwide. Almost all sufferers succumb before 40 years old from cystinosis-related complications.

The April 8-9 symposium was underwritten by the CRF, which is the leading source of grant funds for cystinosis research. It was held at the Arnold and Mabel Beckman Center of the National Academies of Sciences and Engineering, adjacent to the University of California, Irvine.

Pre-clinical investigation by consortium members at The Scripps Research Institute in La Jolla, Calif., has resulted in the significant decrease of cystine in all tissues as well as the prevention or treatment of tissue injury in laboratory mice, a result recently published in the journal "Blood."

Work is under way to develop the necessary preclinical animal model data and translate these results into an FDA-approved clinical trial. The consortium includes well-known researchers in stem cell and gene therapies, molecular biology and clinical pathophysiology.

“We are dedicated to bringing the first stem cell and gene therapy clinical trial for cystinosis to reality. We hope that, if all goes well, there will be a clinical trial for a cure within the next three to four years,” said consortium member Nancy Stack, a co-founder and trustee of the CRF.

Stack added that the initial published findings also have “far-reaching implications for application to other diseases with systemic defects similar to cystinosis.”

Principal Investigator and consortium member Dr. Stephanie Cherqui explained, “Gene therapy adds a functional copy of the faulty gene and delivers it to the appropriate cells of the body. In the case of cystinosis, most of the tissues are damaged because of the lack of the *CTNS* gene. In other human disorders, a person’s own stem cells have already been used safely, and the stem cells could target several tissues. For cystinosis patients, this strategy might create a reservoir of healthy stem cells in the bone marrow for the lifetime of the patient that might respond to the progressive tissue damage of cystinosis and travel to repair the different organs of the patient.”

The symposium was the largest gathering of scientists in this rare-disease field. All of the researchers attending the symposium have been conducting bench and clinical research on cystinosis for several years. The symposium provides them a unique opportunity to meet and discuss their research progress. It fosters collegiality and cooperation which will advance treatments and the search for a cure.

In patients with the disease, cystine accumulates in the tissue and over time destroys various organs including the kidneys, muscles, eyes and central nervous system. Other complications include muscle wasting and difficulty swallowing.

Investigators from the United States, Canada, Belgium, France, Germany, Italy and The Netherlands, all of whom are working with CRF grants, delivered 30 presentations on various aspects of cystinosis research. The two-day event opened with a keynote address by the symposium’s chairman, Dr. Jerry Schneider of the University of California, San Diego, and concluded with remarks by Dr. William Gahl, clinical director of the National Human Genome Research Institute, part of the National Institute of Health. The first symposium was held in 2008.

Since its formation in 2003, the CRF has funded more than \$10.8 million in cystinosis research. Twice a year the CRF puts out a worldwide call to the scientific community for research proposals. Currently, the CRF is funding 31 studies and 10 research fellows in North America and Europe.

Also attending were officials from sponsors Sigma Tau Pharmaceuticals and Raptor Pharmaceuticals Inc. Raptor is scheduled to begin Phase Three trials on slow-release DR Cysteamine, a treatment that was developed with CRF grants awarded to Drs. Schneider and Ranjan Dohil of University of California, San Diego.

Development of DR Cysteamine represents the first treatment breakthrough in 20 years. It cuts the patient's dosing frequency of the medication from six hours to 12 hours. As a result of taking the medication less frequently, compliance improves, side effects are reduced and most importantly, children are able to sleep through the night.

The DR Cysteamine also has demonstrated potential in clinical studies as a treatment for other metabolic and neurodegenerative diseases, including Huntington's Disease, Batten Disease and NASH fatty liver disease.

Stack founded the CRF with her husband, Geoffrey, a managing director of the SARES•REGIS Group, a diversified real estate company in Irvine. The Stacks' daughter Natalie, 19, was diagnosed with cystinosis as an infant.

The Cystinosis Research Foundation is dedicated to finding better treatments to improve the quality of life for those with cystinosis and to finding a cure for this devastating disease. The CRF raises funds to support bench and clinical research aimed at improved treatments and a cure for cystinosis. The CRF also seeks to educate the medical and public communities about cystinosis to ensure early diagnosis and proper treatment.

For more information, call the Cystinosis Research Foundation at 949-223-7610 or visit [www.cystinosisresearch.org](http://www.cystinosisresearch.org).

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