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A 2010 NPKUA Funded Research Project

RESEARCH GRANT SUMMARY
“MATERNAL PKU: OFFSPRING FOLLOW-UP AND
MATERNAL NUTRITIONAL AND PSYCHOLOGICAL STATUS”

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Medicine/Genetics at Children’s Hospital, Boston*

For women affected by PKU, it is essential for the health of their unborn child to maintain a low-phenylalanine (“phe”) diet before and during pregnancy. Though the developing fetus may only be a carrier of the PKU gene, the intrauterine environment can have very high levels of phe. The result is that children born to mothers with PKU are at risk for mental retardation, growth restriction and other health risks unless this diet is strictly followed by the mother during pregnancy. Dr. Levy’s study seeks to describe the health and well-being of women with maternal PKU and their children by examining and performing psychological assessments, providing a larger study on long-term outcome in maternal PKU. Forty children born to PKU mothers and 25 PKU mothers will be included in the study in the New England region.

PKU, once one of the most frequent causes of mental retardations, is now treatable thanks to the implementation of newborn screening. Those affected by PKU can follow a low-phe diet and avoid health issues. Not adhering to a low-phe diet, however, can result in emotional and neurophysical damage. And for women with PKU carrying a child, damage can occur to the child itself as well. Some women with PKU go off the low-phe diet during and post-pregnancy. The damages to the mother vary – including depression, anxiety, phobias, obsessive-compulsive behavior. For the children, behavioral deficits were also noted.

While correct diet therapy for PKU in pregnant women can prevent birth defects, Dr. Levy says a major clinical question remains: What is the long-term outcome in children from treated PKU pregnancies? An additional question is whether children from untreated PKU in the mother during pregnancy function normally. Dr. Levy’s study aims to describe the long-term medical and intellectual outcome in children from maternal PKU pregnancies; to describe the psychological, emotional, and social functioning in children from maternal PKU pregnancies; and to describe the medical and nutritional status and social and emotional functioning of PKU women. Secondary aims include determining which factors of child psychological status may be related to the mother following a low-phe diet during pregnancy to which factors may be environmental; and determining the relationship between previous and current measures of intelligence in children of maternal PKU. The overall goal of the study is to examine the health and well-being of women with maternal PKU and their babies. Both prenatal and postnatal environment factors will also be considered.

Methods for the study of mothers with maternal PKU will include medical and psychological examination, review of nutrition diary and biochemical studies; for the children, a physical, neurological assessment and a psychological assessment will be performed. Data will be analyzed based on characteristics of the mothers and children, including test scores, lab findings, nutrition intake, and background variables.