

Maternal PKU Treatment Plan

Introduction

Maternal PKU is a problem that threatens to reverse the benefits of over 25 years of newborn screening for phenylketonuria. Women with PKU who are not in strict metabolic control prior to and throughout pregnancy are at high risk of having a child with microcephaly, mental retardation, congenital heart disease and low birth weight. Metabolic control depends on strict adherence to a low phenylalanine diet which requires ingestion of a special metabolic formula, avoidance of high protein foods and careful monitoring of food intake and blood levels. Under the best of circumstances the treatment for maternal PKU is very difficult; in situations complicated by psychosocial problems, it becomes an even greater challenge.

This care plan is a comprehensive intervention strategy which was developed by Children's Hospital PKU Program in conjunction with the New England Maternal PKU Planning Group, who have met regularly to consider the problems faced by women with PKU of childbearing age and to develop innovative, holistic approaches to dealing with the dilemma. Involvement in the Maternal PKU Collaborative Study, the PKU CORPS, and the Study of Psychosocial Factors in Maternal PKU has led to the development of a treatment plan which encompasses medical, nutritional and psychosocial aspects of treatment. The goal is to provide consistent, comprehensive assessment and treatment of maternal PKU in order to minimize fetal damage in an efficient, cost-effective manner.

This treatment plan is intended to be used for women who have blood phenylalanine levels over 6 mg/dl ($>360 \mu\text{mol/L}$). If there is uncertainty about whether or not treatment is indicated, refer to guidelines in Appendix 2 - Part 3.

This treatment plan can be used for women with PKU who are planning a pregnancy or already pregnant. ***If the woman with PKU is pregnant when she comes to medical attention, there is urgency in having her evaluated by a metabolic clinic and beginning the diet as soon as possible in order to protect the developing fetus.*** In such cases, the initial assessment, development of a care plan and initial teaching (Treatment Plan sections A-C)) must occur in the initial visit. This is an enormous amount of material to cover, especially if the woman is of lower intelligence or emotionally overwhelmed by an unplanned pregnancy. Hospitalization may be warranted in order to get the phenylalanine level down as quickly as possible, allow adequate time for educating the woman about managing the diet, and coordinate necessary community-based services.

If the woman is planning a pregnancy, but not in the immediate future, the material may be covered over the course of several outpatient visits to the metabolic clinic.

Treatment Plan

A. Initial Assessment- Metabolic Clinic

1. Metabolic/Medical
 - a. PKU history
 - i. Method of ascertainment
 - ii. Previous treatment of PKU and blood phenylalanine levels
 - b. Health
 - i. History
 - ii. Physical exam
 - iii. Primary care provider/health care delivery system identified
 - c. Understanding of risks of maternal PKU and PKU in the offspring
2. Nutritional
 - a. Weight and height
 - i. Weight class before pregnancy (Nutrition Support Protocol; Appendix 2 - Part 4)
 - b. Diet history
 - i. Medical food use
 - ii. Phenylalanine intake and blood levels
 - iii. Usual eating patterns/ likes and dislikes
 - c. Nutritional status
 - i. Laboratory assessment (below)
 - ii. Nutrient analysis of usual intake
 - d. Knowledge of phenylalanine restricted diet and effects of high phenylalanine on pregnancy outcome
 - e. Independence in managing MPKU diet
 - i. Ability to count phenylalanine intake
 - ii. Cooking ability
3. Laboratory
 - a. Serum amino acids (phenylalanine and tyrosine needed within 4 hours if pregnancy is suspected or confirmed; otherwise within 24 hours)
 - b. Albumin
 - c. Ferritin
 - d. Erythrocyte folate
 - e. Vitamin B12
 - f. Cholesterol
 - g. Genotype (optional)
 - h. Quantitative pregnancy test (if pregnancy is suspected)

4. Social/Family
 - a. Socioeconomic assessment
 - i. Age
 - ii. Marital status
 - iii. Other children/pregnancies
 - iv. Education
 - v. Employment (current and history)
 - vi. Religion
 - b. Organizational skills
 - c. Support for maintaining diet and pregnancy
 - i. Spouse
 - ii. Family
 - iii. Community
 - d. System of payment for medical food and protein modified products
 - e. Home environment
 - i. Facilities for cooking
 - ii. Organization
5. Psychological
 - a. Reading and arithmetic ability
 - b. Learning disabilities/learning style
 - c. History of depression, anxiety or emotional problems

B. Care Plan

1. Nutritional
 - a. Diet prescription (Nutrition Support Protocol; Appendix 2 -Part 4)
2. Metabolic/Medical
 - a. System for monitoring and reporting blood phenylalanine
 - b. Schedule of assessments/metabolic visits
 - c. Communication with other care providers
 - i. Obstetrician/Gynecologist
 - ii. Primary Care Physician
3. Psychosocial
 - a. Counseling referrals
 - b. Recommendation for teaching approach based on psychological testing
 - c. Resources to pay for low protein food and formula
 - d. Coordination with health care delivery system (managed care)
4. Community
 - a. Obstetrician/Gynecologist identified and contacted regarding risks of maternal PKU and treatment protocol

C. Initial Teaching- Metabolic Clinic

1. Reproductive decision making
 - a. Recommendations for treating maternal PKU and outcome of treated pregnancies (Appendix 2 -Part 3)
 - b. Probability of offspring having PKU
 - i. Carrier testing of father
 - ii. Detection of PKU in newborn
 - c. Alternatives to childbearing
2. Nutritional instruction for women planning pregnancy
 - a. Review of PKU and diet rationale
 - b. Medical food
 - i. Importance in diet
 - ii. Determining which medical food to use
 - A. Taste
 - B. Volume
 - C. Availability
 - iii. Ways to take medical food
 - A. Beverage (recipes)
 - B. Capsules
 - C. Bars
 - iv. Paying for medical food
 - v. Ordering and obtaining medical food
 - c. Phenylalanine restriction
 - i. Determination of phenylalanine tolerance
 - ii. Methods of counting phenylalanine intake
 - d. Weight gain
 - e. Protein modified products
 - i. Importance for calories and variety
 - ii. How to obtain
 - iii. Paying for protein modified products
 - f. Monitoring blood phenylalanine
 - g. Menu planning
 - h. Grocery shopping
 - i. Cooking
 - j. Record keeping
3. Psychosocial
 - a. Organization and planning
 - b. Supportive measures
 - c. Mental health counseling referral if desired

D. Initial Visit- Community

1. Gynecological/Obstetric
 - a. If planning pregnancy
 - i. Gynecological exam
 - ii. Birth control to be used until metabolic control is achieved
 - iii. Education regarding fertility and pregnancy testing
 - b. If pregnant
 - i. Obstetric exam
 - ii. Ultrasound at 6 weeks or after to determine viability and dating of pregnancy

E. Follow-up care - Metabolic Clinic

1. Metabolic/Nutritional
 - a. When pregnancy is confirmed
 - i. Review of diet and adjustment of intake to meet needs of pregnancy
 - b. Weekly
 - i. Report phenylalanine and tyrosine level and adjust diet as needed
 - ii. Assess weight gain for first 4 weeks after diet is initiated and monthly thereafter if weight gain is appropriate
 - iii. Encouragement and support - phone call
 - c. Monthly
 - i. Assess nutrient adequacy of diet from food records
 - d. Once per trimester (12, 20, and 32 weeks)
 - i. Clinic visit
2. Psychosocial
 - a. Weekly
 - i. Phone call for support
 - ii. Care coordination
 - b. Once per trimester
 - i. Clinic visit
3. Laboratory
 - a. Weekly
 - i. Blood phenylalanine -quantitative until metabolic control is achieved
 - ii. Blood tyrosine until levels > 0.8 mg/dl, then monthly
 - b. Monthly
 - i. Blood tyrosine if in desired range, otherwise weekly
 - c. Per trimester (at 12, 20 and 32 weeks)
 - i. Albumin
 - ii. Serum amino acids
 - iii. Ferritin
 - iv. RBC folate, B12, or cholesterol if indicated by previous low blood levels or if low intake is noted by nutritionist

F. Follow-up care -Community Based

1. Obstetric
 - a. Office visit
 - i. monthly until 32 weeks
 - ii. biweekly until 36 weeks
 - iii. weekly until delivery
 - b. Ultrasound
 - i. Level 1 ultrasound at 6 weeks for viability and dating of pregnancy
 - ii. Referral for level 3 ultrasound at 18-20 weeks for visualization of the heart to rule out heart anomalies
 - c. Prenatal birthing class - complete by 36 weeks

H. Coordination of Care

1. Once pregnancy is confirmed - Team meeting with representation from the metabolic center, obstetric office, health care delivery system and pregnant woman and spouse
 - a. Roles of health care providers defined
 - b. Expectations of pregnant woman and spouse defined
 - i. Weekly blood sampling
 - ii. Daily food record
 - iii. Keeping appointments with metabolic center, obstetrician, home visitor
 - iv. Involvement of spouse
 - v. Degree of self-efficacy
 - c. Case coordinator identified

I . Contingencies

1. If blood phenylalanine level is >10 mg/dl for 2 weeks, or > 15 for 1 week, or if woman is not monitoring bloods, an additional metabolic visit is required.
 - a. Determine causes of high phenylalanine (Appendix 2 - Part 6)
2. If blood phenylalanine level is > 10 mg/dl for another week, or if woman continues not to monitor phenylalanine, refer to community resources including:
 - a. Visiting Nurse or Public Health Nurse
 - i. for monitoring blood, supporting diet, checking weight
 - b. MCH Network
 - i. for public health nutritionist, if available
 - ii. for case manager to identify and coordinate services
 - c. Home health aide
 1. for practical assistance with diet (cooking, making formula, meal planning)

- d. Mental health counselor
 - i. for emotional support
 - e. Respite care (other children)
 - i. to allow more time for woman to manage diet
3. If woman is pregnant and blood phenylalanine level remains >10 mg/dl for yet another week, hospitalize patient for metabolic control (Appendix 2 Part 7)