PHENYLKETONURIA AND PREGNANCY

Part II: Pregnancy Management

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Update A Fact Sheet
for Practitioners

What is Maternal PKU?

An elevated maternal level of phenylalanine that causes damage to a developing fetus is termed "maternal PKU." The term refers to the cause of the damage (high PHE levels in the mother), rather than to the resulting damage to the baby. Most often, the baby of a woman with PKU does not actually have PKU.

There is a 1.5:1 gradient of PHE across the placenta. Thus, the fetus is exposed to a higher concentration of PHE than that which exists in maternal blood. Because of this gradient, serum PHE levels that are safe for adults are harmful to the fetus. The higher the serum PHE concentration, the greater the degree of damage to the fetus.

Who is at increased risk for having a baby damaged because of elevated maternal PHE levels?

Any woman who has serum PHE levels greater than 6 mg/dl—even if she does not carry the diagnosis of PKU—is at risk.

Can Maternal PKU be treated to improve reproductive outcomes?

Yes. If maternal serum PHE levels are <4 mg/dl prior to conception and during pregnancy, the risk to the fetus is decreased. The risk of damage to the fetus cannot be entirely eliminated by treatment during pregnancy. Optimum management of PKU for a pregnant woman requires huge emotional, physical and financial commitments by the woman and is extremely labor-intensive for her health care providers. Optimum management begins months before conception. It is important for any woman with PKU who is contemplating pregnancy to understand that optimum management will lower the chance that her baby will be born with damage because of her PHE levels, but will not assure that her baby will be healthy.

Abbreviations: PHE - phenylalanine, PKU - phenylketonuria KU.

What Is A Reasonable Pregnancy Management Protocol for Women with PKU?

All pregnancies should be carefully planned. Women with PKU who are considering pregnancy should be referred to a regional genetics center. Pregnancy management should be a joint effort of the woman and her support system, health care providers, health agencies, and the metabolic team. Nutritional management is the key to pregnancy treatment for women with PKU. "Safe" maternal serum PHE levels cannot be achieved without the use of medical food (formula), and formula is essential to healthy maternal weight gain. The formula limits PHE intake, but still provides enough PHE for fetal growth and development. Formula should provide 80-90% of protein and energy intake.

Step 1: Pre-Pregnancy Counseling

The woman with PKU and her partner should participate in pre-pregnancy counseling with an experienced genetic counselor. They should understand the risks involved with the pregnancy, and the treatment. Even with the best dietary control, normal pregnancy outcome cannot be guaranteed. Elevated maternal serum PHE levels may cause spontaneous abortion, microcephaly, congenita cardiac anomalies, poor intrauterine growth, and mental retardation. Serum PHE levels at conception are critical to the organ development of the fetus. Effective treatment requires extraordinary commitment from the woman with PKU and a strong relationship with a team of professionals who are familiar with the treatment of high risk, PKU pregnancies. The team should include an experienced obstetrician and nutritionist.

Issues to be discussed:

- risks of pregnancy
- commitment required for treatment
- financing of treatment and formula

Step 2: Plasma Phe Levels to <6 mg/dl

If the woman is planning to become pregnant, she should bring her plasma PHE levels to <6 mg/dl; this may take several weeks. She must maintain blood levels in this range for several weeks to understand the effort required and to establish new patterns of formula intake, meals and cooking.

Issues to be discussed:

- effort required to lower blood PHE levels
- effects of treatment on other family members

Step 3: Plasma PHE Levels to 1-4 mg/dl

If the couple is still considering pregnancy, plasma PHE levels must be lowered to 1-4 mg/dl. Levels should be maintained in this range for 2-3 months to establish new patterns of food choices and meals, and increased formula intake.

Issue to be discussed:

- effort required to lower and maintain appropriate blood PHE levels

Step 4: More Pre-Pregnancy Counseling

The couple should meet again with the genetic counselor. At this point they need to be sure that they understand the risks of maternal PKU and pregnancy and are prepared for the rigors of management of a high-risk pregnancy. They also need to be sure that the financial arrangements are made for formula, and medical and nutritional monitoring. The couple should be counseled that as for any other couple, conception may take months or even years.

Issues to be discussed:

- pregnancy risks, efforts required to maintain blood PHE levels
- financial arrangements
- fertility issues
- increased risk for birth defects
- screening, testing methods during pregnancy

Step 5: Continuous Monitoring During Pregnancy

Monitoring a high-risk pregnancy requires weekly plasma PHE levels and nutrition consultations, monthly amino acid profiles, careful record keeping of food and formula intake, monitoring of weight gain and overall nourishment. Ultrasound assessments of fetal

growth and measurement of other standard biochemical parameters of pregnancy are also required. The physiologic changes of pregnancy may pose difficulties for effective dietary management, especially nausea and vomiting associated with early pregnancy.

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Resources

National PKU News Webpage – *http://www.pkunews.org*

University of Washington PKU Program – *http://depts.washington.edu/pku/*

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