



Maternal Hyperphenylalaninemia

Introduction

This booklet is for young women with either a metabolic condition called Phenylketonuria (PKU) or hyperphenylalaninemia. It is also for families and friends of women with such conditions.

This will inform you about the problems of pregnancy which may affect 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, their cause and treatment.

Maternal Hyperphenylalaninemia

- Another name for Hyperphenylalaninemia is hyperphe.
- This condition is the special problem of pregnant women who have too much phenylalanine (PHE), an essential amino acid, in their blood.
- Affected 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.
- 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.

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

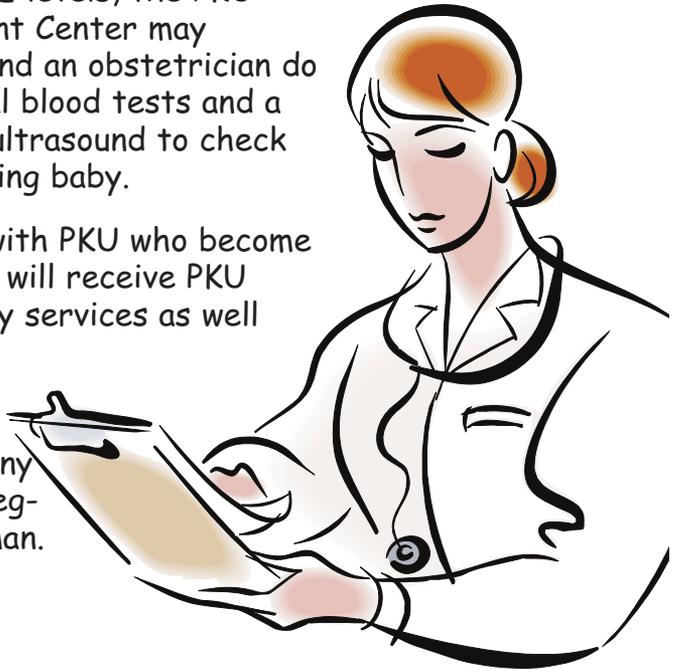
- Doctors believe that lowering the blood PHE level of a woman with hyperphe will help to greatly decrease the chance that her unborn baby will have serious defects.
- **A baby's body parts, including its heart and brain, begin to grow in the first weeks of pregnancy before the woman even knows she is pregnant.**
- Lowering blood PHE can be done by eating foods low in PHE. For women with hyperphe, this can be done by eating a low PHE diet, which includes low PHE formula. The dietitian at your PKU Treatment Center will prescribe a low PHE diet.
- The medication, Kuvan (Sapropterin), may be helpful during your pregnancy. Kuvan may or may not be effective in lowering blood PHE levels. Speak to your PKU treatment team about possibly using this drug during pregnancy. Kuvan is designated as a Pregnancy Category C drug. The safety of this drug is not completely known. This drug will only be prescribed for you if you and your treatment team decide that the benefits outweigh the potential risk to your fetus.
- The best way for a woman with high blood PHE to try and protect her 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. This is particularly so 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.
- Some choose adoption.
- Knowing about the different kinds of birth control allows 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.



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

4401 Penn Avenue
Pittsburgh, PA 15224
412-692-8631

Children's Hospital of Philadelphia

Clinical Metabolism
34th Street and Civic Center Boulevard
Philadelphia, PA 19104
267-426-5158
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.