



MATERNAL HYPERPHENYLALANINEMIA

INTRODUCTION

This booklet was written for young women with Phenylketonuria (PKU), a metabolic condition, or hyperphenylalaninemia and their families and friends.

It is to help people learn about the problems of pregnancy which may face some women with PKU. These problems are called Maternal Hyperphe. Maternal Hyperphe will apply to all women with extra phenylalanine in their blood (those with classical PKU, moderate hyperphe, and mild hyperphe).

PKU and hyperphenylalaninemia are complicated conditions, unknown to most people. Therefore, we suggest you read this book along with a companion book *Phenylketonuria - PKU*, which offers general information about the disorders, the cause and treatment.

MATERNAL HYPERPHENYLALANINEMIA

- Another name for Hyperphenylalaninemia is hyperphe.
- Describes the special problem of pregnant women who have too much phenylalanine (PHE), an essential amino acid, in their blood.
- These women may have either Phenylketonuria (PKU), a metabolic condition, or other types of hyperphe.
- Birth defects are found more often in babies of mothers with PKU or hyperphe. Their babies may have low birth weight, unusually small heads, heart problems, and they may be mentally retarded.
- The chance that the baby will have these birth defects appears to be related to the amount of PHE in the mother's blood during pregnancy.
- Mothers with higher PHE levels are more likely to have babies with birth defects than mothers with lower PHE levels, especially during first trimester.
- It is recommended that a woman's PHE levels remain between **2-6 mg/dl** before and during pregnancy.



WOMEN WITH CLASSICAL PKU OR HYPERPHE

- Must continue to eat a low PHE diet and special formula, **before and during** pregnancy under medical supervision. Most insurances will pay for special formula.
- Need to contact their PKU Treatment Center for updated information before becoming pregnant.

CHILDREN OF WOMEN WITH CLASSICAL PKU OR HYPERPHE

- Can be exposed to their mother's high PHE levels during pregnancy.
- Can have serious birth defects, such as mental retardation, heart defects, and a small head.
- Most likely will not have PKU and will not be harmed by the mother's special formula during pregnancy.*
- Do not have to eat a special low PHE diet unless they have PKU.

*Chances are about one in 100 that the baby will have either PKU or hyperphe.

INDIVIDUALS (boys or girls) WITH CLASSICAL PKU OR HYPERPHE:

- Have high PHE levels because they have inherited two PKU genes from their parents. (See the booklet *Phenylketonuria-PKU*)
- Must eat a low PHE diet to prevent damage to the brain during childhood.

WHAT WOMEN WITH MATERNAL HYPERPHE CAN DO

- Doctors believe that lowering the blood PHE level of a woman with hyperphe will help to greatly decrease the chance that her unborn baby will have serious birth defects.
- Lowering blood PHE can be done by eating foods low in PHE. For women with PKU and some women with hyperphe, this can be done only by eating a low PHE diet and drinking low PHE medical food (formula) like that eaten by children with PKU and hyperphe.
- The dietitian at your PKU Treatment Center will prescribe a low PHE diet.



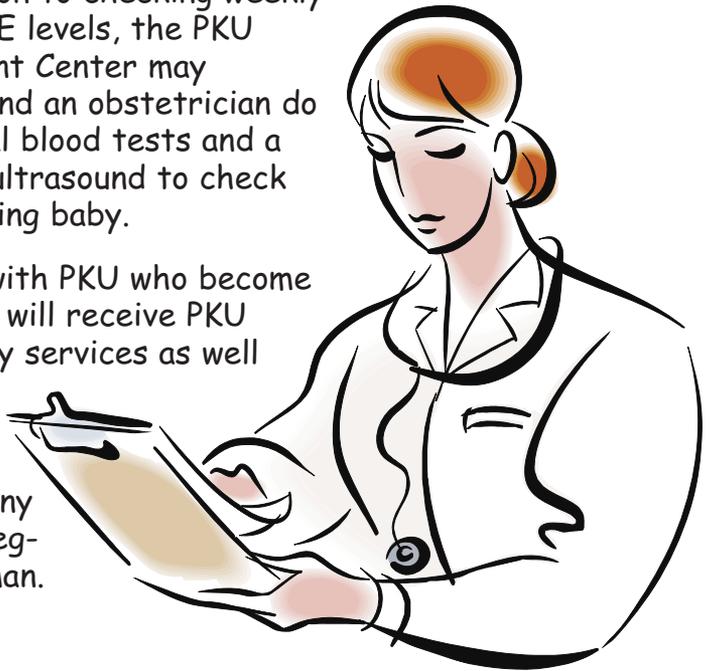
A baby's body parts, including its heart and brain, begin to grow in the first weeks of pregnancy before the woman even knows she is pregnant.

- The best way for a woman with high blood PHE to try and protect her unborn baby is to begin the low PHE diet **BEFORE** she becomes pregnant, and to continue the diet until the baby is born. That way the baby might be protected from the very beginning of its growth.

- Lowering the PHE levels during pregnancy reduces the chances of birth defects and particularly if this treatment is started before the mother becomes pregnant and if she receives special care during the pregnancy.

Make sure your obstetrician knows you have PKU. Most obstetricians (doctors who care for pregnant women) have never cared for a pregnant woman with PKU.

- PKU Treatment Centers will help a woman with PKU to find an obstetrician in her area who knows about her special problems.
- They can help plan a pregnancy to be the safest possible for mother and baby.
- In addition to checking weekly blood PHE levels, the PKU Treatment Center may recommend an obstetrician do additional blood tests and a level II ultrasound to check the growing baby.
- Women with PKU who become pregnant will receive PKU pregnancy services as well as the same prenatal care as any other pregnant woman.



CHOICES FOR WOMEN WITH PKU

- PKU Treatment Centers can help women with PKU learn more about all the choices of having children. The following are some choices:
- Some women decide they are well satisfied with their lives, husband, families, friends, school, job, community activities, hobbies, travel, etc. and choose not to have children.
- Adoption
- Knowing about the different kinds of birth control allow women to plan when or when not to have children.
- If you plan on becoming pregnant, call a PKU Treatment Center immediately to make plans for the safest possible pregnancy.
- After learning all the choices, women can choose the way that best satisfies themselves and their families.



IF A WOMAN WITH PKU WANTS TO BECOME PREGNANT

- Call the PKU Treatment Center **IMMEDIATELY** to make plans for the safest possible pregnancy.
- The risk of complications is greatly reduced when blood PHE levels are low during pregnancy for a woman with PKU.



If you have PKU and think you are pregnant, call a PKU Treatment Center **IMMEDIATELY** to start special pregnancy care right away.

PENNSYLVANIA PKU TREATMENT CENTERS

St. Christopher's Hospital for Children

Erie Avenue at Front Street
Philadelphia, PA 19134
(215) 427-5485

Milton S. Hershey Medical Center

Department of Pediatrics
500 University Drive
Hershey, PA 17033
(717) 531-8006

Children's Hospital of Pittsburgh

3705 Fifth Avenue
Pittsburgh, PA 15213
(412) 692-8631

Children's Hospital of Philadelphia

Clinical Metabolism
34th Street and Civic Center Boulevard
Philadelphia, PA 19104
(267) 426-5158
1-800-986-2229



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To connect children with special needs and their families
with a wide variety of services, call the Special Kids
Network Line at 1-800-986-4550.