Gene Therapy Researchers Form Consortium to Bring Cystinosis Findings to Clinical Trials; Disease Reversed in Laboratory Tests

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IRVINE, CA -- (Marketwire) -- 05/17/10 -- A group of leading researchers in stem cell and gene therapies, molecular biology and clinical pathophysiology have formed the CRF Cystinosis Gene Therapy Consortium to find successful therapies for cystinosis, a rare and fatal metabolic disorder that afflicts about 2,000 persons, mostly children, worldwide.

Pre-clinical investigation by members of the consortium at The Scripps Research Institute, 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." The goal of the CRF Cystinosis Gene Therapy Consortium is 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 at The Scripps Research Institute to develop the necessary preclinical animal model data and translate these results into an FDA-approved clinical trial.

The studies are being funded privately by the Cystinosis Research Foundation (CRF). "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. She added that the initial published findings also have "far-reaching implications for application to other diseases with systemic defects similar to cystinosis."

Principal Investigator 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."

In patients with cystinosis the amino acid cystine accumulates in the tissue due to the inability of the body to transport cystine out of one of the compartments of the cell. Over time, cystine destroys various organs including the kidneys, muscles, eyes and central nervous system. Other complications include muscle wasting and difficulty swallowing. Most cystinosis sufferers succumb to the disease or its complications by age 40.

This new consortium was created based on the promising pre-clinical studies performed in the mouse model for cystinosis by Dr. Cherqui's laboratory team. Transplantation of healthy bone marrow stem cells led to a significant decrease of cystine in all the tissues tested as well as prevention or correction of tissue injury. According to Dr. Cherqui, "The critical next step is to develop the strategies necessary to successfully deliver the CTNS gene to patients' bone marrow stem cells using gene therapy. Finally, the translation of these studies to humans will require extensive safety studies as well as the expertise of well-established researchers and physicians to lead to the first clinical trial of stem cell and gene therapy for cystinosis."

At present, the consortium is comprised of eight members. At The Scripps Research Institute are Dr. Stephanie Cherqui, who established the proof of concept for bone marrow stem transplantation in the mouse model for cystinosis, and Dr. Daniel R. Salomon, who has many years of experience in transplantation, stem cells and the immunology of gene therapy including membership on safety monitoring boards for several ongoing gene therapy clinical trials and regulatory experience with the FDA.

At the University of California, Los Angeles, are Dr. Donald B. Kohn, who is internationally recognized as a leader in stem cell and gene therapy, a former President of the American Society of Gene and Cell Therapy and has several gene therapy clinical trials in progress, and Dr. Alan Ikeda, who

specializes in blood and marrow transplantation. Dr. Ikeda is the site principal investigator for multiple cooperative studies in bone marrow transplantation and collaborates in designing and serving as a clinical investigator for the clinical trials involving gene and cell therapy.

Other consortium members are Dr. Corinne Antignac at Inserm U983, Paris, a specialist in the molecular pathophysiology of cystinosis; Dr. William Gahl of the National Institutes of Health and Dr. Jerry Schneider of the University of California, San Diego, who are world authorities on the diagnosis, therapy and clinical manifestations of cystinosis; and Nancy Stack, a director of the CRF and the mother of an affected child.

Since its formation in 2003, the CRF has provided more than $10.8 million in funding for71 cystinosis research and fellowship grants. Twice a year the CRF announces worldwide calls to the scientific community for research proposals. Currently, the CRF is funding 41 studies including 10 research fellows in the United States, Canada, France, Italy, Belgium, Germany and The Netherlands.

Read more: http://www.andhranews.net/Business/2010/May/17-Gene-Therapy-Researchers-Form-Consortium-19069.asp#ixzz0q5m78JLm