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2011 Research Grants and Fellowship awarded by National PKU Alliance

Tomahawk, Wisconsin – January 28, 2011 – The National PKU Alliance (NPKUA) announced today its 2011 research grantees who are working to improve treatment options for people with phenylketonuria (PKU) and pursue a cure.

Roberto Gramignoli, D.Sc., will continue his NPKUA post-doctoral fellowship to explore the viability in using transplanted liver cells to cure PKU; Harvey L. Levy, M.D., will further explore the relationships between maternal Phe levels and outcomes on their children; Denise M. Ney, Ph.D., will continue studying the effect of dietary glycomacropeptide (GMP) on bone development, and Cary Harding, M.D. will be supported in his work on gene therapy to cure PKU. All of these research awards are made possible by the local fundraising efforts of the nineteen NPKUA member organizations around the country.

“The NPKUA Scientific Advisory Committee of the NPKUA evaluated and selected proposals that best continue to improve the understanding of this genetic disorder and develop new treatment options. Our ultimate goal is to accelerate the timeline for a cure,” said Christine Brown, Executive Director of the National PKU Alliance.

Postdoctoral Fellowship

Dr. Gramignoli is a Visiting Scholar in the Department of Pathology at the University of Pittsburgh School of Medicine, which is internationally renowned in the field of liver and hepatocyte transplantation. Hepatocyte transplantation has been successfully used to treat other metabolic disorders, but its efficacy in treating PKU had not yet been studied. During the last year, he has made significant progress in developing the mouse PKU model to test hepatocyte transplantation as a cure for PKU. Dr. Gramignoli and his team possess a level of commitment, excitement, and expertise that was evident in a successful site visit to the University of Pittsburgh lab last fall. Based on this visit, the NPKUA is pleased to support this post-doctoral fellowship award for a second year.

Research Grants

Dr. Levy, Professor of Pediatrics at Harvard Medical School and Senior Physician in Medicine/Genetics at Children’s Hospital Boston, will continue a study entitled “Maternal PKU: Offspring Follow-Up and Maternal Nutritional and Psychological Status.” This is the first study to examine post-natal influences as well as prenatal predictors of offspring outcome. Preliminary findings suggest that greater attention should be given to the pre- and post-natal environment of children born to mothers with PKU, as well as

to speech and language issues in these children. Ultimately, this study will change the standard of care and create a better understanding of outcomes in the children of PKU mothers. Dr. Levy's work is being funded through the Pepsi Refresh Project – a grant competition that the NPKUA won in August 2010 to support PKU research.

Dr. Ney, Professor in the Department of Nutritional Sciences at the University of Wisconsin-Madison, will continue to evaluate GMP, synthetic amino acids, and casein control diets on osteopenia in the PKU mouse model and wild type litter mates. Dr. Ney has already demonstrated that GMP diets lower the Phe in both blood and brain in the PKU mouse model. There have been observations documented that osteopenia does occur in PKU patients. Dr. Ney's hypothesis is that the GMP diet provides a source for more substantial systemic protein synthesis, a reduction in acid levels that would favor bone formation and more proline to support cartilage and scaffolding to enhance bone formation.

Dr. Harding, Associate Professor of Molecular and Medical Genetics at Oregon Health & Science University in Portland, Oregon, has been working on gene therapy to cure PKU in the PAH mouse models for more than 10 years with support from NIH. In order to translate this research into the clinic, vectors carrying the human PAH cDNA must be designed and carefully evaluated in a preclinical study. Specific gene therapy methods will be incorporated to improve the liver PAH expression from the human PAH cDNA and will be compared head-to-head in a short 8-week trial in PAH mice following portal vein injections and measuring the blood phe levels. A second specific aim is to use sequences from the human 28S ribosomal RNA gene (rDNA) in an effort to get permanent integration with the recipient genome so PAH expression will not diminish over time and thus could lead to a cure for PKU in humans.

NPKUA Research Selection Process

The overall funding strategy of the NPKUA is to support projects that will promote advances in the treatment and management of PKU, with the long-term goal of facilitating the development of a cure and to facilitate the growth and expansion of young, innovative researchers working in the inherited metabolic disease field. The NPKUA's Scientific Advisory Board is made up of eminently qualified physicians, researchers, and clinicians who are leaders in their fields to evaluate proposals, including Dr. Thomas Franklin, PhD; Dr. Emil Kakkis, MD, PhD; Dr. Harvey Levy, MD; Kathryn Moseley, MS, RD, Dr. Ray Stevens, PhD, and Dr. Bryan Hainline, MD. Each year this board goes through a rigorous evaluation process to select those proposals that will meet the above funding strategy.

“The NPKUA Scientific Advisory Board ensures that funds from the PKU community will go to support the most promising, rigorous research in the field,” said Scientific Advisory Board President, Dr. Tom Franklin. “These proposals have been thoroughly vetted by scientists and clinicians who are familiar with the current state of PKU research and recognize opportunities for advancement.”

Research grants and post-doctoral fellowships are awarded on an annual basis. Please visit www.npkua.org for more information.

About PKU

PKU, or phenylketonuria (pronounced *fen-il-Key-to-New-ree-uh*) is a lifelong genetic disorder in which a deficient enzyme prevents the body from metabolizing an essential amino acid, called Phenylalanine (Phe), which is found in most foods, including meat, bread, eggs, dairy, nuts, and some fruits and vegetables. When left untreated, PKU patients who consume too much Phe are at risk for severe neurological complications, including IQ loss, memory loss, concentration problems, mood disorders,

and in some cases, severe mental retardation. PKU affects approximately 13,000 people in the U.S. and 50,000 people worldwide.

To manage their disorder and avoid neurological complications, PKU patients must reduce their blood Phe levels by adhering to a Phe-restricted diet, which requires patients to monitor their daily intake. A Phe-restricted diet is supplemented with medical foods and specially produced low-protein foods and has to be maintained for a lifetime.

About the National PKU Alliance

Established in 2008, the National PKU Alliance is a coalition of the many local, state, and regional organizations that support PKU families. The National PKU Alliance is dedicated to improving the lives of individuals and families affected by PKU and pursue a cure. Visit www.npkua.org to learn more.

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