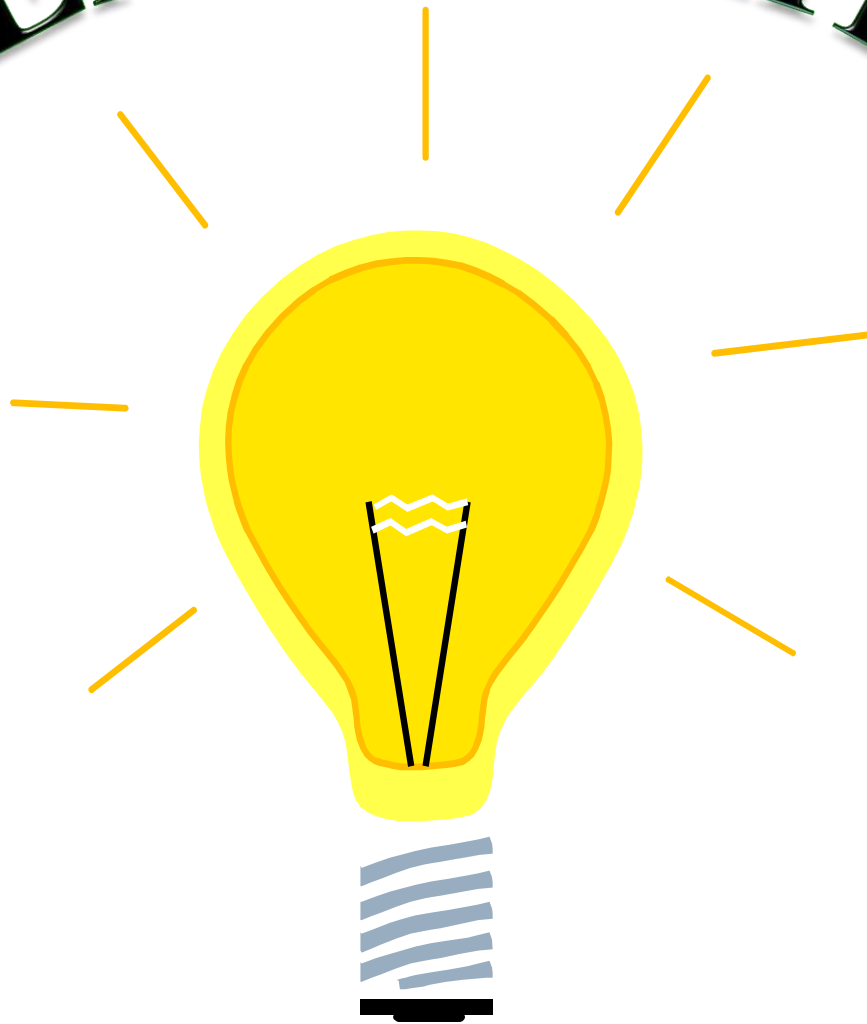


# EAT RIGHT



# STAY BRIGHT™

## Guide For Hyperphenylalaninemia

Laurie Bernstein, MS, RD, FADA

Cindy Freehauf, RN, CGC



The Children's Hospital

Affiliated with  
University of Colorado Denver

# **AUTHORS & CONTRIBUTORS**

## **Laurie Bernstein, MS, RD, FADA**

Fellow of the American Dietetic Association  
Assistant Professor- Department of Pediatrics  
Director, IMD Nutrition  
The Children's Hospital, Aurora CO

## **Cindy Freehauf, RN, CGC**

Assistant Professor- Department of Pediatrics  
Clinical Coordinator, IMD Clinic  
The Children's Hospital, Aurora CO

A special thank you to:

## **Kathleen M. Martin, BS, BA**

for her enthusiasm for learning and excellent graphic skills.  
Intern, IMD Clinic  
The Children's Hospital, Aurora CO

## **Second Edition Review Committee:**

### **Casey Burns, RD**

Metabolic Nutritionist  
The Children's Hospital, Aurora CO

### **Janet A. Thomas, MD**

Associate Professor, Pediatrics  
Director, IMD Clinic  
The Children's Hospital, Aurora CO

### **Sommer Myers, RD**

Metabolic Nutritionist  
The Children's Hospital, Aurora CO

### **Erica L. Wright, MS, CGC**

Certified Genetic Counselor  
The Children's Hospital, Aurora CO

### **Shannon L. Scrivner, MS, CGC**

Certified Genetic Counselor  
The Children's Hospital, Aurora CO

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# CHAPTER FOUR



## Maternal PKU

# INTRODUCTION

The Inherited Metabolic Clinic at The Children's Hospital in Aurora, CO serves the Rocky Mountain Plains Region and at least 130 individuals with hyperphenylalaninemia (PKU). Children and families require a great deal of complex information, most often new and alien to their experience, in order to establish and maintain consistent and effective treatment. Our experience with the process of sharing such information with families motivated us develop this anticipatory guidance book with teaching aids. We also found it useful to develop a checklist to be certain our delivery of service is consistent and thorough. We hope that this guide will prove to be a useful tool for you in your clinic.

THIS EDUCATIONAL TOOL IS DIVIDED INTO FOUR CHAPTERS:

1. Birth to Five Years
2. The Elementary School Years
3. Adolescent Years

## 4. Maternal PKU

EACH CHAPTER IS SUBDIVIDED INTO FOUR SECTIONS:

### **Clinic Encounter Check Lists**

Contains forms to be utilized during each clinic appointment in an effort to ensure that appropriate key issues are discussed at each clinic visit.

### **Experience and Thoughts**

We share insights from our experience. This section can be read independently, however, superscript items on the clinic encounter checklists refer to specific topics.

### **Teaching Aids and Handouts**

Find the materials designed to assist in counseling and teaching.

### **Resources**

Other useful and generally available teaching aids and information on acquiring those publications.

Keep in mind that all chapters have been developed as an anticipatory guidance tool with patient education and improved patient compliance as its main goal. We urge you to copy, individualize, and add to any and all of the sections. Whatever your approach, we hope this educational tool assists you in your clinic setting. New innovative methods are always helpful in our roles as health care providers.

This book has been developed with contributions from many professionals and students within The IMD clinic. There are some teaching aids that are available in one or more variations; we hope this complements your teaching style and facilitates the learning of new information.

# TABLE OF CONTENTS

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# MATERNAL PKU

Our final chapter has been designed for maternal PKU. It continues to reflect our multi-disciplinary approach to the treatment of hyperphenylalaninemia. Handouts have been adapted from previous chapters as well as teaching tools that have been designed specifically for maternal PKU. As professionals, maternal PKU is one of our greatest challenges.

It continues to be our goal to ensure that PKU related mental retardation and secondary complications are eradicated through early diagnosis and diet therapy.

# CHECKLIST: Maternal PKU

## General Maternal PKU Information

- Maternal PKU
  - Treated
  - Untreated
- Fetal development
  - Period of critical development of organs commonly affected by untreated maternal hyperphenylalaninemia (see handout)
  - Treatment range for maternal hyperphenylalaninemia (see handout)
- Emotional response to diagnosis of pregnancy<sup>1</sup>
  - Joy
  - Ambivalence
  - Fear
  - Denial
- Reproductive options<sup>2</sup>

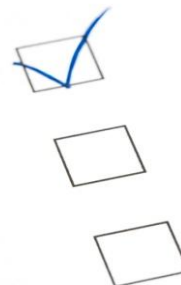
## Review of General Hyperphenylalaninemia Information<sup>3</sup>

- Biochemistry (see handout)
- Autosomal recessive disorder (see handouts)
  - Recurrence risk
  - Spousal carrier testing options
  - Prenatal diagnosis options
  - Newborn diagnostic testing
  - Reproductive options
- Principles of dietary management

## The Prescription (Rx)

- Principles of prescription
- Tyrosine supplementation
- Kuvan (see References)
- Gram scale—hands on demonstration with the individual
- Twenty-four hour clock—explain with use of diet records
- Formula preparation—measuring and mixing
- Diet records (see handout)

Superscript numbers throughout the Clinic Encounter Checklists refer to the Experience and Thoughts section.



# CHECKLIST: Maternal PKU

## Phenylalanine Levels, Interim History

- Interim phenylalanine levels
- Interim tyrosine levels
- Gestational age

## Daily Living Routine

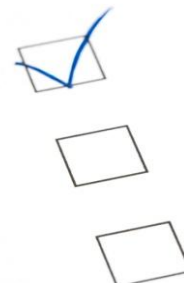
- Cooking/recipes
- Availability of low protein foods
- Diet records
- Blood draws
- Setting
  - Home
  - Work

## Daily Living Issues

- Adjusting to possible re-implementation of diet<sup>4</sup>
- Work
- Finances
- Impact of diet on family life

## Prenatal and Newborn Care<sup>5</sup>

- Team effort
- Ultrasound dating
- High risk pregnancy
- Newborn testing





# CHECKLIST: Maternal PKU

## Formula Coverage

- Each geographic region has its own laws regarding coverage of formula. When financial coverage of formula is not guaranteed via state law, clinic involvement might be necessary to facilitate coverage. Examples: Women Infant Children's Program (WIC), state's health department, solicitation to insurance companies of medical necessity. (see MEDICAL FOODS LETTER)

## The Clinic Routine

- Blood draws
  - Procedure
  - Frequency
- Laboratory results
  - Procedure
  - Frequency
- Diet records
  - Procedure
  - Frequency
- Weights
  - Procedure
  - Frequency
- Diet prescription changes
  - Procedure
  - Frequency
- Appointments
  - Frequency of visits
  - Flow at visits
- Clinic staffs' contact numbers
  - Routine
  - Emergency

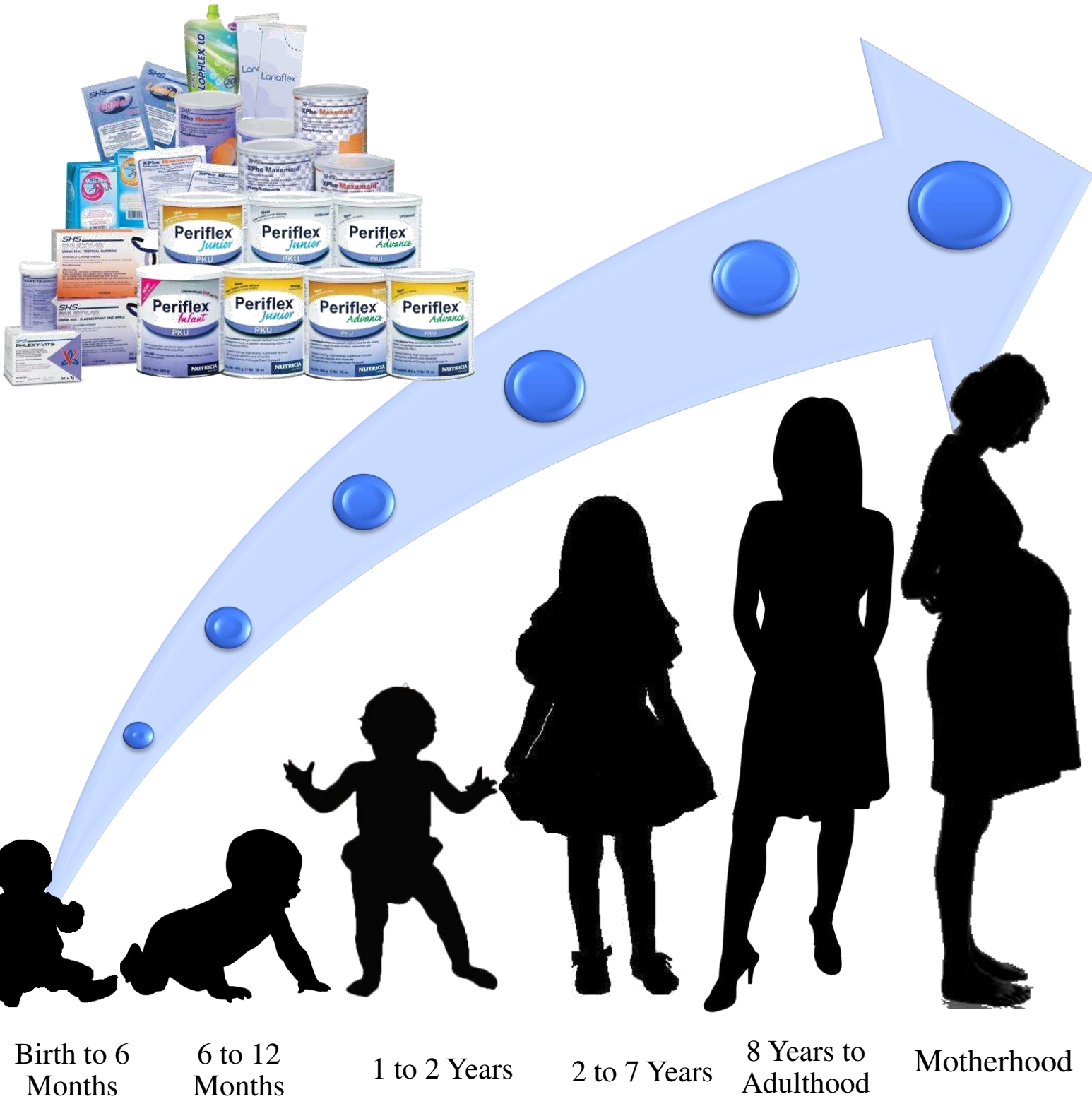


# EXPERIENCE & THOUGHTS

1. Dependent upon the circumstances, the emotional responses will vary from patient to patient and from time to time throughout the pregnancy.
2. Our team comes from both a metabolic and genetic prospective. With respect to genetic counseling, the approach is nondirective. Risks and possible outcomes are identified and discussed so the family may make an informed decision with respect to reproductive options.
3. It has been our experience that an adult woman who has maintained contact with our clinic is slightly more “savvy” with respect to the biochemistry and genetics of hyperphenylalaninemia. For these individuals, a brief overview may be all that is required. For those women whose clinic contact has been limited, an in-depth review may be required. Our multidisciplinary team allows us to address all of the issues listed in this section.
4. A patient who has been off diet can be a challenge. It is our approach to offer a variety of choices, including some that are nontraditional. The options that are now available to our patients include powders, bars, modular components and pills. The ultimate goal is to have levels within treatment range.  
It is important for the clinician to assess the cognitive level of the patient. If this is a patient who has been off diet and has returned to clinic because of her pregnancy there is a strong possibility that her desire for compliance will not be balanced with her ability to achieve compliance. Year of non-compliance and high levels will most likely have an effect on her learning curve. Modifications to teaching approaches and expectations may need to be made.
5. Depending on patient compliance and state screening methods, prenatal and newborn testing will vary. Care and interaction with prenatal and pediatric teams must be adjusted accordingly. Typically, we include articles on maternal PKU for the teams’ review.

# PRINCIPLES OF DIET PRESCRIPTION

## Throughout a Lifecycle



**At Every Age, Medical Food Is An Important Part Of Your Daily Nutrition!**

# PRINCIPLES OF DIET PRESCRIPTION

## PKU and Pregnancy

### Medical Food

*Circle One*

Lophlex

Lophlex LQ

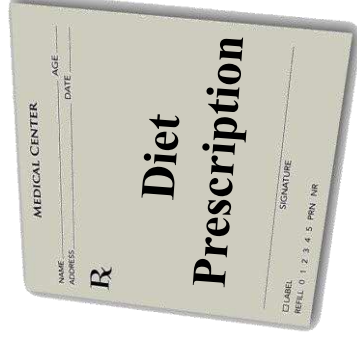
Periflex Advance

XPhe Maxamum

+



=



Phe, Amino Acids, Vitamins, Minerals & Calories

If a pregnant woman only eats what is allowed on a low phenylalanine diet without medical food, they and their baby may be deficient in protein, calories, essential vitamins and minerals. Medical food may provide most of the daily protein, essential vitamin and mineral requirements.

**See Reference:** “PKU & Pregnancy, For Women With PKU.” March of Dimes.

# PRINCIPLES OF DIET PRESCRIPTION

## PKU and Pregnancy

### Medical Food

*Circle One*

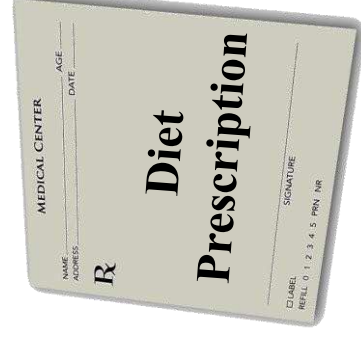
- Lophlex
- Lophlex LQ
- Periflex Advance
- XPhe Maxamum
- PhenylAde 40
- PhenylAde Drink Mix
- PKU Coolers
- PKU Express
- CaminoPro PKU 15
- Phenex-2
- Phenyl-Free 2



+



=



Phe, Amino Acids, Vitamins, Minerals & Calories

If a pregnant woman only eats what is allowed on a low phenylalanine diet without medical food, they and their baby may be deficient in protein, calories, essential vitamins and minerals. Medical food may provide most of the daily protein, essential vitamin and mineral requirements.

**See Reference:** “PKU & Pregnancy, For Women With PKU.” March of Dimes.



# DAILY DIET PRESCRIPTION

## PKU and Pregnancy



Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

Name: \_\_\_\_\_ DOB: \_\_\_\_\_

### Medical Food/Formula:

**Step 1:** Measure Medical Food/Formula:

\_\_\_\_\_ of \_\_\_\_\_  
 Amount Unit of Measure Medical Food

\_\_\_\_\_ of \_\_\_\_\_ Add to hand shaker.  
 Amount Unit of Measure Medical Food

**Step 2:** Add water to make a total volume of \_\_\_\_\_  
 Amount Unit of Measure

**Step 3:** Shake vigorously

**Step 4:** Refrigerate, complete within 24 hours.

### Regular and Low Protein Food:

\_\_\_\_\_

mg Phe gm Protein # Exchanges

*Circle One*

**Medical food may provide complete nutrition without any Phe.**  
 Solid and low protein foods provide additional amino acids, vitamins, minerals, and calories.

# 24 HOUR DIET DIARY

Name: \_\_\_\_\_

Dates Covered: \_\_\_\_\_

Date of Birth: \_\_\_\_\_

Age: \_\_\_\_\_

Weight: \_\_\_\_\_

Gestational Age: \_\_\_\_\_ (# of weeks since last period)

Levels: \_\_\_\_\_ Phe \_\_\_\_\_ Tyr

Medical Food/Formula	Amount

Kuvan:
Tyrosine:
Multivitamin:
Other:

Add water to make \_\_\_\_\_ ml ( \_\_\_\_\_ fl. oz.)

Phe or protein \_\_\_\_\_ mg or g

**Before obtaining a blood specimen, please record the food eaten for 3 consecutive days.**

Date	Time	Foods or Liquid Eaten	Amount Eaten	Phe (mg)	Energy (kcal)
<b>Totals</b>					

Appetite today was \_\_\_\_\_ better than usual \_\_\_\_\_ usual \_\_\_\_\_ poor

Felt ill today? \_\_\_\_\_yes \_\_\_\_\_no      Morning sickness? \_\_\_\_\_yes \_\_\_\_\_no

If ill, was medication required? \_\_\_\_\_yes \_\_\_\_\_no

If yes, name and amount of medication prescribed: \_\_\_\_\_

Regurgitation of food or formula? \_\_\_\_\_yes \_\_\_\_\_no      Diarrhea? \_\_\_\_\_yes \_\_\_\_\_no

# MEDICAL SERVICES LETTER

THIS IS A TEMPLATE TO AID YOU IN CREATING ONE THAT WORKS FOR YOUR INSTITUTION

RE:

DOB:

[Hospital]# : \_\_\_\_\_

To whom it may concern:

[Name] is a woman insured with you. We are writing at her request to tell you about medical services that will be needed before and during pregnancy.

[Name] has Hyperphenylalaninemia (PKU), which is an inherited metabolic disorder that results from a deficiency in the enzyme phenylalanine hydroxylase. Individuals treated early in life with a special diet low in phenylalanine have normal growth and development, whereas those untreated become mentally retarded and may have seizures and a variety of other problems. As a result of early treatment, women with PKU are now having children. This has led to what is known as Maternal PKU syndrome. Strict dietary treatment and monitoring before and during pregnancy is necessary to prevent detrimental effects to the fetus.

In a survey of maternal PKU, 92% of the non-phenylketonuric children of PKU mothers not on diet were mentally retarded, 73% were microcephalic, 40% had low birth weight, and 12% had congenital heart disease. These deleterious effects result from the high phenylalanine levels in maternal blood. The placenta actively transports these amino acids to levels in the fetal circulation that can be up to two times greater than in the maternal circulation.

A national collaborative study confirms the extent of the positive effects of dietary treatment on pregnancy outcome. There is strong evidence that dietary therapy instituted prior to conception and maintained throughout pregnancy results in a positive prognosis and a healthy baby. For this reason, we strongly recommend that [name] be placed on diet and carefully monitored before and during each pregnancy.

The diet will consist of a special formula and calculated amounts of food. Monitoring will consist of weekly serum phenylalanine and tyrosine levels, periodic additional blood work, and periodic ultrasounds, in addition to routine prenatal care.

The low phenylalanine diet requires a special formula (Medical Food) which provides all vitamins, minerals, energy and protein (without phenylalanine). Foods are given to meet a prescribed amount of phenylalanine intake per day. The majority of cost comes from the use of the special formula. The cost per day, depending on the formula needed to provide necessary nutrients, can range from \$8.80 - \$17.00 per day. Therefore, the cost per year would be between \$3200 - \$6200; which includes three months pre-conception management plus nine months pregnancy.

We appreciate your consideration of this situation. Please feel free to contact us at [phone number of metabolic clinic].

Sincerely,

[name of metabolic nutritionist]

[name of metabolic clinic director]



# MEDICAL FOODS LETTER

THIS IS A TEMPLATE TO AID YOU IN CREATING ONE THAT WORKS FOR YOUR INSTITUTION

RE: \_\_\_\_\_

BD : \_\_\_\_\_

TCH : \_\_\_\_\_

To Whom It May Concern:

This letter will address the use of special medical foods (also called metabolic formula) in Phenylketonuria (PKU).

PKU is an inherited enzymatic defect transmitted on an autosomal recessive basis. Affected individuals have difficulty in metabolism of phenylalanine, one of the essential amino acids. PKU is treated with a diet restricted in phenylalanine, designed to provide just enough phenylalanine for growth, development, and physiologic needs, while keeping blood phenylalanine levels in a narrow acceptable range. Individuals with PKU who are not placed on dietary treatment for this condition early in life will become mentally retarded with seizures and behavior disturbance. Dietary treatment has been shown to be very effective if it is instituted before one month of life, and maintained and monitored over time.

Indefinite continuation of dietary management is recommended to all patients with PKU. We also recommend reinstatement of dietary restriction to all patients whom diet was discontinued in childhood during the 1970's, when that was the usual practice. These recommendations are based on the growing body of evidence, including a report from the PKU Collaborative Study (Koch et al., *Journal of Pediatrics*, 1982; 100:870-875), indicating that there is a decline in average IQ and development of difficulties in school performance after diet discontinuation. There are also reports (e.g. Butler et al., *JIMD* 1987; 12:451-457) describing the development of neurologic abnormalities including paralysis in young adults who have gone off diet; in some patients, the neurologic damage was reversible when returned to diet control, but in some, the damage was permanent. Finally, adult women with PKU must have dietary control to prevent adverse effects in pregnancy. It has been known since the 1950's that PKU, uncontrolled in pregnancy, causes severe mental retardation and birth defects (severe heart and intestinal defects are most common) in the developing fetus. The Maternal PKU Collaborative Study demonstrated that perfect control of phenylalanine levels coupled with attention to the multiple problems of nutrition in pregnancy on an artificial diet may permit the birth of healthy children (Widaman KF et al *Pediatrics* 2003;112:1537-1543).

One of a number of proprietary formulas (see table) provides the primary protein constituent for the PKU dietary treatment regimen. Use of these medical foods is absolutely essential for the normal intellectual development of these patients and their ongoing neuropsychologic health. Patients who receive this formula must be under the care of a doctor and a metabolic nutritionist.

These metabolic formulas are used in combination with ordinary foods in restricted, monitored amounts. The patient's diet, growth and serum phenylalanine levels must be carefully monitored and adjusted as indicated.

# MEDICAL FOODS LETTER

These medical foods are an artificial replacement for the normal protein-containing foods that we all require for growth and cell replacement. The use of medical foods may cause growth retardation, malnutrition, and neurologic disease if not meticulously prescribed and carefully monitored. Inappropriate use or poor monitoring can result in malnutrition and irreversible brain damage. For this reason, medical foods should only be dispensed by prescription.

Medical foods fall into a special category based on an agreement between the FDA and the producers of these metabolic formulas. They do not fall strictly in the category of pharmaceuticals; however, they are not “food supplements.” Responsible pharmacists, despite the lack of laws preventing dispensation without prescription, will insist upon a prescription to document appropriate medical use of and monitoring of these medical foods.

We request that you approve coverage of medical foods for the management of PKU for [name]. We would prescribe the amount of specific medical food and of phenylalanine from natural foods; and monitor [name]’s clinical status using laboratory studies (phenylalanine levels and measures of nutritional adequacy to avoid iatrogenic deficiency of protein or other nutrients) and clinical evaluations. The medical food could be dispensed through [name of metabolic clinic] or any other pharmacy; laboratory studies would utilize [name of metabolic clinic laboratory] for phenylalanine levels and [name of metabolic clinic laboratory] or other laboratory for other needed surveillance tests. If you have any questions regarding any of this information, please contact us at [phone number of metabolic clinic].

Sincerely,

Laurie Bernstein MS, RD, FADA  
Assistant Professor- Department of Pediatrics  
Director- IMD Clinical Nutrition

Janet Thomas, MD  
Associate Professor- Department of Pediatrics  
Director- IMD Clinic

## Addendum: List of formulas used to manage maternal PKU

### **Nutricia:**

[www.nutricia.com](http://www.nutricia.com)

- Lophlex
- Lophlex LQ
- Periflex Advance
- XPhE Maxamum

### **Applied Nutrition:**

[www.medicalfood.com](http://www.medicalfood.com)

- PhenylAde 40
- PhenylAde Drink Mix

### **VitaFlo:**

[www.vitaflousa.com](http://www.vitaflousa.com)

- PKU Coolers
- PKU Express

### **Cambrook Foods:**

[www.cambrookefoods.com](http://www.cambrookefoods.com)

- CaminoPro PKU 15

### **Abbott Nutrition:**

[www.abbottnutrition.com](http://www.abbottnutrition.com)

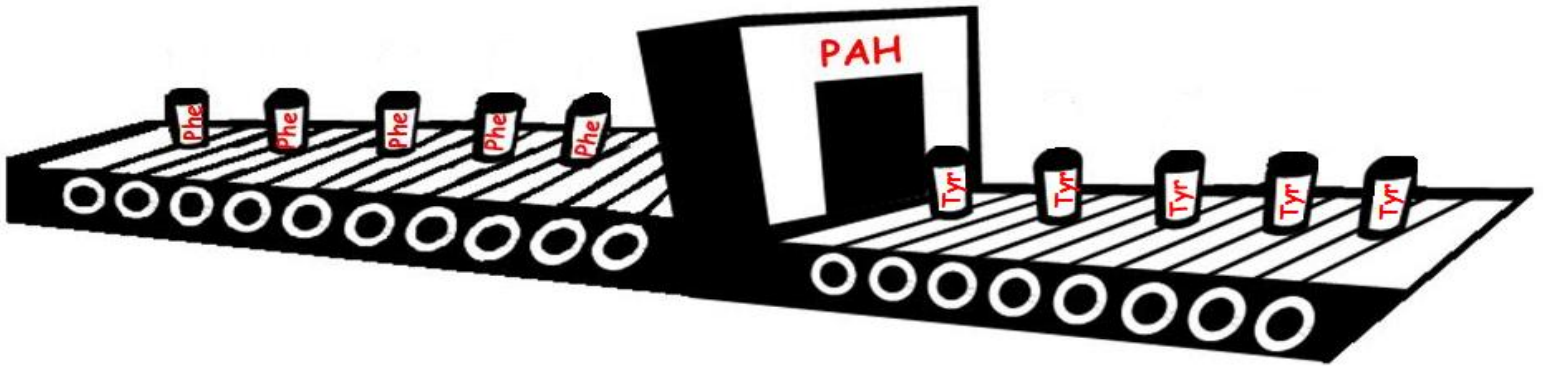
- Phenex-2
- Phenyl-Free 2

Adapted from *Medical Director’s Resource Guide to Metabolic Disorders*, Ross Laboratories

# Hyperphenylalaninemia

THE BODY IS SIMILAR TO A FACTORY...

When the body has enough **PAH** it converts **Phe** to **Tyr**.



If the body does not have enough **PAH**,  
the **Phe** is not converted to **Tyr**.



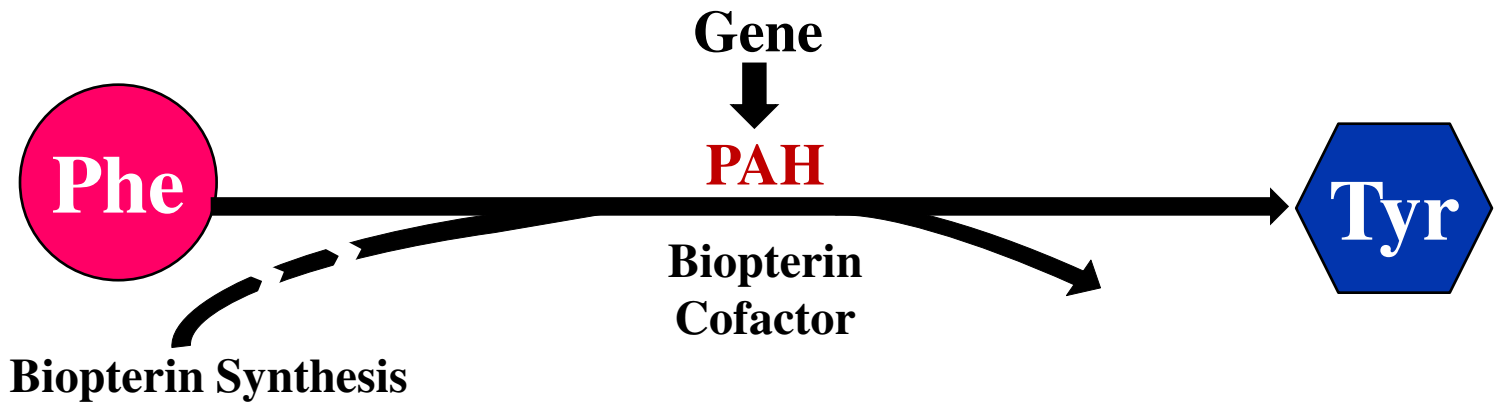
The Result: Too much **Phe** and not enough **Tyr**.

**PAH:** Phenylalanine Hydroxylase    **Phe:** Phenylalanine    **Tyr:** Tyrosine

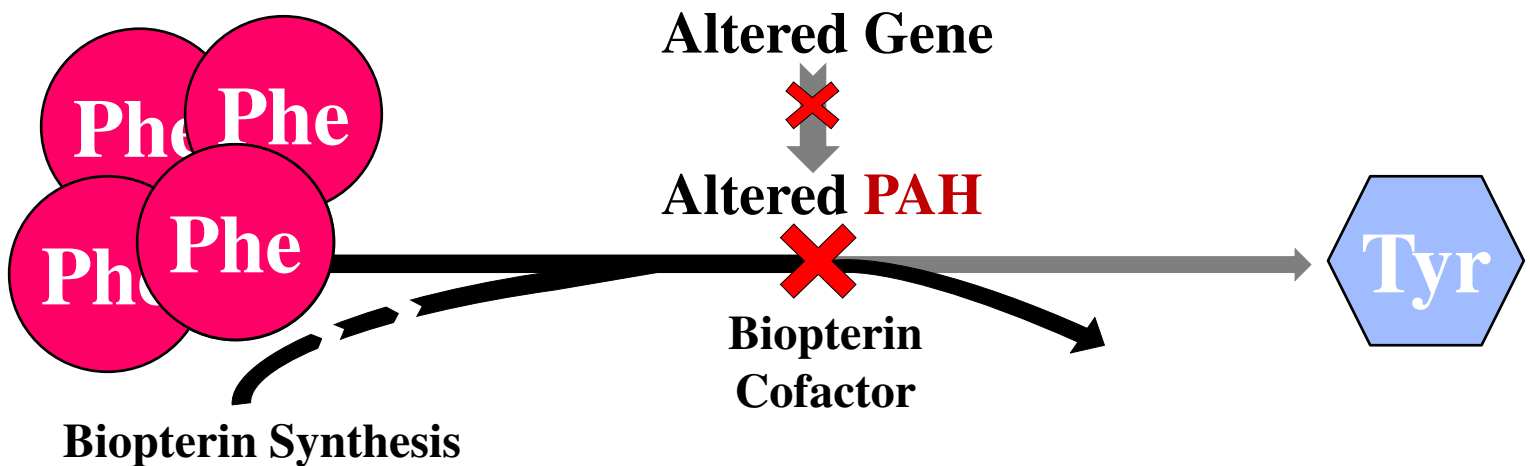
# Pathways

**Phe** = Phenylalanine    **PAH** = Phenylalanine Hydroxylase    **Tyr** = Tyrosine

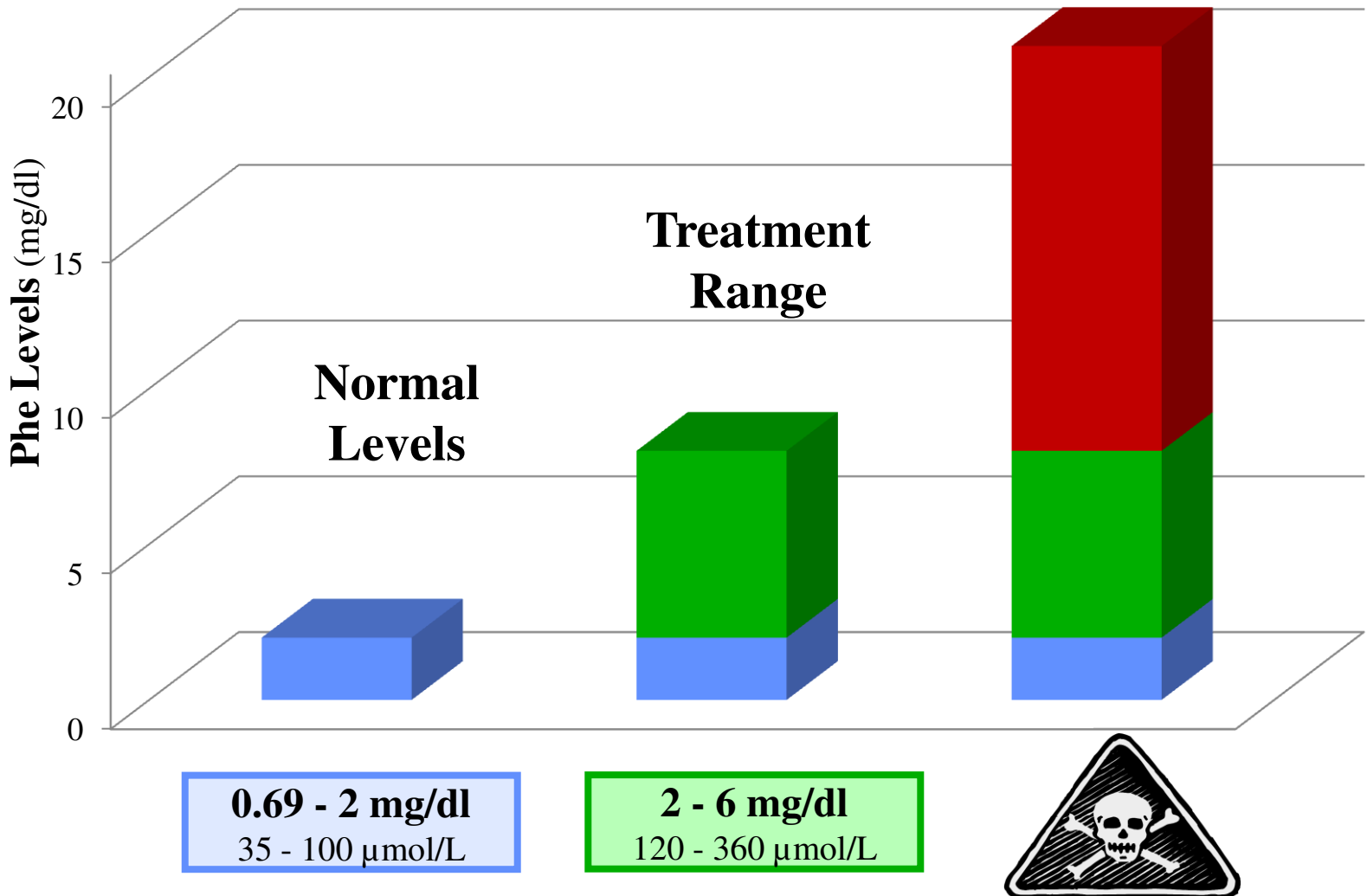
## Normal Phenylalanine Pathway



## Classic PKU Pathway



# Phenylalanine (Phe) Levels



## High Phe Levels:

When the level of phenylalanine is greater than the treatment range.

# Phenylalanine (Phe) Levels

## Treatment Range

120 - 360  $\mu\text{mol/L}$  (2 - 6 mg/dl)

Low	Normal	Non- PKU HPA	Mild PKU	Classic PKU
< 35 $\mu\text{mol/L}$ ( < 0.5 mg/dl )	35 - 120 $\mu\text{mol/L}$ ( 0.5 - 2 mg/dl )	120 - 600 $\mu\text{mol/L}$ ( 2 - 10 mg/dl )	600 - 1200 $\mu\text{mol/L}$ ( 10 -20 mg/dl )	> 1200 $\mu\text{mol/L}$ ( > 20 mg/dl )

## Hyperphenylalaninemia (HPA)

[-----hyper-----][-----phenylalanin-----][-----emia-----]  
*high levels of* → *phenylalanine* → *in the blood*

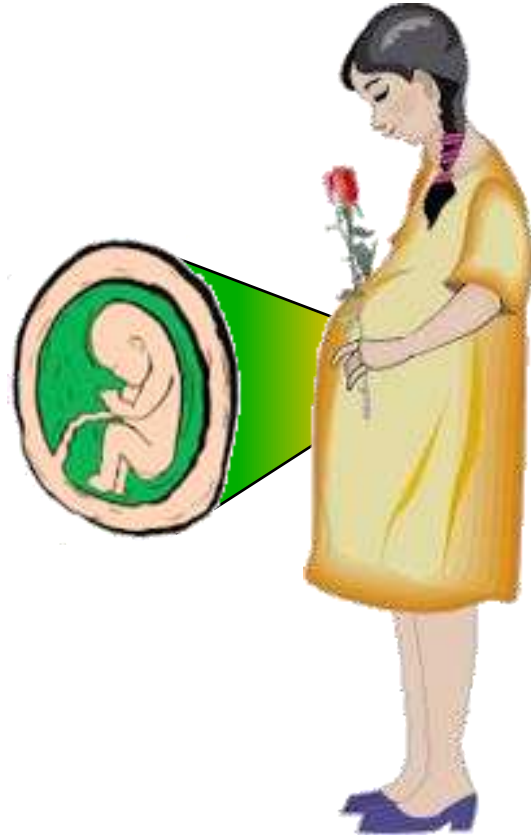
## Phenylketonuria (PKU)

[-----phenylketon-----][-----uria-----]  
*phenylketone* → *in the urine*

# PREGNANCY TREATMENT RANGE

**2.0-6.0 mg/dl**  
or  
**120 - 360  $\mu\text{mol/L}$**

Phenylalanine  
levels are within  
treatment range



**DANGER!!!**



Phenylalanine  
levels greater than

**6.0 mg/dl**

or

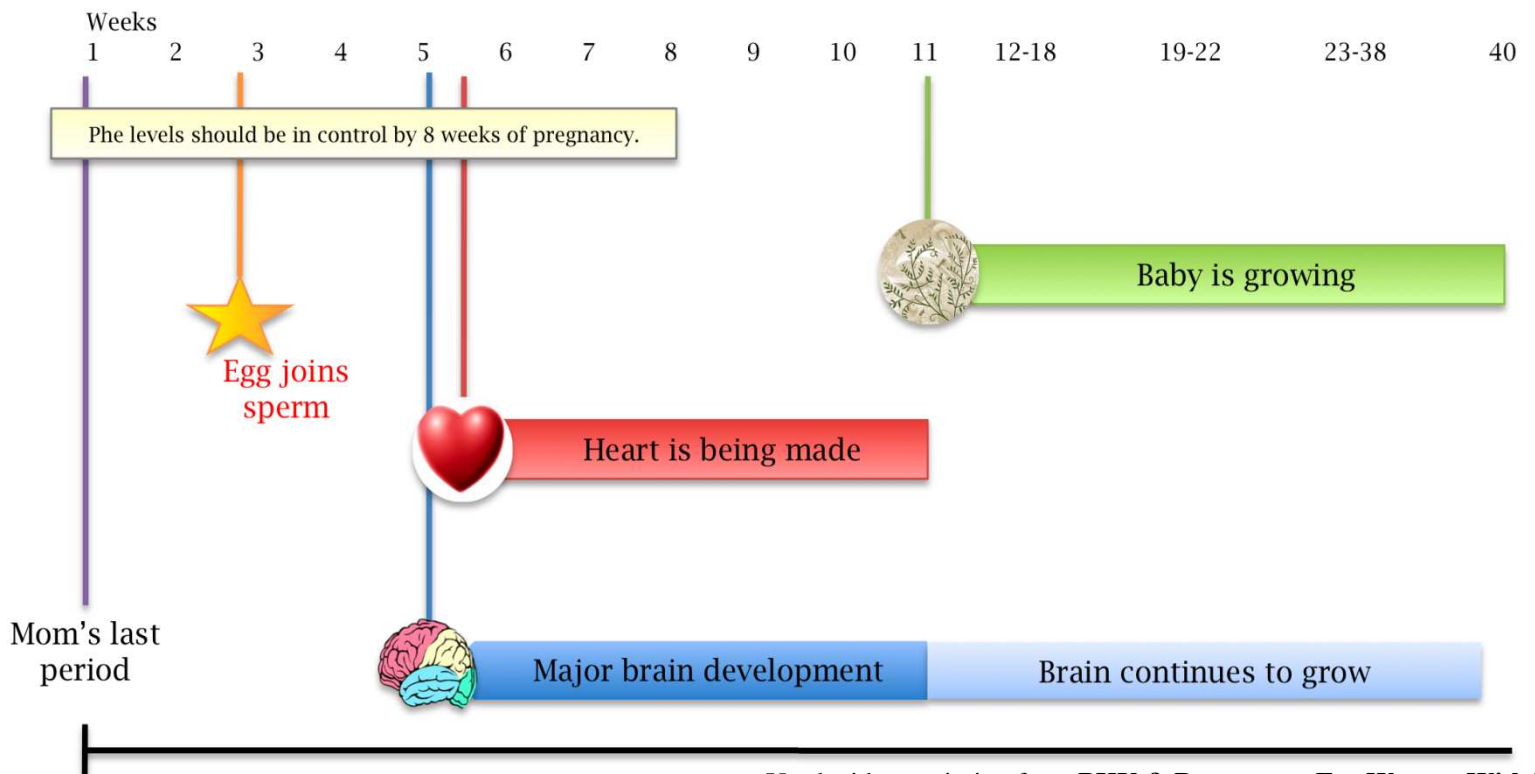
**360  $\mu\text{mol/L}$**

lead to increased  
risk for birth  
defects in the fetus!

# PREGNANCY DEVELOPMENT TIMELINE

**Untreated maternal hyperphenylalaninemia is known to cause the following birth defects:**

- ❖ Heart Defects
- ❖ Esophageal Defects
- ❖ Microcephaly
- ❖ Mental Retardation
- ❖ Low Birth Weight



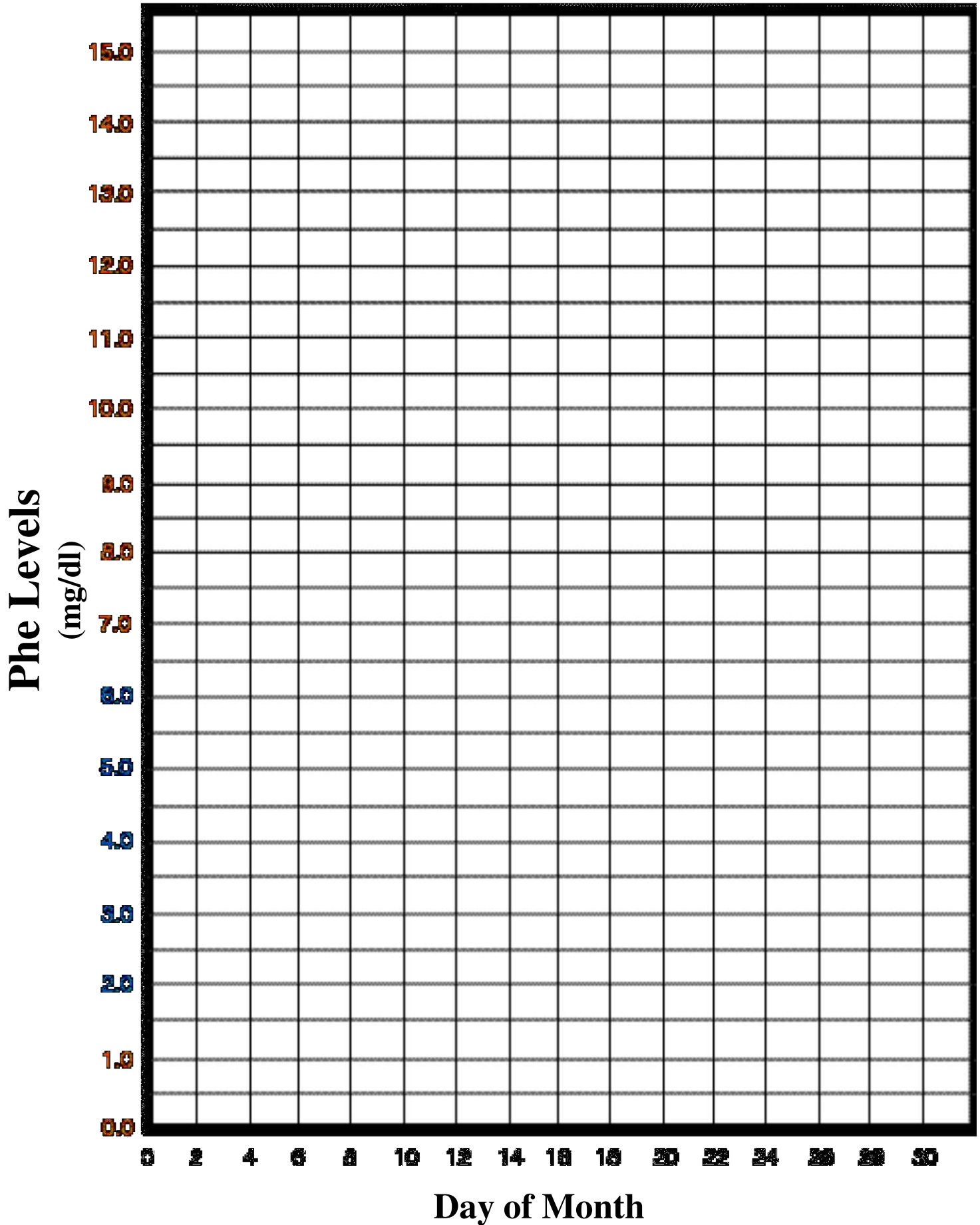
## Additional Remarks:

- There is a link between high Phe levels and risk for birth defects. Early control of Phe levels decreases these risks.
- These ranges do not represent strict cut-offs for the development of these organs, they are approximations. Each pregnancy is different, and development rates vary.
- There are other systems developing concurrently that are not represented on this diagram.
- There is a 3-5% risk for birth defects in ANY pregnancy.



# PHENYLALANINE LEVELS GRAPH

Month \_\_\_\_ of Pregnancy



# PREGNANCY CALENDAR

## FIRST TRIMESTER

Week	Phe Level	Tyr Level	Description of Development
1			One week after last menstrual period
2			Conception takes place
3			Implantation of fertilized egg into uterus
4			Placenta begins to form
5			Heart and cardiac system begins to form; formation of the neural tube
6			Embryo is 1/5 of an inch in length; primitive heart is beating and pumping blood; head, eyes, ears, and mouth begin to form
7			Formation of lenses of the eye, middle parts of ear
8			Eyelids begin to form; ears are taking shape; wrists and elbows become evident; fingers begin to form
9			Formation of the pancreas, bile ducts, gall bladder, and internal reproductive organs
10			Embryo is 1 inch in length; beginning of all major organs are formed; formation of the skeleton; facial features begin to take shape
11			Neck develops; external genitalia begins to be gender specific
12			Fingernails appear; gender differences are apparent

# PREGNANCY CALENDAR

## SECOND TRIMESTER

Week	Phe Level	Tyr Level	Description of Development
13			Nose begins to develop bridge; fetus begins to excrete urine into amniotic fluid
14			Fetus is 3 inches long and weighs almost an ounce; muscles begin to develop
15			<p><b>By 22 Weeks of Gestation:</b></p> <ul style="list-style-type: none"> <li>• Fetus weighs approximately ½ pound and is 10 inches long</li> <li>• Eyebrows and scalp hair have begun to form</li> <li>• Skin is covered with vernix (a protective coating) and lanugo (fine hair)</li> </ul>
16			
17			
18			
19			
20			
21			
22			
23			
24			

# PREGNANCY CALENDAR

## THIRD TRIMESTER

Week	Phe Level	Tyr Level	Description of Development
25			<ul style="list-style-type: none"><li>• Surfactant covers the inner lining of the air sacs of the lungs, which allows them to expand easily; blood vessels are rapidly developing in the lungs and brain; fetus can inhale, exhale, and cry; tongue now has taste buds and eyes are completely developed.</li><li>• The lungs and brain are continuing to mature; the fetus can open and close its eyes; in the male, the testicles are descending from an area near the kidneys to the scrotum; in the female, the labia are still underdeveloped and do not completely cover the relatively prominent clitoris.</li><li>• In the lungs, fluid is being absorbed so the fetus can breathe following birth; there is a surge of fetal hormones that may function in maintaining blood pressure and blood sugar levels following birth; in the male, the testicles have fully descended into the scrotum.</li></ul>
26			
27			
28			
29			
30			
31			
32			
33			
34			
35			
36			
37			
38			
39			
40			

# DNA BLUEPRINT



Instructions are normal.  
Phenylalanine hydroxylase  
(PAH) is made. Works  
efficiently.



Instructions have an error.  
Phenylalanine hydroxylase  
(PAH) is made, however it  
does not work as well.

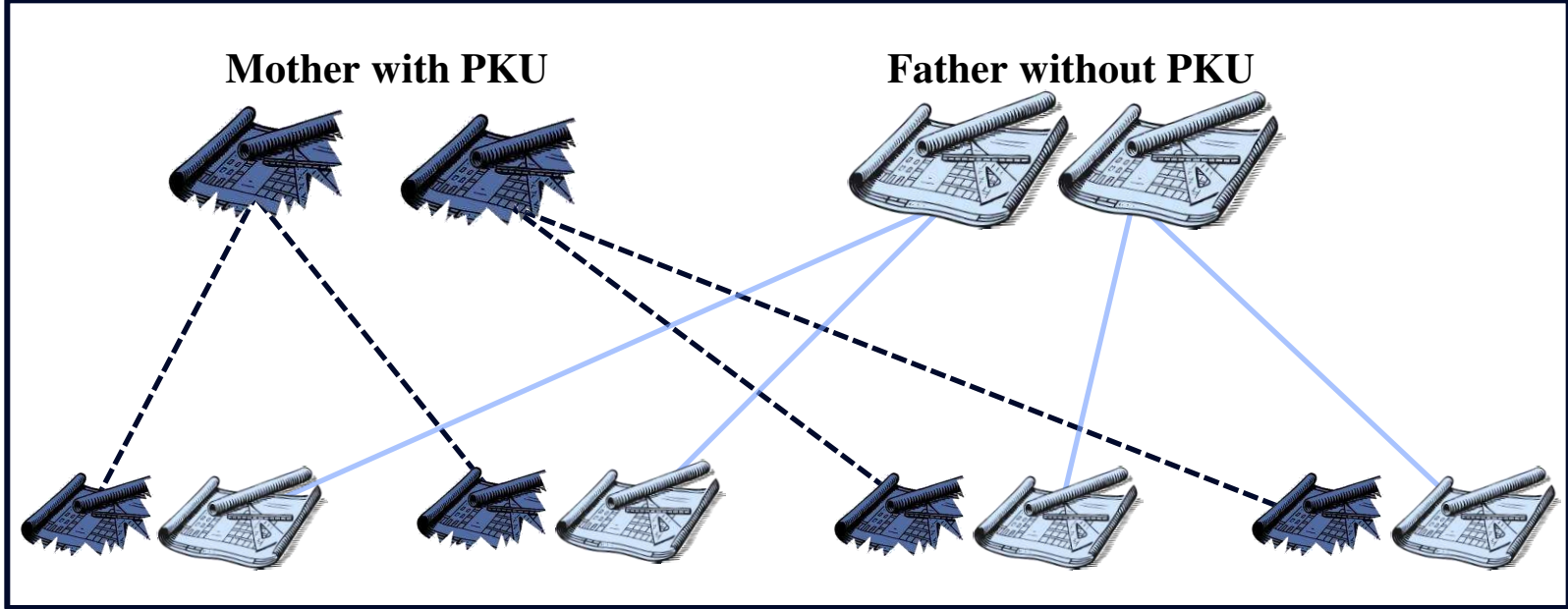


A significant amount of  
instructions are missing. No  
phenylalanine hydroxylase  
(PAH) can be made.

# AUTOSOMAL RECESSIVE INHERITANCE

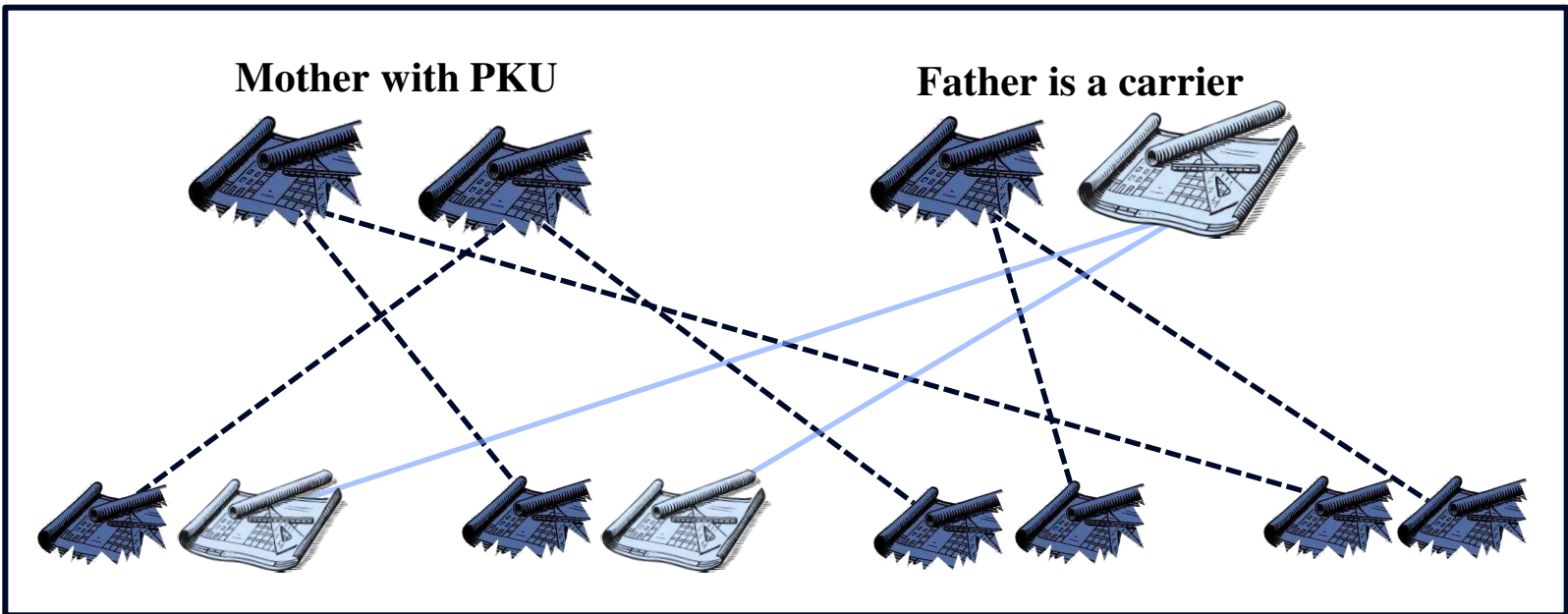
Mother with PKU

Father without PKU



Mother with PKU

Father is a carrier



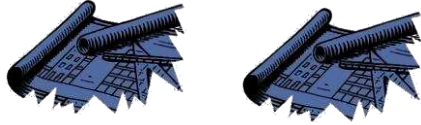
Gene for normal or full phenylalanine hydroxylase (PAH) activity



Gene for altered phenylalanine hydroxylase (PAH) activity

# AUTOSOMAL RECESSIVE INHERITANCE

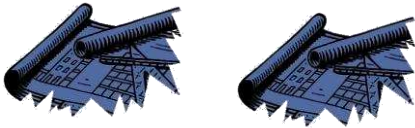
**Mother with PKU**



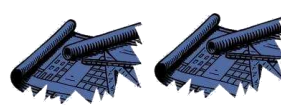
**Father without PKU**



**Mother with PKU**



**Father who is a carrier**



Gene for normal or full phenylalanine hydroxylase (PAH) activity



Gene for altered phenylalanine hydroxylase (PAH) activity

# AUTOSOMAL RECESSIVE INHERITANCE

## Chromosome #12 and the PKU Gene

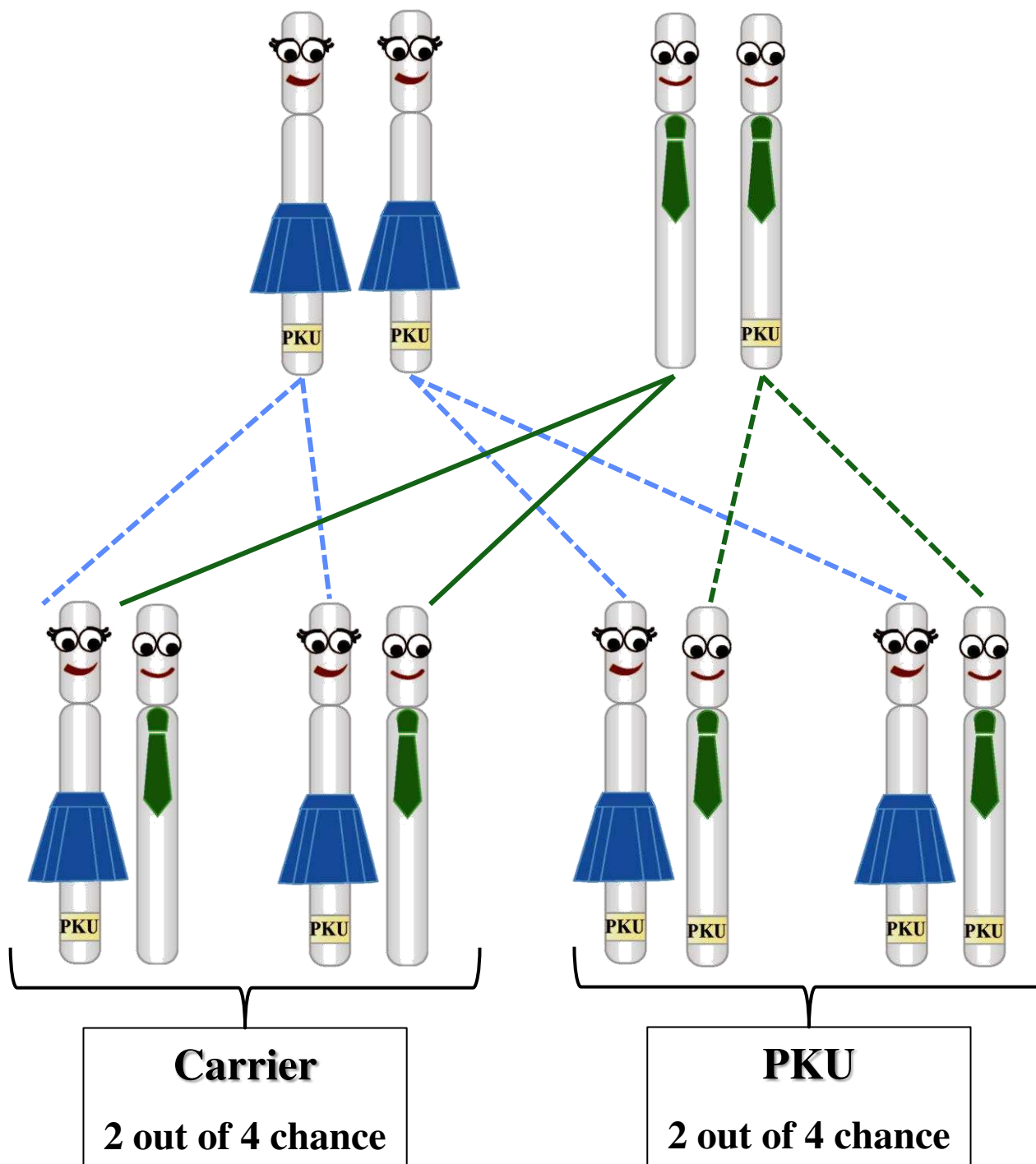
Our genes are organized on our body on Chromosomes.

Mrs. Chromosome has PKU and Mr. Chromosome is a carrier; see their chances of having PKU based on Autosomal Recessive Inheritance.

**Mrs. Chromosome**    **Mr. Chromosome**

**has PKU**

**is a carrier.**





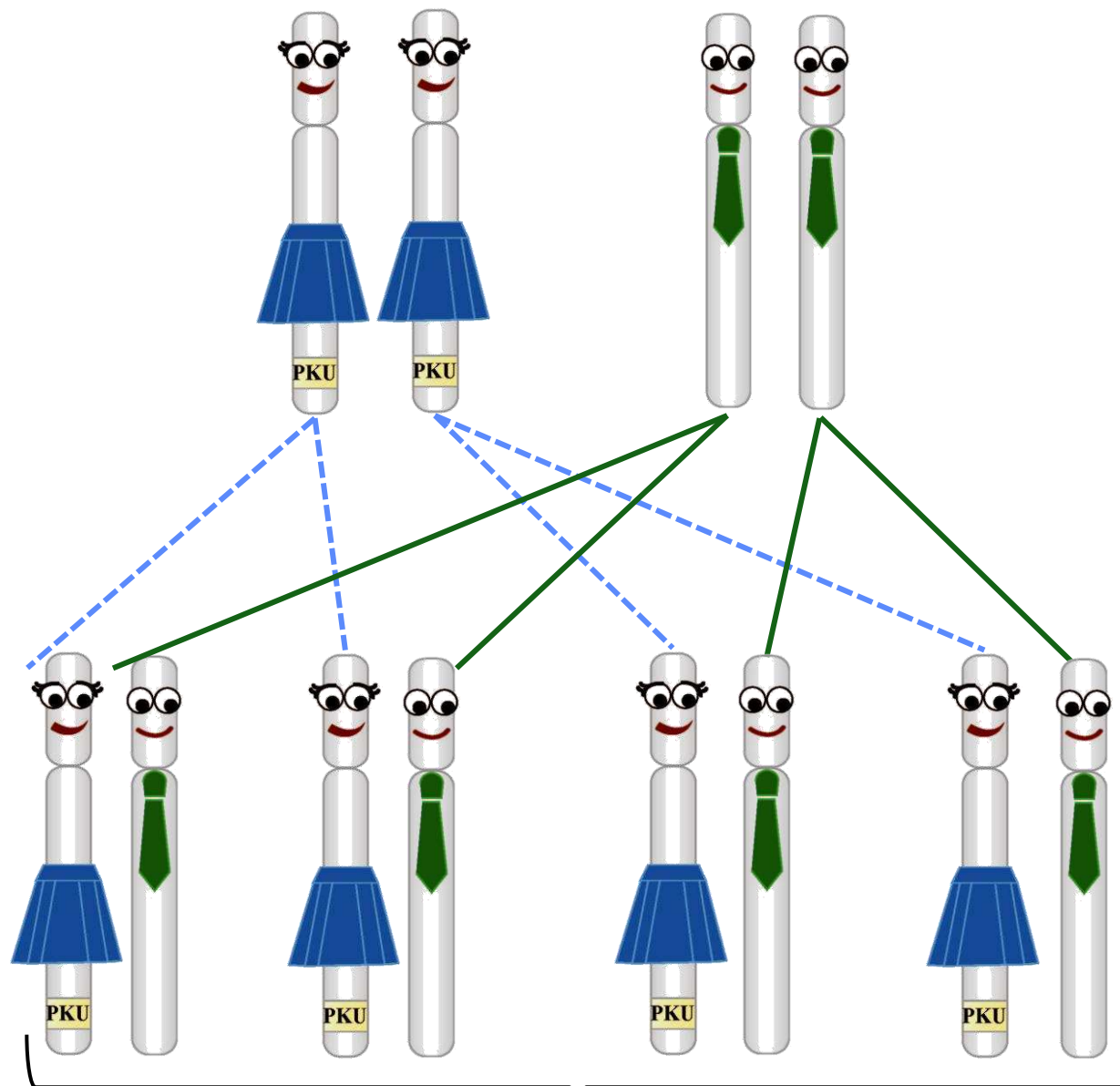
# AUTOSOMAL RECESSIVE INHERITANCE

## Chromosome #12 and the PKU Gene

Our genes are organized on our body on Chromosomes.

Mrs. Chromosome has PKU and Mr. Chromosome doesn't; see their chances of having PKU based on Autosomal Recessive Inheritance.

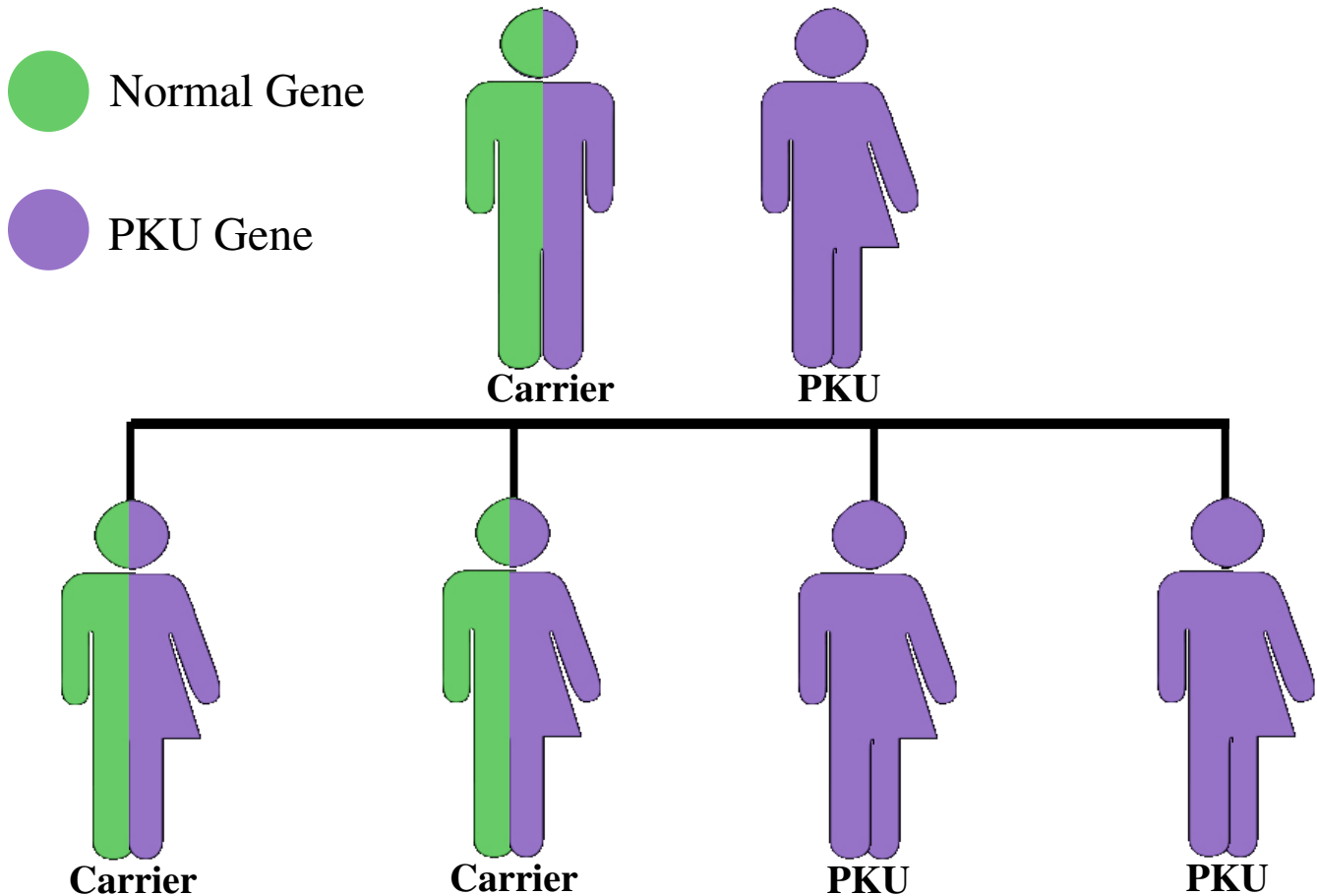
**Mrs. Chromosome**    **Mr. Chromosome**  
**has PKU**            **doesn't have PKU**



**Carrier**  
**4 out of 4 chance**

# AUTOSOMAL RECESSIVE INHERITANCE

Dad is a carrier. Mom has PKU.



## WHAT DOES AUTOSOMAL RECESSIVE INHERITANCE REALLY MEAN?

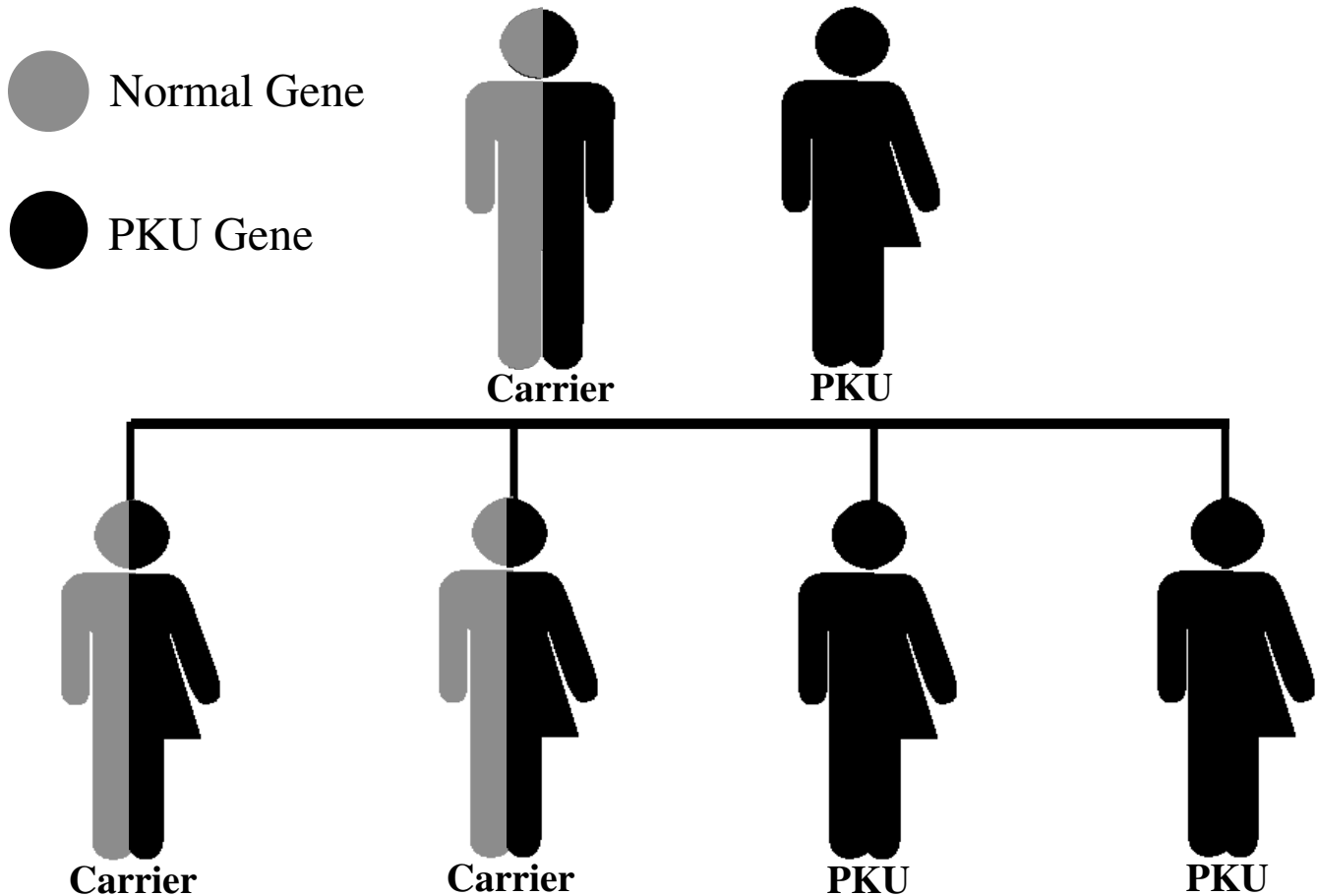
Autosomal recessive inheritance means you have to receive two copies of an altered gene, one from each parent, to have the condition. Individuals who have only one copy of an altered gene are called carriers and do not have the condition.

If a woman has PKU she will always pass a PKU gene down to her children. If her partner is a carrier, there is a 50% chance that he will pass a PKU gene down.

There is a 50% chance that a child will have PKU and a 50% chance that the child will be a carrier.

# AUTOSOMAL RECESSIVE INHERITANCE

Dad is a carrier. Mom has PKU.



## WHAT DOES AUTOSOMAL RECESSIVE INHERITANCE REALLY MEAN?

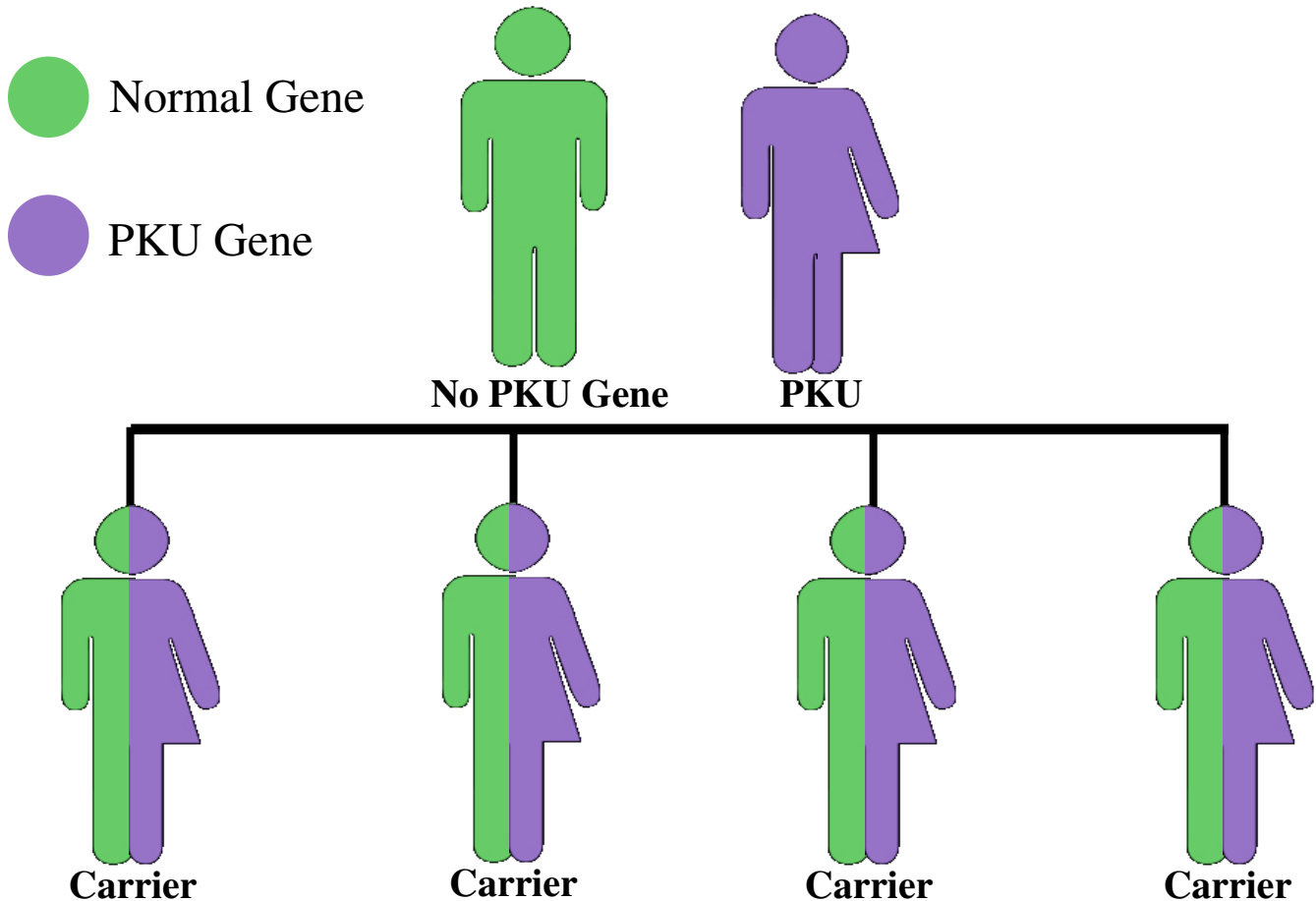
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There is a 50% chance that a child will have PKU and a 50% chance that the child will be a carrier.

# AUTOSOMAL RECESSIVE INHERITANCE

Dad does not have PKU and is not a carrier. Mom has PKU.



## WHAT DOES AUTOSOMAL RECESSIVE INHERITANCE REALLY MEAN?

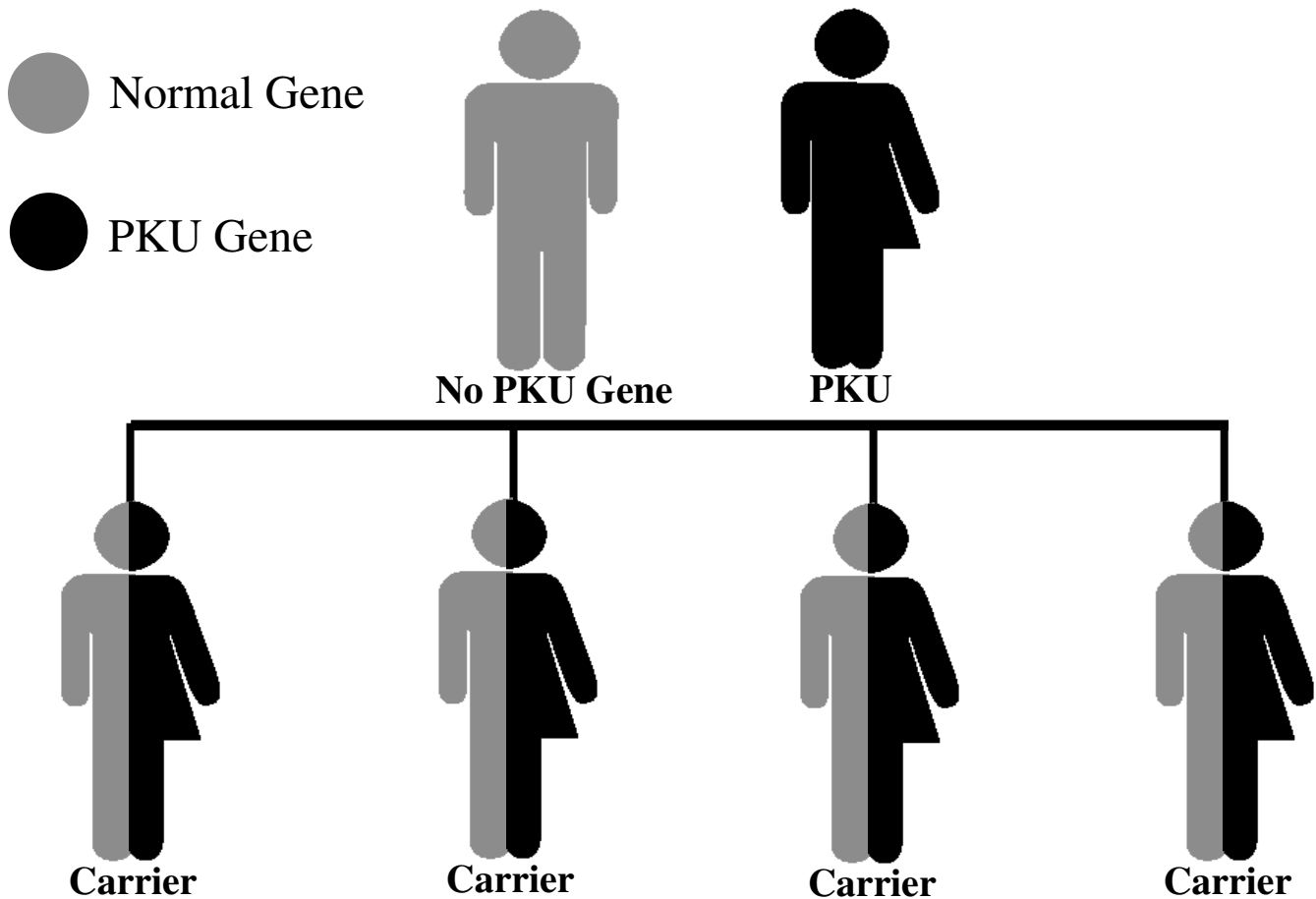
Autosomal recessive inheritance means you have to receive two copies of an altered gene, one from each parent, to have the condition. Individuals who have only one copy are called carriers. They do not have the condition.

If a woman has PKU she will always pass a PKU gene down to her children. If her partner does not have PKU and is not a carrier, he will never pass a PKU gene down.

All offspring will be carriers, but none will have PKU.

# AUTOSOMAL RECESSIVE INHERITANCE

Dad does not have PKU and is not a carrier. Mom has PKU.



## WHAT DOES AUTOSOMAL RECESSIVE INHERITANCE REALLY MEAN?

Autosomal recessive inheritance means you have to receive two copies of an altered gene, one from each parent, to have the condition. Individuals who have only one copy are called carriers. They do not have the condition.

If a woman has PKU she will always pass a PKU gene down to her children. If her partner does not have PKU and is not a carrier, he will never pass a PKU gene down.

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**More Phe, More Choices: Think Healthy During Pregnancy!** Laurie Bernstein, Sommer Myers, Doug Neuschwanger. IMD Clinic, The Children's Hospital, Aurora, CO. 303-724-2338

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