



## Enrollment Procedures

1. Eligibility and Informed Consent
2. Treatment Plan with a Resource Mother
3. Treatment Plan without a Resource Mother
4. Randomization and Assignment of a Resource Mother

## **ELIGIBILITY AND INFORMED CONSENT**

### **ELIGIBILITY**

Any woman with phenylketonuria (PKU) who states she is planning to become pregnant or is already pregnant. Her blood phenylalanine level (when not treated) must be >6 mg/dl.

By agreeing to participate in the study and signing the informed consent, the woman agrees to being randomly assigned to the treatment group or the control group.

### **INFORMED CONSENT**

The informed consent for each institution must be read to the potential enrollee. A copy of the signed informed consent should be sent to the coordinating center.

A summary of what participation in the study involves (both with and without a Resource Mother) is included in your packet.

### **QUESTIONS/ANSWERS**

1. Does the Resource Mother have to come to my home?

The RM can usually help the most if she visits your home, where questions about food are easily addressed and cooking demonstrations can easily take place. We would prefer that a RM visit your home. However, if you do not wish the RM to come to your home, you can meet at another location (a park, library, restaurant, etc). Often, the RM will be in touch via phone or email. Visits usually last about an hour.

2. Why do I need an IQ test?

First, it will tell us what your learning strengths are. This will help the clinical staff present information in the best possible manner for you to learn—there is a lot to learn when returning to the diet! Secondly, there are many people who think that how well you do on diet depends on your IQ. We do not think this is necessarily true. We believe there are other factors, such as how much support you have and your attitudes toward the diet, which are more important than IQ.

3. What if I don't like my Resource Mother?

We have every reason to believe that you will. The RM's are women who are interested in helping other women with PKU. However, if a problem arises, please speak to the study coordinator for your clinic.

## **TREATMENT PLAN FOR WOMEN ASSIGNED A RESOURCE MOTHER**

### **When diet is initiated:**

Metabolic Clinic Visit

Assessment of Nutrient Intake

Laboratory assessments including:

Amino acids, ferritin, CBC, Pre-albumin, Vitamin B12, RBC folate, Serum cholesterol

Evaluation of Support Systems (questionnaires)

Intelligence testing if none of record

### **Weekly throughout pregnancy:**

Blood phenylalanine (and in some cases, tyrosine)

Feedback from metabolic clinic

### **Twenty times during pregnancy:**

Home visitation by a Resource Mother – a mother who has a child with PKU herself, is familiar with the diet and has been trained to provide assistance to women on the diet

### **Monthly throughout pregnancy:**

Nutrient analysis of dietary intake

### **Each trimester: \***

\* This is a minimum number of clinic visits; many clinic protocols include more frequent visits

Metabolic Clinic Visit

Assessment of Nutrient Intake

Laboratory Assessment (same as when diet is initiated)

### **Obstetrician visits:**

The number of visits is determined by the obstetrician – usually once per month until the last month of pregnancy and then once per week

### **Ultrasounds:**

Two ultrasounds are recommended – one at about 6 weeks to determine how far along the pregnancy is, and the second between 18-20 weeks when the heart and other organs can be visualized

### **Other services:**

Referrals to community health and nursing programs will be made as needed

### **Formula and low protein food:**

Each state and clinic has its own method for coverage of formula and low protein food. Emergency funds are available, if all other means of coverage have been exhausted

### **Birth and After:**

Newborn Pediatric Exam

Developmental Testing at age 1 year

Referral to Early Intervention

## **TREATMENT PLAN FOR WOMEN NOT ASSIGNED A RESOURCE MOTHER**

### **When diet is initiated:**

Metabolic Clinic Visit

Assessment of Nutrient Intake

Laboratory assessments including:

Amino acids, ferritin, CBC, Pre-albumin, Vitamin B12, RBC folate, Serum cholesterol

Evaluation of Support Systems (questionnaires)

Intelligence testing if none of record

### **Weekly throughout pregnancy:**

Blood phenylalanine (and in some cases, tyrosine)

Feedback from metabolic clinic

### **Monthly throughout pregnancy:**

Nutrient analysis of dietary intake

### **Each trimester: \***

\* This is a minimum number of clinic visits; many clinic protocols include more frequent visits

Metabolic Clinic Visit

Assessment of Nutrient Intake

Laboratory Assessment (same as when diet is initiated)

### **Obstetrician visits:**

The number of visits is determined by the obstetrician – usually once per month until the last month of pregnancy and then once per week

### **Ultrasounds:**

Two ultrasounds are recommended – one at about 6 weeks to determine how far along the pregnancy is, and the second between 18-20 weeks when the heart and other organs can be visualized

### **Other services:**

Referrals to community health and nursing programs will be made as needed

### **Formula and low protein food:**

Each state and clinic has its own method for coverage of formula and low protein food  
Emergency funds are available, if all other means of coverage have been exhausted

### **Birth and After:**

Newborn Pediatric Exam

Developmental Testing at age 1 year

Referral to Early Intervention

## **RANDOMIZATION**

Once the informed consent has been signed, contact the study coordinator in Boston (Fran Rohr) and let her know whether the woman with PKU began the diet before or during pregnancy. The woman will then be assigned to the treatment group (Resource Mothers) or the control group (no Resource Mothers).

The study coordinator will usually be able to let you know the randomization within a day. If the study coordinator will be away for more than a day, she will email the contributing coordinators at each clinic and let them know whom to call in her absence.

### **Assignment of Resource Mothers**

If the woman with PKU is assigned a Resource Mother, the coordinator at the contributing center will determine who the Resource Mother will be. This is generally a matter of which Resource Mother is the closest geographically, though other factors, such as whether or not the Resource Mother has another case, may be considered.

The contributing center coordinator will provide information about the woman with PKU to the Resource Mother, including contact information, basic background information, and the diet that has been prescribed. The Case Information Form may be used for this purpose.

If the woman with PKU is pregnant at the time of enrollment, the Resource Mother should make the first visit within a week. If the woman with PKU is planning a pregnancy, the Resource Mother should make the first visit within 2-3 weeks, but should call the woman with PKU as soon as possible to establish contact.

Let the study coordinator know who the Resource mother will be so that Dianne Sullivan can be contacted. Dianne Sullivan will call the Resource Mother prior to the initial visit to offer support.

### **Frequency of RM Visits**

The RM should visit a maximum of 20 times during the pregnancy. A log for keeping track of visitation is included in this packet. If the woman with PKU is already pregnant, the visits will occur approximately every other week. If the woman with PKU needs more frequent help in the beginning of the pregnancy, more visits may be scheduled early in the pregnancy, and less frequent ones near the end.

If the woman with PKU is planning a pregnancy, the RM should visit initially one or two times, and then determine with the clinic coordinator how frequently subsequent visits are needed, keeping in mind that the model for this program dictates a maximum of 20 visits. There is much flexibility in this aspect of the study – some women need more help as they attempt to resume diet.