Rare, Life-Threatening Childhood Disease is the Focus of CIRM's Most Recent Investment

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San Diego, CA – Cystinosis is a rare disease that usually strikes children before they are two years old and can lead to end stage kidney failure before their tenth birthday. Current treatments are limited, which is why the CIRM Board today approved $5.2 million for research that holds the possibility of a safe, effective, one-time life-long treatment.

Cystinosis is caused by a genetic mutation that allows an amino acid, cysteine, to build up in and damage the kidneys, eyes, liver, muscles, pancreas and brain of children and adults. There is a Food and Drug Administration (FDA)-approved therapy, cysteamine, but this only delays progression of the disease, has severe side effects and people taking it still require kidney transplants, and develop diabetes, neuromuscular disorders and hypothyroidism.

University of California, San Diego researcher Stephanie Cherqui, Ph.D. and her team think there is a better approach. Their goal is to take blood stem cells from people with cystinosis, genetically-modify them to remove the mutation that causes the disease, then return them to the patient. The hope is that the modified blood stem cells will create a new, healthy, blood system free of the disease.

The CIRM funding approved today will enable Cherqui to do the pre-clinical work needed to show the approach appears to be both safe and effective, paving the way for FDA approval to test this in people.

"Orphan diseases like cystinosis may not affect large numbers of people but they are no less deserving of our support," says C. Randal Mills, Ph.D., President and CEO of CIRM. "Current treatments are expensive and limited. We want to push beyond and help find a life-long treatment, that could prevent kidney failure and the need for kidney transplant. In this case, both the need and the science were compelling."

Immediately following the Board meeting CIRM also launched its Roadshow, a multi-city tour that is focused on raising awareness about funding opportunities. The Roadshow is targeting the leading stem cell researchers, in both academia and industry, and informing them about the new ways to get CIRM funding, and support, in advancing their research.

"Our goal is to create a pipeline of projects heading towards clinical trials," says Dr. Mills. "We want to remind researchers that they have access to unrivaled resources here in California. We are not just investing money, though we do anticipate making an additional $900 million to put to work in new programs, we are also offering support in helping researchers get approval for clinical trials as well as designing and running those trials."

Through its Accelerating and Translating Centers, CIRM is creating a system that provides expertise and support in areas that researchers may lack experience in, such as applying to the FDA for approval to start a new clinical trial.

"We want to do everything we possibly can to shorten the length of time it takes to move a promising therapy from the laboratory bench to the patient's bedside," says Dr. Mills. "We believe the Accelerating and Translating Centers will do that. And anytime you create a faster, more effective system it is better for everyone, particularly the patients."

About CIRM

At CIRM, we never forget that we were created by the people of California to accelerate stem cell treatments to patients with unmet medical needs, and act with a sense of urgency to succeed in that mission.

To meet this challenge, our team of highly trained and experienced professionals actively partners with both academia and industry in a hands-on, entrepreneurial environment to fast track the development of today's most promising stem cell technologies.

With $3 billion in funding and approximately 300 active stem cell programs in our portfolio, CIRM is the world's largest institution

dedicated to helping people by bringing the future of cellular medicine closer to reality.

For more information, go to www.cirm.ca.gov.
