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PROPIONIC ACIDEMIA FOUNDATION SUPPORTS RESEARCH EFFORTS AT UT SOUTHWESTERN

DALLAS – March 5, 2003 – More than 900 people from across the United States and the United Kingdom have contributed to a research fund established at UT Southwestern Medical Center at Dallas by the Propionic Acidemia Foundation to find a cure for propionic acidemia, a genetic disease that typically kills children before age 10.

One in every 50,000 children is born with propionic acidemia (PA), a genetic disorder that disrupts the metabolism of branched-chain amino acids. The work of Dr. Toru Miyazaki, associate professor in the Center for Immunology at UT Southwestern, will be supported by the fund, which to date totals nearly $90,000.

"Dr. Miyazaki is the only researcher in the country looking at viable gene therapy to treat and, hopefully, one day cure this disease," said Janice Stoebner Boecker of Austin, vice president-research for the Propionic Acidemia Foundation and mother of a 3-year-old girl with PA. "The main course of treatment up until now has been dietary modification, but that is not very effective as children often still develop serious complications such as heart problems, brain damage, other organ damage, etc."

Children with PA must restrict their protein intake; otherwise they build up propionic acid and other toxins in their bodies. PA patients are deficient in propionyl-CoA carboxylase (PCC), an enzyme responsible for utilizing certain amino acids found in protein. Newborns with the disease immediately have problems consuming milk, often resulting in dehydration and death.

Currently, only a few states require that newborns be screened for PA, and the disease is often misdiagnosed. Researchers say it is likely that hundreds of sudden infant death syndrome-attributed fatalities may be the result of PA or other metabolic disorders.

Dr. Kern Wildenthal, president of UT Southwestern, applauds the work of the PA Foundation. "We are grateful to the foundation and its members for their generous support of Dr. Miyazaki's innovative research. An important part of our role at UT Southwestern is to discover

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new ways to fight diseases such as propionic acidemia, with the ultimate goal being to find cures," he said.

The gene that causes propionic acidemia was identified about 15 years ago, Miyazaki said. Most research since has focused on gene-mutation analysis. Miyazaki's work, however, centers on finding new therapies for the disease through construction of a mutant mouse model with PA. Studying mice with propionic acidemia allows researchers to observe gene manipulation in an animal with the disease. Once "corrected" genes have been transferred into mice, his research team can evaluate how they respond.

The Propionic Acidemia Foundation was established in 2002 by five families of PA patients. The foundation learned of UT Southwestern's research when one of its members saw an article by Miyazaki. Foundation leaders were so impressed with his work that they began collecting money to support his efforts.

During the past two years, Miyazaki has successfully cured PA-afflicted mice of most of the disease's symptoms. Treated mice are able to consume a normal high-protein diet, as well as grow, develop, procreate and have a normal lifespan. If his research continues to progress, Miyazaki sees the possibility of a cure for humans.

"Hopefully, we will finish the mice experiment in two to three years," he said. "Then we can think about human applications.

"I am very grateful to the Propionic Acidemia Foundation for helping me with my research efforts. This is a very difficult disease, and these parents really want to find better therapies and a cure for their children."

PA Foundation president Jill Franks of Highland Park, Ill., whose 3-year-old son has PA, said, "As parents, we recognize the importance and urgency of finding a cure for PA. We understand that economics play an important role in deciding which medical disorder gets researcher attention. We are doing our part to make sure that PA isn't neglected.

"And, we already see the benefits of our research funds beginning to pay off. We think that as Dr. Miyazaki is successful, more researchers will become interested in PA and our efforts will be able to fund their work."

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