



FOR IMMEDIATE RELEASE

Jan. 7, 2009

Cystinosis Research Foundation Awards \$1.75 Million

In Scientific Grants To Find A Cure For Deadly, Genetic Disease

The [Cystinosis Research Foundation of Irvine](#), Calif., announced it awarded \$1.75 million in 11 new grants to researchers in the United States, Canada, Belgium and Italy who are working to find a cure for cystinosis, a rare, deadly metabolic and genetic disease that afflicts about 500 children and young adults in the U.S. and 2,000 worldwide.

“This brings the amount of funded research in 2009 to \$3.3 million, the largest one-year award total ever. With more than \$10.8 million in grants for [cystinosis research](#) issued by the CRF since 2003, our foundation is the leading funding source for bench and clinical investigations worldwide. Every day, CRF-funded researchers around the world get closer to unlocking the mysteries of this terrible disease. We believe we will find a cure in five to 10 years,” said Nancy Stack, CRF president.

The largest grant in this latest round of funding was \$308,602. It was awarded to Dr. Paul Goodyer, M.D., at the Montreal Children’s Hospital in Quebec and Dr. Francesco Emma, M.D., at the Bambino Gesù Children’s Hospital in Rome for a two-year study on “Stem Cell Microvesicles Rescue Cystinosis in Vitro.”

Eight CRF grants went to researchers on the San Diego and San Francisco campuses of the University of California and the Scripps Research Institute in La Jolla, Calif. Researchers in Belgium were awarded grants for two separate studies.

Cystinosis is a metabolic disease that slowly destroys every organ in the body, including the liver, kidneys, eyes, muscles, thyroid and brain. There is a medicine that prolongs the children’s lives, but there is no cure. Almost all sufferers succumb before 40 years old.

In patients with cystinosis, the amino acid cystine accumulates in the tissue due to the inability of the body to transport cystine out of the cell. This causes development of crystals, resulting in early cell death.

“The advancements achieved so far are the result of the CRF’s focused efforts and targeted approaches to research. The CRF is funding the best and brightest scientists at world-renown institutions who are focused on better treatments and a cure for cystinosis. This strategy has produced a greater understanding of the disease and the first improved treatment now in clinical trials in more than 25 years,” Stack said.

In the spring 2009 round of funding, \$1.6 million in grants were issued to researchers in the United States and France.

The CRF currently is funding more than 30 research studies, including 10 research fellows worldwide. In 2008, the CRF funded 12 studies with grants totaling \$1,741,889. The CRF also has directed significant funds to stem-cell and gene-therapy research while supporting researchers whose work is translatable into new treatments.

The CRF has scheduled its second International Cystinosis Research Symposium for April 8-9, 2010 at the Arnold and Mabel Beckman Center of the National Academies of Engineering and Science adjacent to the campus at the University of California, Irvine, Calif. The first symposium was held in 2008. It facilitated the formation of collaborative relationships among its research teams. More than 60 cystinosis researchers from the United States and Europe attended the symposium.

Nancy Stack and her husband, Geoffrey, an owner of the SARES•REGIS Group, an Irvine real estate company, have a daughter, Natalie, 18, with cystinosis.

The CRF has raised more than \$12 million, all of which is committed for medical research. All grants are awarded based on evaluations by the CRF's Scientific Review Board, which is comprised of leading doctors and research scientists in the field.

The 11 newly funded research studies, totaling \$1,754,629 are:

Bruce Barshop, M.D., Ph.D., Principal Investigator

University of California at San Diego

“Improvement of Intracellular Cystine Measurement”

\$62,930 – 1 year study

Bruce Barshop, M.D., Ph.D.

University of California at San Diego

“Tandem Mass Spectrometer Support”

\$160,604 – 1.5 years

Bruce Barshop, M.D., Ph.D., Mentor

Ilya Gertsman, Ph.D., Research Fellow

University of California at San Diego

“Proteomic Based Identification of Cysteinylylated Proteins in Cystinotic Cells”

\$124,214 – 2 year fellowship

Stephanie Cherqui, Ph.D., Principal Investigator

The Scripps Research Institute, La Jolla, California

“Stem Cell and Gene Therapy for Cystinosis”

\$163,340 – 2 year study

Stephanie Cherqui, Ph.D., Mentor

Brian Yeagy, Ph.D., Research Fellow

The Scripps Research Institute, La Jolla, California

“Stem Cell and Gene Therapy for Cystinosis”

\$138,485 – 2 year fellowship

Pierre J. Courtoy, M.D., Ph.D., Principal Investigator

de Duve Institute, Brussels, Belgium

“Lessons from Cystinotic Mice: Vital Imaging of Protein Handling and Lysosomal Function, Reciprocal Interactions with Regulatory Kinases, and Regeneration Potential by Transdifferentiation”

\$236,000 – 2 year study

Ranjan Dohil, M.D., Principal Investigator

University of California at San Diego

“A Study to Evaluate Enteric-Coated Cysteamine Therapy in Patients with Cystinosis”

\$27,226 – 1 year study

Paul Goodyer, M.D., Principal Investigator

Montreal Children’s Hospital, Quebec, Canada

Francesco Emma, M.D., Co-Investigator

Bambino Gesù Children’s Hospital and Research Institute, Rome, Italy

“Stem Cell Microvesicles Rescue Cystinosis in Vitro”

\$308,602 – 2 year study

Elena Levtchenko, M.D., Ph.D., Principal Investigator

University Hospital Leuven, Belgium

Roos Masereeuw, Ph.D., and Lambertus van den Huevel, Ph.D., Co-Investigators

Radboud University, Nijmegen, The Netherlands

“Role of P-glycoprotein Expression and Function in Cystinotic Proximal Tubular Cells”

\$83,999 – 1 year study

Robert Mak, M.D., Ph.D., Principal Investigator

University of California at San Diego

“Energy Homeostasis and Muscle Wasting in Nephropathic Cystinosis”

\$150,000 – 2 year study

Holger Willenbring, M.D., Ph.D., Principal Investigator

University of California at San Francisco

“Pluripotent Stem Cells as a Source of Immunocompatible Renal Progenitor Cells for Therapy of Nephropathic Cystinosis”

\$138,625 – 1 year study

The Cystinosis Research Foundation is the largest non-profit provider of funds for cystinosis research in the world. For more information, call Zoe Solsby at (949) 223-7610 or visit www.cystinosisresearch.org.

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