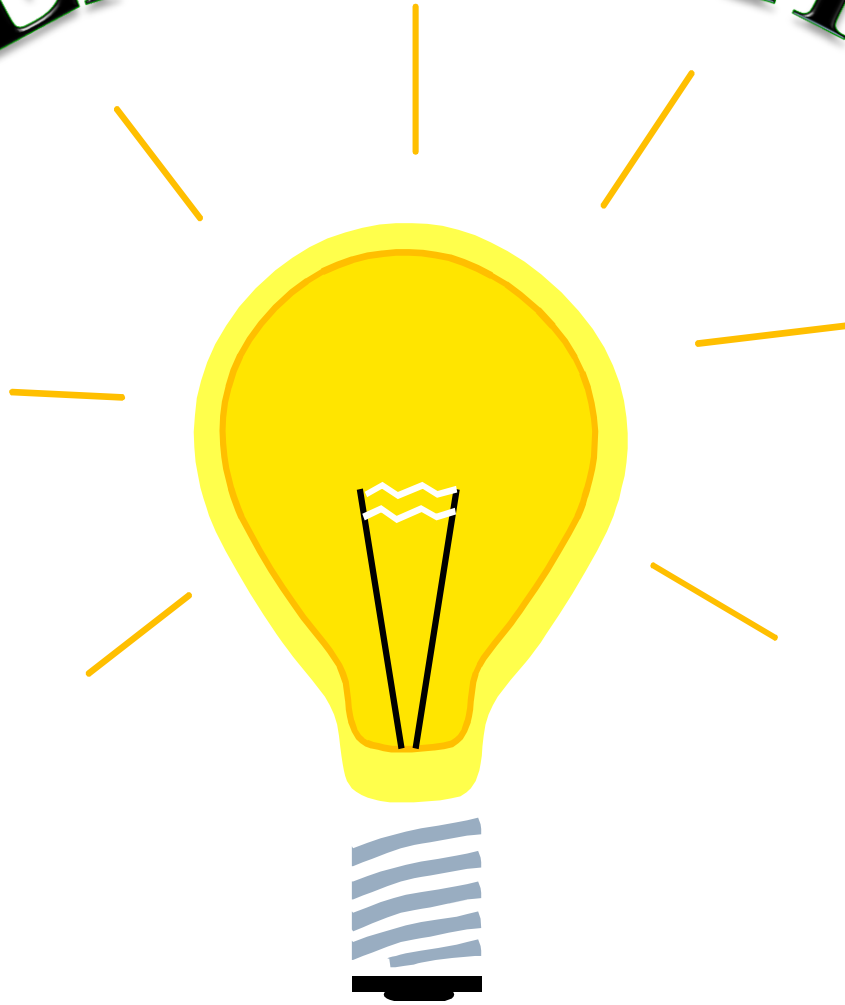


EAT RIGHT



STAY BRIGHT™

Guide For Hyperphenylalaninemia



The Children's Hospital

Affiliated with
University of Colorado Denver

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CHAPTER ONE



Birth to Five Years Old

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.

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CHECKLIST: Preparation for First Encounter

- ☐ Establish relationship with primary medical care provider

Query

- ☐ Nuclear family members/support
- ☐ Geographic location
- ☐ Insurance status
- ☐ Breast fed versus formula fed, source of formula if formula fed
- ☐ What information has been given to the family

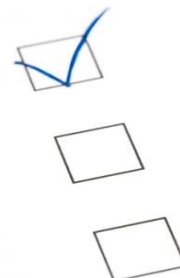
Information To Be Given To The Family

- ☐ Expected length of first clinic visit
- ☐ Bring 24 hr diet record to first visit
- ☐ No change in diet until clinic visit

Practical Issues

- ☐ Registration
- ☐ Laboratory Notification And Result Tracking

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



CHECKLIST: The First Encounter

General Hyperphenylalaninemia Information

- ☐ Biochemistry (BIOCHEMISTRY)
 - Hepatic phenylalanine hydroxylase deficiency
 - Biopterin cofactor abnormalities
 - Phenotype and molecular heterogeneity
- ☐ Autosomal recessive disorder (GENETICS)
 - Recurrence risk
 - Quantitative testing for future siblings (see form letter)
 - DNA carrier testing
 - Family history
 - Identify family member(s) in whom hyperphe/carrier testing is indicated/desired
- ☐ Principles of dietary management
- ☐ Outcome (OUTCOMES OF TREATMENT)
 - Untreated
 - Treated
- ☐ Diet is positive¹
- ☐ Diet for life
- ☐ Emotional response to diagnosis of chronic metabolic disease
 - Denial
 - Fear
 - Anger
 - Guilt
 - Over involvement
 - Sorrow/sadness

The Prescription (Rx)

- ☐ Principles of prescription (DIET)
- ☐ Gram scale:
 - Hands on demonstration with the family
- ☐ 24 hr clock
 - Explain with use of diet records
- ☐ Formula preparation (FORMULA PREPARATION)
 - Measuring and mixing



CHECKLIST: The First Encounter

Daily Living Issues²

- ☐ Family reorganization
 - New family member
 - New family member with a chronic disease
 - “Why Can’t I Eat That?”, *pg.* 113-127
- ☐ Need to maintain lifestyle normalcy
- ☐ Otherwise normal child
- ☐ Siblings³
 - “Why Can’t I Eat That?”, *pg.* 113-127
- ☐ Parents plans to return to work or school
- ☐ Child care⁴
- ☐ Loss of privacy

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 form letter - handout section.

Resources

- ☐ Extended family
- ☐ Parent support groups
- ☐ Community agencies
- ☐ Spiritual support



CHECKLIST: The First Encounter

Primary Medical Care Provider (PMP)

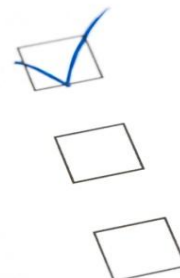
- ☐ Team player (parent, metabolic clinic, PMP)
- ☐ Open lines of communication
- ☐ General health issues to be directed by PMP

The Clinic Routine (THE CLINIC ROUTINE)

- ☐ Blood draws
 - Procedure
 - Frequency
- ☐ Laboratory results
 - Procedure
 - Frequency
- ☐ Diet records
 - Procedure
 - Frequency
- ☐ Weights/length
 - Procedure
 - Frequency
- ☐ Diet prescription changes
 - Procedure
 - Frequency
- ☐ Appointments
 - Frequency of visits
 - Flow at visits
 - Option to meet other PKU families at next clinic appointment
 - Group clinics
- ☐ Clinic staffs' contact numbers
 - Routine
 - Emergency

Handouts

- ☐ A Caregivers Guide
- ☐ Include your specific clinic handouts



CHECKLIST: Birth to Four Months Old

Review

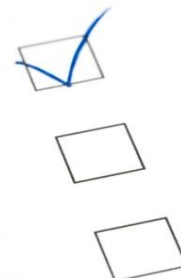
- ☐ Biochemistry
 - Bipterin cofactor results
- ☐ Genetics (plans for future pregnancies)
- ☐ Principles of dietary management
- ☐ Principles of diet prescription

Phe Levels, Growth Charts, and Interim History

- ☐ Intercurrent levels
 - Phenylalanine (Phe)
 - Tyrosine (Tyr)
- ☐ Immunizations
- ☐ Teething
- ☐ Intercurrent illness
- ☐ Lengths and weights

Daily Living Routine

- ☐ Weighing, measuring and preparing formula
- ☐ Diet records/24 hr clock
- ☐ Feeding schedule
- ☐ Blood draws



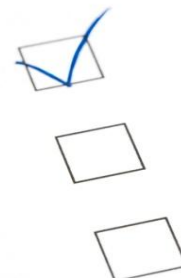
CHECKLIST: Birth to Four Months Old

Psychosocial Adjustment

- ☐ Process of adjusting to diagnosis
- ☐ Process of adjusting to diet
- ☐ Validate feelings
- ☐ Support system
- ☐ Plans to return to work or school
- ☐ Child care
- ☐ Start process of communication⁵
- ☐ Trigger questions:
 - Are you angry (if so, at whom)?
 - Have you bonded to your baby?
 - Are there shared responsibilities with the diet?
 - How is your relationship?
 - Do you feel overwhelmed?
 - Do you feel responsible or guilty?

Nutrition Intervention

- ☐ Developmental readiness
 - Preparing for solid foods
- ☐ Nutrition education
 - Phe from food
 - Low Phe food lists (supply order forms)
 - Low protein foods (supply order forms)
 - Label reading (example: NutraSweet in medications)



CHECKLIST: Four to Six Months Old

Review

- ☐ Biochemistry
- ☐ Genetics (plans for future pregnancies)
- ☐ Principles of dietary management
- ☐ Principles of diet prescription

Phe Levels, Growth Charts, and Interim History

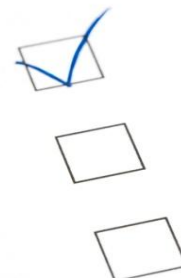
- ☐ Intercurrent levels
 - Phenylalanine (Phe)
 - Tyrosine (Tyr)
- ☐ Immunizations
- ☐ Teething
- ☐ Intercurrent illness
- ☐ Lengths and weights

Daily Living Routine

- ☐ Weighing, measuring and preparing formula
- ☐ Diet records/24 hr clock
- ☐ Feeding schedule
- ☐ Blood draws

Psychosocial Adjustment

- ☐ Process of adjusting to diagnosis and diet
- ☐ Validate feelings
- ☐ Support system
- ☐ Plans to return to work or school⁷
- ☐ Child care
- ☐ Relationship with the clinic
- ☐ The parents role as child advocate



CHECKLIST: Four to Six Months Old

Psychosocial Adjustment...



Trigger questions:

- How do you think having a baby has changed your life? How do you think having a baby with PKU has changed your life?
- Have there been any major changes or stress in your family since last visit?
- How are you balancing your roles of parents and partners?
- Do you have extended family support?

Nutrition Intervention



Developmental readiness

- Introduction of solid foods



Nutrition education

- Sequencing of solid food
- Suggested meal patterns (SUGGESTED MEAL PATTERN)
- Appropriate and inappropriate feeding practices (see handouts)
- The meal time mess⁸



Educate siblings, family members and friends

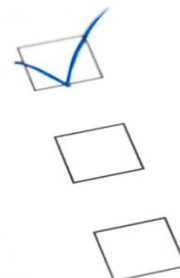
Additional Handouts



Why Is Mary On A Diet? (for siblings)



Online Tool: Denny the Dragon and His Magic MILK (for siblings)



CHECKLIST: Seven To Twelve Months Old

Review

- ☐ Biochemistry
- ☐ Genetics (plans for future pregnancies)
- ☐ Principles of dietary management
- ☐ Principles of diet prescription

Phe Levels, Growth Charts, and Interim History

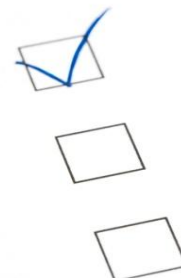
- ☐ Intercurrent levels⁹
- ☐ Immunizations
- ☐ Teething
- ☐ Intercurrent illness
- ☐ Lengths and weights

Daily Living Routine

- ☐ Weighing, measuring and preparing formula
- ☐ Diet records/24 hr clock
- ☐ Feeding schedule
- ☐ Blood draws

Psychosocial Adjustment

- ☐ Process of adjustment to diagnosis¹⁰ and diet
- ☐ Validate feelings
- ☐ Support system
- ☐ Plans to return to work or school
- ☐ Relationship with the clinic
- ☐ The parents role as child advocate
- ☐ Diet button/manipulative behavior



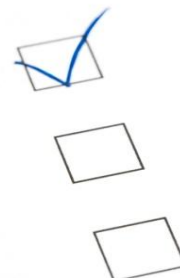
CHECKLIST: Seven To Twelve Months Old

Psychosocial Adjustment ...

- ☐ Trigger questions:
 - Do you have time for yourself?
 - Who do you turn to when you need some help?
 - What are your thoughts on discipline?

Nutrition Intervention

- ☐ Introduction of low protein foods/free foods
 - Order forms
 - Food lists
 - Cook books
- ☐ Suggested meal patterns (SUGGESTED MEAL PATTERN)
- ☐ Introducing the “Yes” and “No” food concepts
- ☐ Educating siblings, extended family, friends¹¹
- ☐ First birthday party--offer ideas for low protein cake and ice cream (PARTY TIME RECIPES)
- ☐ Developmental readiness
 - Finger foods (CHOOSE YOUR FOODS)
 - Cup drinking ¹²
- ☐ The mobile child



CHECKLIST: One to Three Years Old

Review

- ☐ Genetics (plan for future pregnancies)
- ☐ Principles of dietary management
- ☐ Principles of diet prescription

Phe Levels, Growth Charts, and Interim History

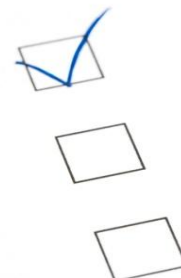
- ☐ Intercurrent levels
- ☐ Immunizations
- ☐ Intercurrent illness
- ☐ Heights and weights

Daily Living Routine

- ☐ Weighing, measuring and preparing formula
- ☐ Weighing, measuring and preparing food
- ☐ Diet records/24 hr clock
- ☐ Blood draws

Psychosocial Issues¹³

- ☐ Living with diagnosis and diet
- ☐ Stress management
- ☐ Support system
- ☐ Plans to return to work or school
- ☐ Relationship with the clinic
- ☐ The parents role as child advocate
- ☐ Impact of child's increasing autonomy¹⁴



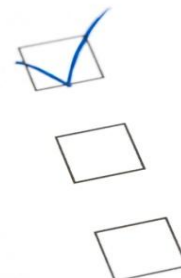
CHECKLIST: One to Three Years Old

Parenting Skills and Chronic Disease

- ☐ Self-concept¹⁵
- ☐ Use of positive language when communicating with child
- ☐ Over involvement
 - “Why Can’t I Eat That?” pg. 16-22
- ☐ Participation in individualizing the child’s health care plan
- ☐ Trigger questions
 - How important are neatness and efficiency?¹⁶
 - Do you feel exhausted?
 - Do you let your child know if a food is allowed/not allowed on the diet? If so, how?
- ☐ Teaching through reading¹⁷ (see references)

Nutrition Intervention

- ☐ Feeding independence
- ☐ Issues with non-compliance, formula acceptance
- ☐ Revisit use of low protein foods/free foods
 - Cookbooks
 - Recipes
- ☐ Variety ¹⁸
- ☐ Educate the child; “Yes” and “No” foods
- ☐ Educate siblings, family, friends
- ☐ The mobile child (exploring his surroundings)



CHECKLIST: Three to Five Years Old

Review

- ☐ Genetics (plan for future pregnancies)
- ☐ Principles of dietary management
- ☐ Principles of diet prescription

Phe Levels, Growth Charts, and Interim History

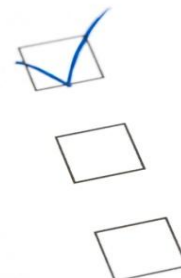
- ☐ Intercurrent levels
- ☐ Immunizations
- ☐ Intercurrent illness
- ☐ Heights and weights

Daily Living Routine

- ☐ Weighing, measuring and preparing formula
- ☐ Weighing, measuring and preparing food
- ☐ Diet records/24 hr clock
- ☐ Blood draws

Psychosocial Issues¹⁹

- ☐ Living with diagnosis and diet
- ☐ Stress management
- ☐ Support system
- ☐ Plans to return to work or school
- ☐ Daycare/ Preschool
- ☐ Relationship with the clinic
- ☐ The parents role as child advocate



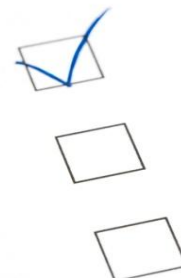
CHECKLIST: Three to Five Years Old

Parenting Skills and Chronic Disease

- ☐ The egocentric child ²⁰
- ☐ The negotiating child ²¹
- ☐ Building trust ²²
- ☐ Over involvement
 - “Why Can’t I Eat That?” pg. 16-22
- ☐ The child’s ambassador ²³

Nutrition and Genetic Education ^{20, 21, 22}

- ☐ “Yes” and “No” foods (RED LIGHT!! GREEN LIGHT!!)
- ☐ Similarities and differences (WAYS WE ARE ALIKE & WAYS WE ARE DIFFERENT)
- ☐ Role playing (ROLE PLAYING)
- ☐ Saying “No” (FISHING FOR PHE)



EXPERIENCE & THOUGHTS

1. We stress from the first encounter that diet is positive. We often remind the family that before the development of newborn screening and without diet therapy, their child would have been mentally retarded.
2. The initial clinic visit is an excellent time to set the tone for your clinic's philosophy regarding management. For example, we have many new families who state they will change their life style and become "vegetarian" to make it easier for their child. Our clinic philosophy is to stress life maintenance of a normal lifestyle. The families' acceptance of this philosophy can best be promoted by the following observations:
 - There is no reason for the parent to apologize for the diet; without the diet, their child would be mentally retarded.
 - Their child has a metabolic disease; they do not. The treatment for the disease is diet. If their child was born with a heart disorder for which the treatment was daily medication, would they feel compelled to take the same medication as their child was prescribed.
 - Their child will be on a metabolic formula, the rest of the family will not. The metabolic formula will provide their child with the necessary amino acids, vitamins and minerals for life. Family members, whether they are vegetarian or not, must ensure there is an adequate intake of amino acids, vitamins and minerals from the food they eat. Therefore, unless parents plan to make themselves and the rest of the family nutritionally deficient, their diet must be different from their child's, independent of whether they are vegetarian or not.
 - Their child will not be living with them forever. She/he will soon be exposed to the "protein eating" world. Their child needs to learn how to manage the emotional and practical problems which accompany any special diet. This can best be taught at home where there is a supportive, loving environment.=-

Tailor your responses to parents' concerns or questions to support your clinic's philosophy.

3. Siblings need to be included. Sharing the family experience allows them to feel more involved. Using their questions as guidelines helps to establish age appropriate dialogue.
4. It has been our experience that providing potential care providers an opportunity to come to clinic, to learn the philosophy and the "nuts and bolts" of the diet from the staff, in addition to learning from the immediate family, increases compliance.
5. Parents talk to their child during baths, diaper changes, and play time. It has been our practice to recommend to parents that talking to their infants about their diet is a learning experience. It makes it easier for parents to talk to their child when she/he is older because they have always been doing it. Talking to their baby is natural.
6. It has been our experience that couples often feel angry about having to call the clinic when their child is hungry or when their appetite changes. It is a natural reaction to be angry when the most innate behavior (to feed your child) is supervised so closely. Our recommendation is to be supportive and validate their feelings. Keeping lines of communication open, usually leads to a good long term relationship with the family.

EXPERIENCE & THOUGHTS

7. It has been our experience that parents often indicate one care provider plans to remain at home. Frequently by four months of age financial issues may cause reconsideration.
8. Encourage families to feed their baby like any other baby. It's normal when introducing solids to have a MESS. This is taken into consideration when calculating the diet prescription. Reminding families that mess is normal reduces the risk of "**Phe-neurosis**", a term we coined to describe the fear that develops (with our help) regarding total Phe intake from food.
9. **Staff reminder:** high levels do not necessarily mean family and/or clinic staff is doing a "bad" job; similarly low levels do not necessarily mean family is doing a "good" job.
10. Frequently, parents breathe a sigh of relief when their child reaches a year of age. By this time their child's normal growth and development is evident and reassuring. Now is a good time to revisit the fact that diet for a child with hyperphe is as essential as medication for heart failure or insulin for diabetes. We have found that playing down the fact that their child does have a metabolic disease has led to poor compliance, especially in teenage years. This is the time to reinforce that their child does indeed have a metabolic disease which, if untreated, causes mental retardation. With diligence, compliance, and education the outcome can continue to be positive.
11. It is the parents' choice whether people having occasional contact with their child need to know about their child's diagnosis and special diet. For example, perception that the diet is due only to a food allergy rather than a metabolic disease minimizes the importance of the diet therapy. Allergies are often perceived as a diagnosis of over concerned parents and nonorganic. On the other hand, lengthy explanations are not always needed.
12. It has been our experience that parents resist the transition from bottle to cup because of the message we have given them regarding the importance of consuming ALL the formula within their 24 hour clock. A fear develops (**Phe-neurosis**) around the normal behavior that a child exhibits when they learn to drink from a cup (i.e., turning the cup upside down). This is a delicate issue because there is no easy solution. Depending on the family and their child, various approaches may be needed to attain the goal of cup drinking without compromising the diet or driving the parents over the edge. One suggestion may be to put the metabolic formula without the infant formula in the cup. This will introduce the concept of drinking "milk" from a cup, but not risking the diet prescription.
13. At this age, we change the heading from psychosocial adjustment to psychosocial issues. It has been our experience that adjustment to the diagnosis and the diet has usually occurred by one year of age. It appears that once our families have adjusted they can then allow themselves to honestly acknowledge that there will be ongoing psychosocial issues due to the chronic nature of the disease.
14. The physical and cognitive maturity of a child this age allows for the development of autonomy. The resistance and negativism exhibited is not defiance, but a normal developmental stage; recognize this as transitional.
15. Do the parents understand their own feelings?
16. Are parents telling you, "I have to make this formula because I do it right. They do not measure properly, they make a mess, and it's easier if I just do it!" Ask the primary care taker, "Is this the healthiest decision for the family?"

EXPERIENCE & THOUGHTS

17. Reading is an educational activity and fun for both children and parents. Family's home library and the public library have numerous books that can be used to teach a variety of situations and choices. For reading materials useful in addressing "yes" and "no" foods, role playing, friendships, and the body, see the PKU literature listing on the PKU Homepage at <http://www.wolfnet.com/~kommel/books/html>.

18. Variety touches on many issues:

- Financial - Can the family afford low protein foods?
- Emotional - How does the family feel about saying no to foods they are eating?
- Lifestyle - Is there time to prepare different recipes? Restaurant eating? Fast food junkies? Is there an interest in cooking?

This is a good time for the nutritionist to review the recipe book with the family and provide additional easy to make snacks etc. Review policy on Low Protein foods reimbursement. Discuss menu options at local restaurants. Variety doesn't always mean more work. It is our belief that an early introduction to variety will save time, and decrease trouble with non compliance later on.

19. At three to four years of age we begin having clinics in a group setting. Although we continue to do individual intakes and address any individual concerns with the families in private, the focus of the clinic encounter is to provide an interactive educational environment. Initially, both child and parent(s) are involved in the same setting; this arrangement avoids potential separation anxiety. It had been our experience that parents in the setting glean insights regarding the management of their child's disease. Through sharing, observing, and networking, the families gain invaluable knowledge and support. Despite the long distances that many families travel and the additional cost of lodging, the families have been overwhelmingly supportive. Starting groups at this age helps form life long friendships for child and parent alike.

20. At approximately three years of age their child is becoming aware of the world around them, but is still an egocentric individual. Role playing is a tool for incorporating what is being learned and how it relates to his/her world. Symbolic and imaginative play are just two of the approaches we use in our groups for this age. An example of this would be using plastic play food and asking each child to set the table, select a meal, and identify "yes" and "no" foods.

21. With the development of language their child is now able to negotiate. Choices are now a critical learning tool. Without choice, a child's actions are mandated, often leading to power struggles. Keeping all choices acceptable helps to ensure a positive outcome for all participants. Example: *"Which finger do you want poked for your blood draw?"*

22. Trust is a fragile thing. Most children have learned to trust their parents. Honesty and consistency will help nurture the trust and the relationship that the child and the parents have both with each other and the clinic. Casual deception and broken promises can seriously jeopardize it. When a child asks, "Why can't I have some hamburger?" a short direct answer may be emotionally difficult for the parents, but an evasive answer is not beneficial to their child. "Because you have hyperphage, and hamburgers are not good for you," is an example of a direct answer. A response of "Not today dear" is evasive and implies that in the future it may be possible.

23. If their child has started preschool, parents must now provide information on their child's metabolic disease and diet to the school staff. Written material that supports verbal communication will support positive outcome. Ongoing communication and clarification of expectations is necessary for a working relationship.

THE CLINIC ROUTINE

THIS IS NOT YOUR CLINIC ROUTINE

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

The clinic routine refers to the overall flow. It is indicative of both what happens during an actual visit and what is expected for maintaining patient care.

Blood Draws

- Blood is drawn either at The Children's Hospital at the outpatient laboratory or it can be drawn at home. Technique for drawing home levels can be taught at the outpatient lab and tubes, labels, etc. can be mail ordered.
- Blood is drawn on a daily basis until levels are in treatment range. Within the first 6 months levels are drawn weekly and then bi-monthly until a year of age. Levels remain at bi-monthly after a year of age and once a month between the ages of 2-10 years of age. Frequency of levels can increase or decrease depending on the patient's compliance.
- Results of laboratory tests will be phone called to the family or a results letter will be sent.
- Blood is drawn 2-4 hours after a meal. It is not recommended that blood be drawn after a fast of more than 4 hours in young children.
- Older Children can choose to have their blood drawn first thing in the morning, after a bed-time fast.

Laboratory Results

- All blood specimens must be received Monday and Thursday by 11:00 am.
- Laboratory results are available twice a week on Tuesdays and Fridays.
- Special laboratory runs (i.e. not Tues/Fri) are available if needed (i.e. for a newborn or following a holiday).

Diet Records

- Initially, diet records are kept on a daily basis. Exact amounts of what your child eats or drinks needs to be recorded.
- A complete 3 day diet record should accompany all blood levels.

THE CLINIC ROUTINE

Lengths and Weights

- Initially, lengths and weights are done twice a week. With age the frequency decreases. During growth spurts or overt changes in appetite a request for a length /weight might be made on an “as need” basis.

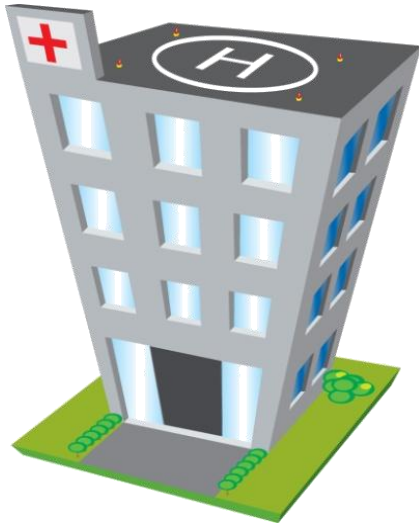
Appointments

- Patients are seen at 2 months, 4 months, 6 months, 9 months and at 1 year of age. they are then seen every 4 months until 3 years of age when they are then seen every 6 months until adulthood.
- At 3 years of age, they will begin group education following an individual clinic visit.
- Clinic appointments are on Monday and Thursday mornings. Plan on being in clinic from 9:00 am until 12 noon.
- During your clinic appointment you will see the metabolic nutritionist, the physician, a nurse or a genetic counseling student. This is a teaching hospital which means that your clinic appointments will continually have new faces popping in. They will be in the form of medical students, residents, fellows, genetic counseling students, dietetic interns and some visiting faculty from other institutions.
- You are always encouraged to interact with other families. As your child grows you will be formally placed in groups and given an opportunity to share ideas, questions and suggestions with other parents that have a child with the same diagnosis.
- Your children will have an opportunity to share with their peers in a safe learning environment.

Clinic Staffs Contact Numbers

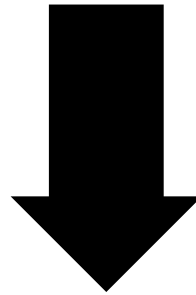
- There is a hospital number that you may call to reach a physician on call 24 hours a day.
- Emergencies are rare in PKU. Please remember that most emergencies should be directed towards your Primary Care Provider.

WHAT LEADS YOU TO US...



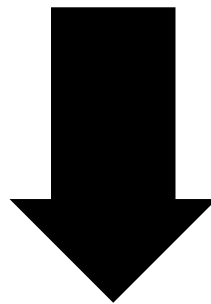
NEWBORN SCREENING

Newborn screen is positive for phenylketonuria (PKU)
Baby's phenylalanine level is high.
Baby's level needs to be rechecked.



LABORATORY TESTING

Secondary phenylalanine level is also high.
Confirmation that baby has PKU.
Baby should be seen at the metabolic clinic.



METABOLIC CLINIC

We are here to help you and answer your questions:

“What is phenylalanine?”

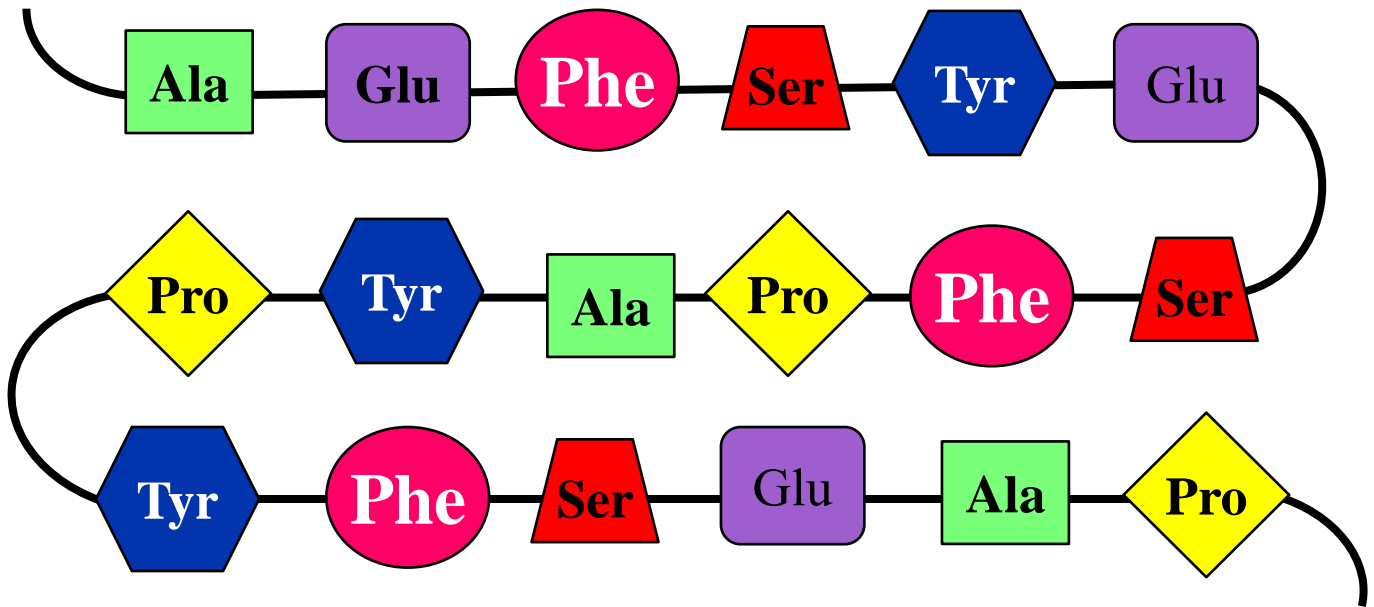
“What is PKU and hyperphenylalaninemia?”

“What causes it and how can it be treated?”



PROTEIN

All protein, including the protein in food we eat and the protein that makes up our body, consists of many amino acids hooked together much like beads on a string.



In this illustration each amino acid bead is a different shape and color.

The pink circles represent phenylalanine.



Phenylalanine is one of the amino acids that is present in protein. In total, there are twenty two different amino acids.

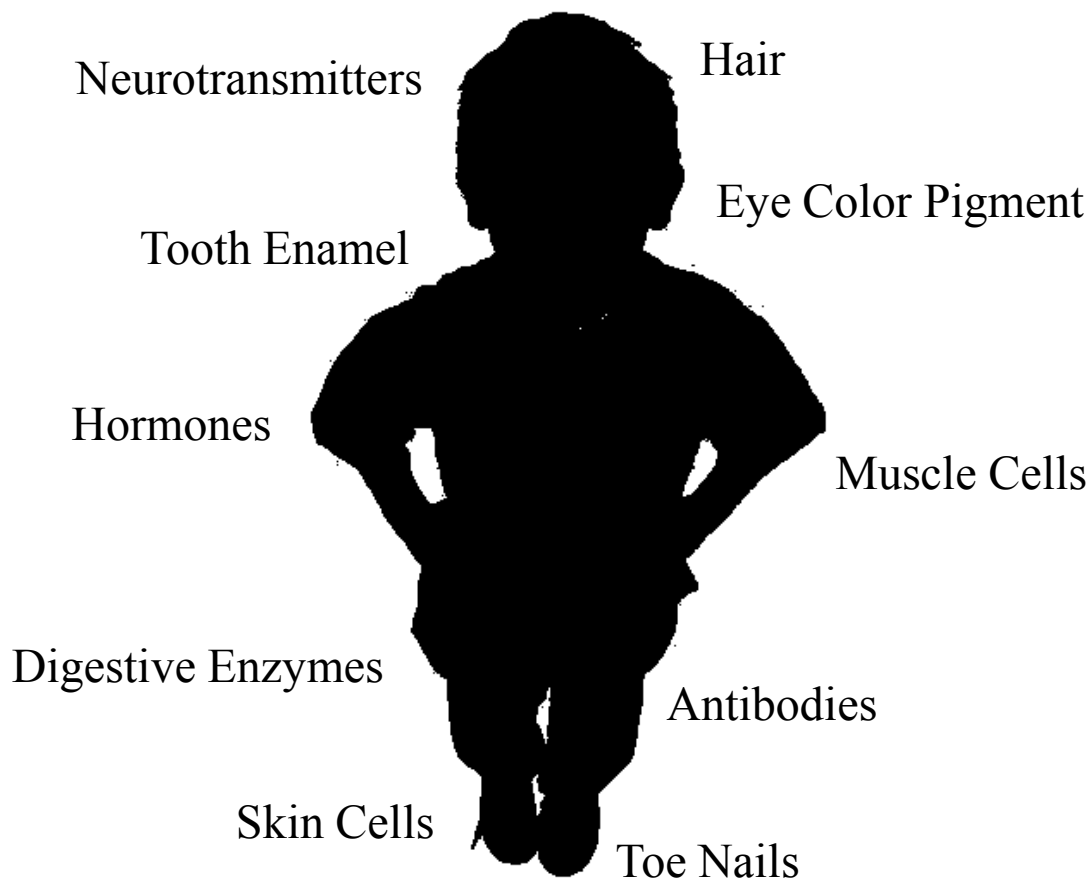
*We often shorten the word “*Phenylalanine*” to “*Phe.*”

Why Do We Need Phenylalanine?

Phenylalanine is one of many essential nutrients.

It is used by the body to make cells and other materials which are necessary for the body to work properly.

OUR BODIES REQUIRE A CERTAIN AMOUNT OF PHENYLALANINE TO MAKE:

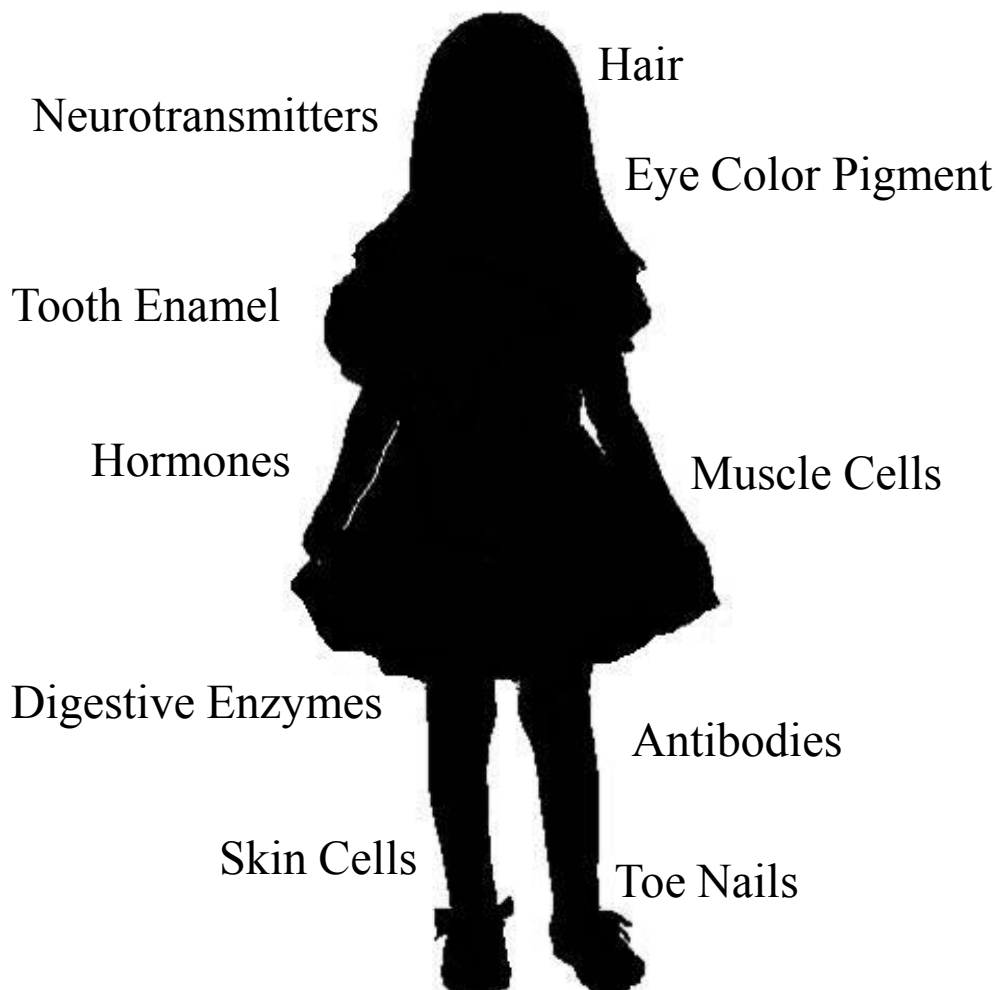


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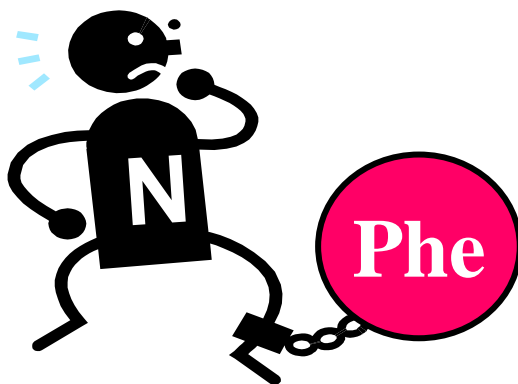
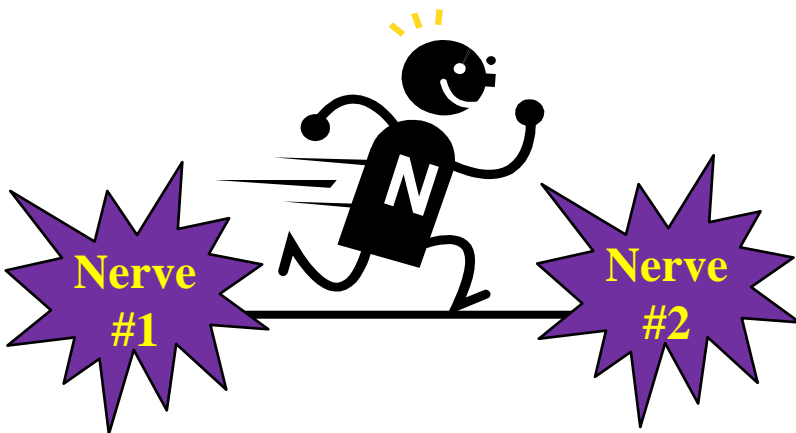
OUR BODIES REQUIRE A CERTAIN AMOUNT OF PHENYLALANINE TO MAKE:



Mr. Neurotransmitter



Mr. Neurotransmitter



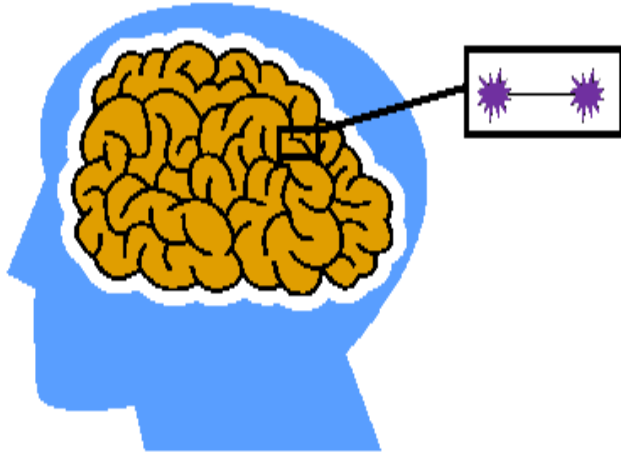
The cells in our brain make chemicals called neurotransmitters.

Neurotransmitters transmit messages from one nerve cell to another within our brain.

The body needs a certain amount of phenylalanine to make these chemicals.

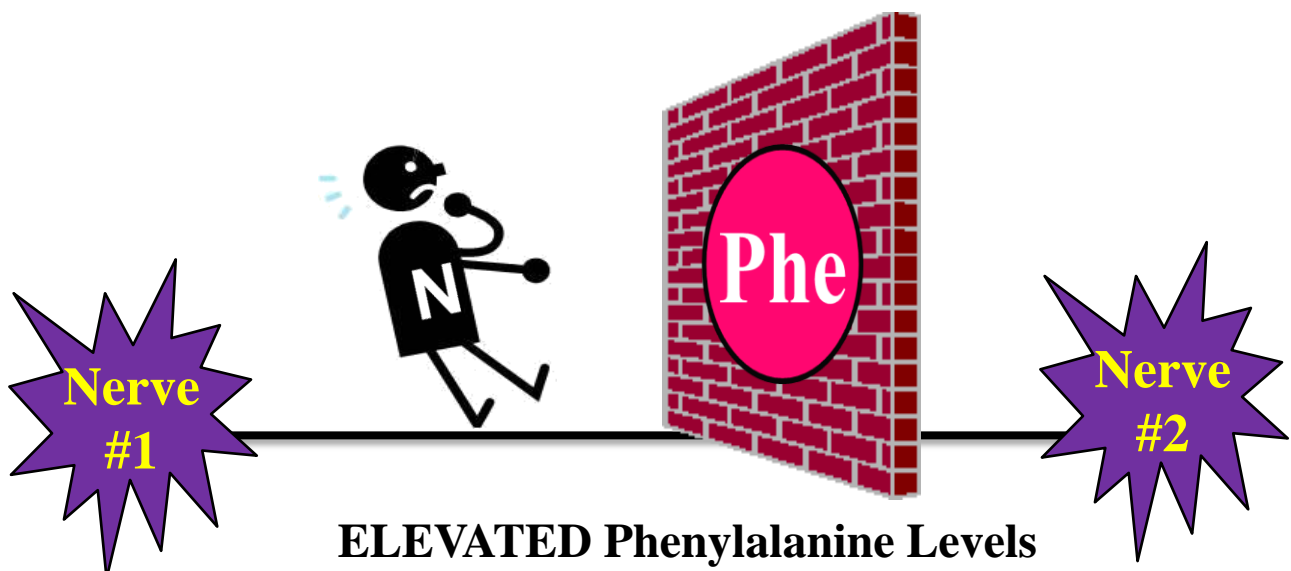
Too much phenylalanine and not enough tyrosine interferes with the formation and performance of neurotransmitters.

ALTERED NEUROCHEMISTRY

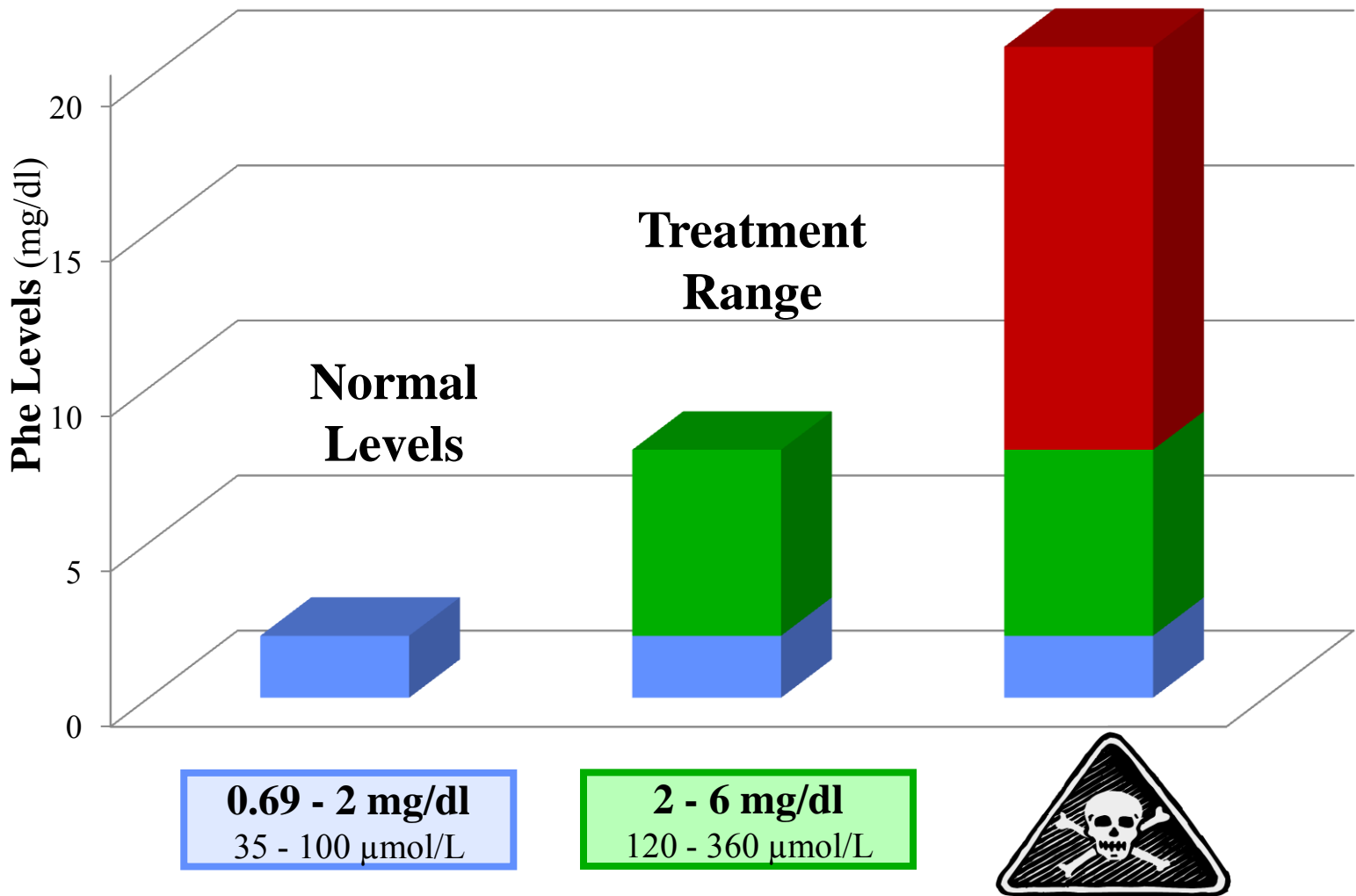


When phenylalanine levels are in treatment range, neurotransmitters can deliver clear messages.

When phenylalanine levels are **ELEVATED**, neurotransmitters **CANNOT** deliver clear messages.



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 \rightarrow *phenylalanine* \rightarrow *in the blood*

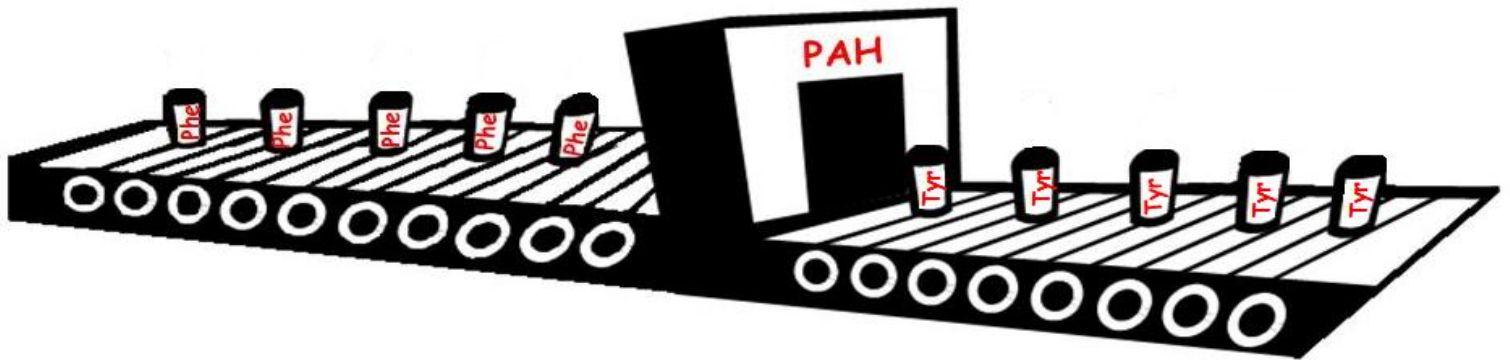
Phenylketonuria (PKU)

[-----phenylketon-----][-----uria-----]
phenylketone \rightarrow *in the urine*

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

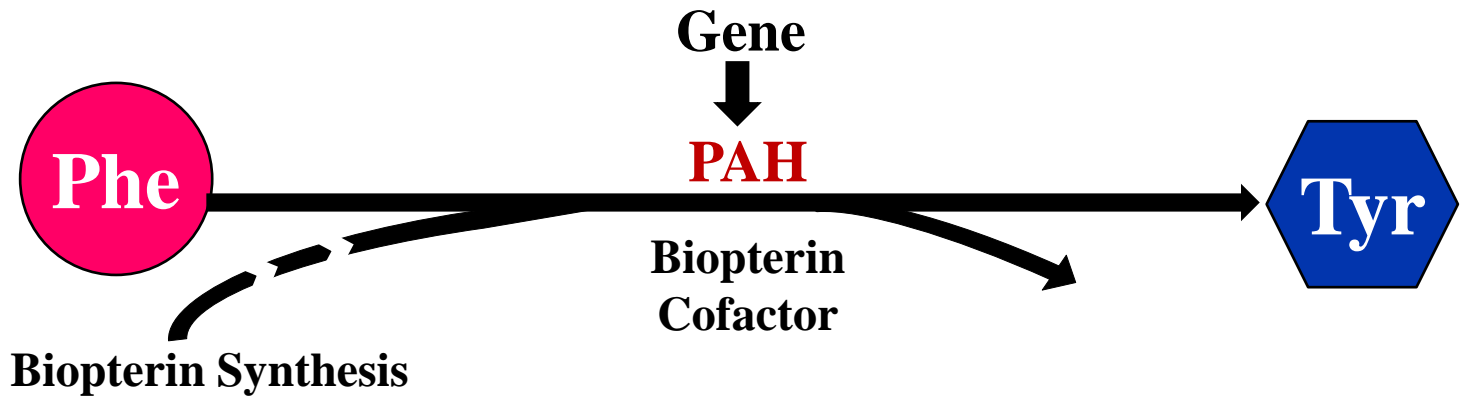
Pathways

Phe = Phenylalanine

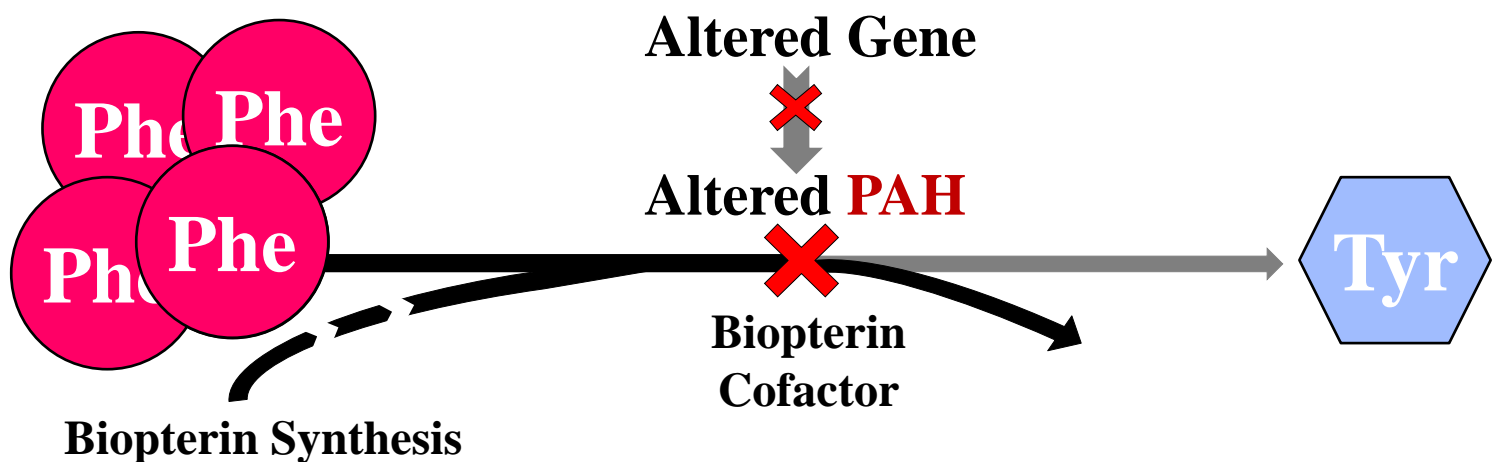
PAH = Phenylalanine Hydroxylase

Tyr = Tyrosine

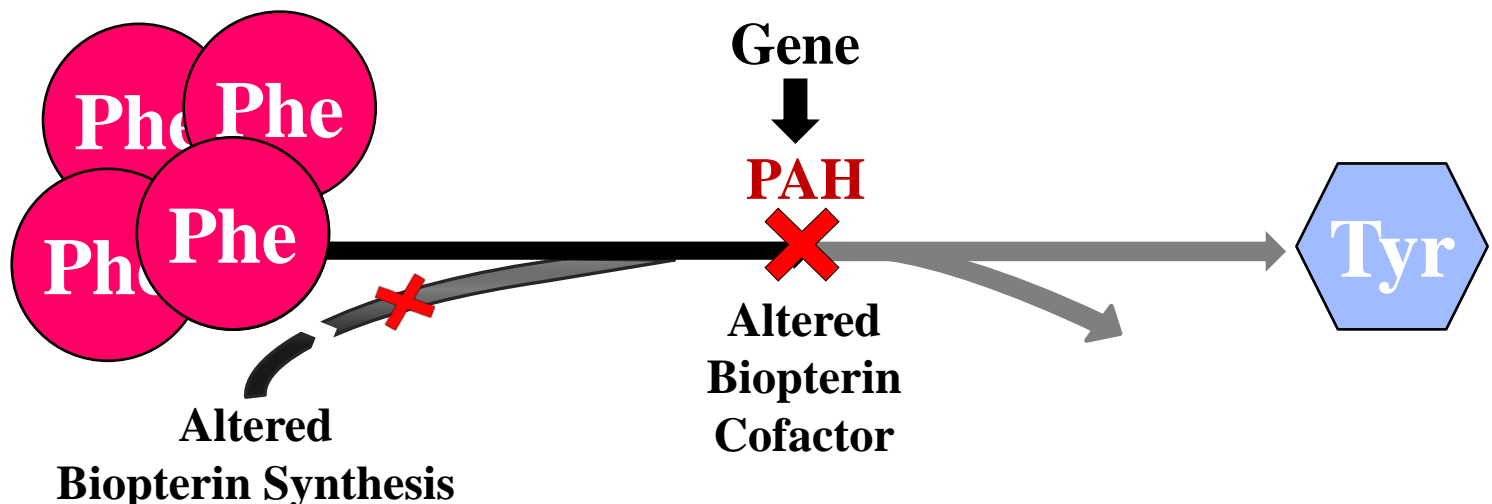
Normal Phenylalanine Pathway



Classic PKU Pathway



Biopterin Cofactor Defect Pathway



DNA BLUEPRINT



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

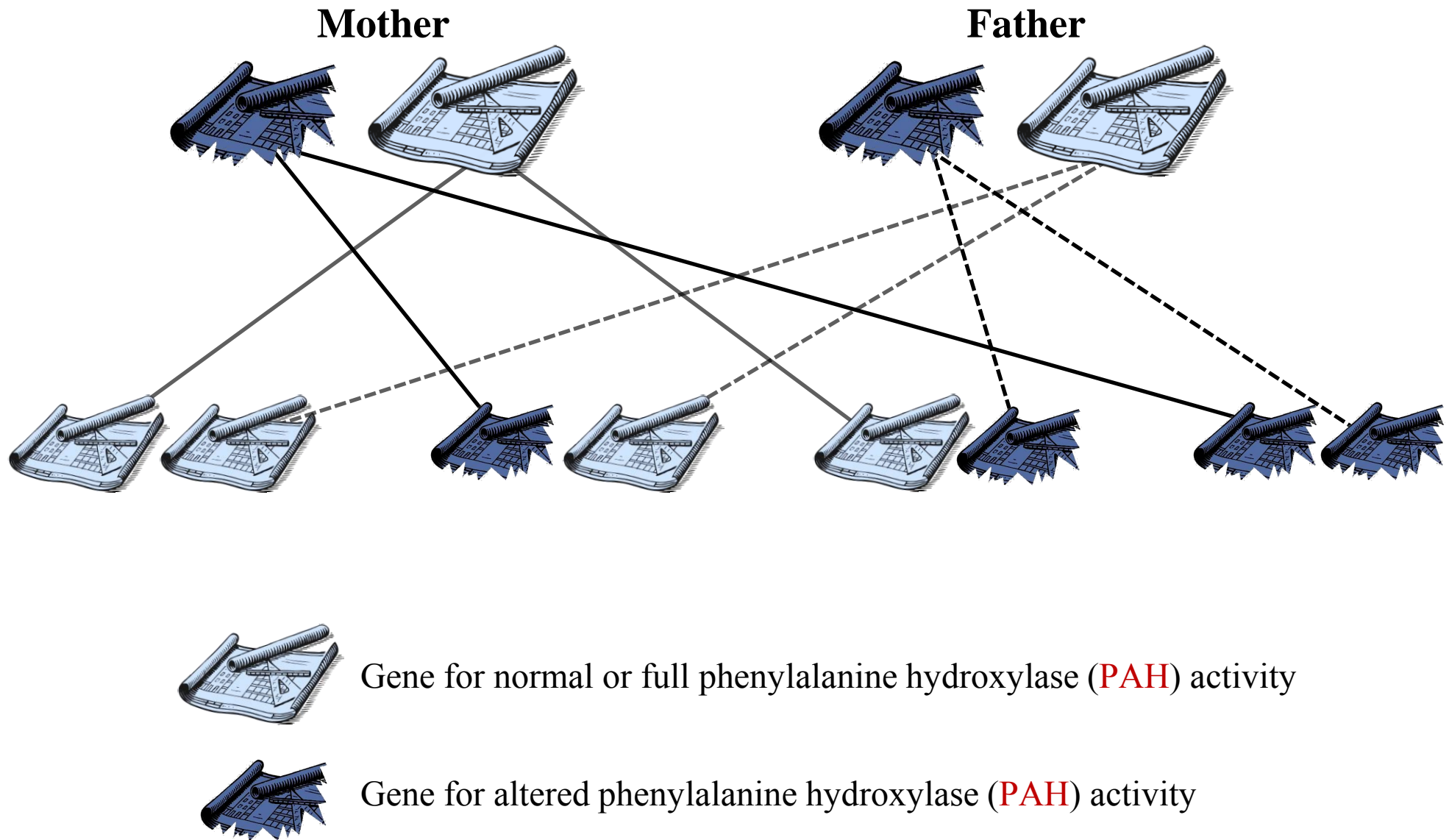


Instructions have an error.
Phenylalanine hydroxylase
(PAH) is made, however it
does not work as well.



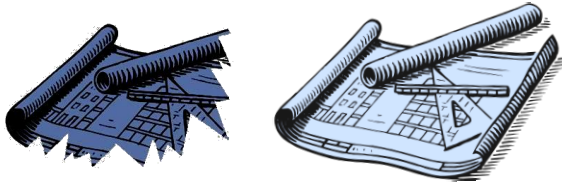
A significant amount of
instructions are missing. No
phenylalanine hydroxylase
(PAH) can be made.

AUTOSOMAL RECESSIVE INHERITANCE

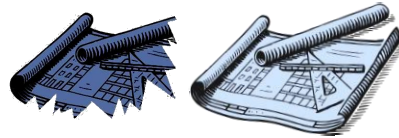
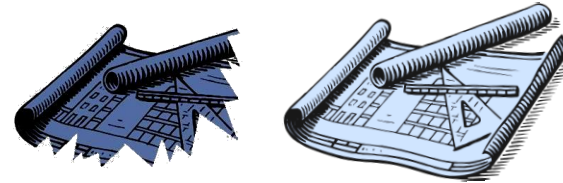


AUTOSOMAL RECESSIVE INHERITANCE

Mother



Father



Gene for normal or full phenylalanine hydroxylase (**PAH**) activity



Gene for altered phenylalanine hydroxylase (**PAH**) activity

AUTOSOMAL RECESSIVE INHERITANCE

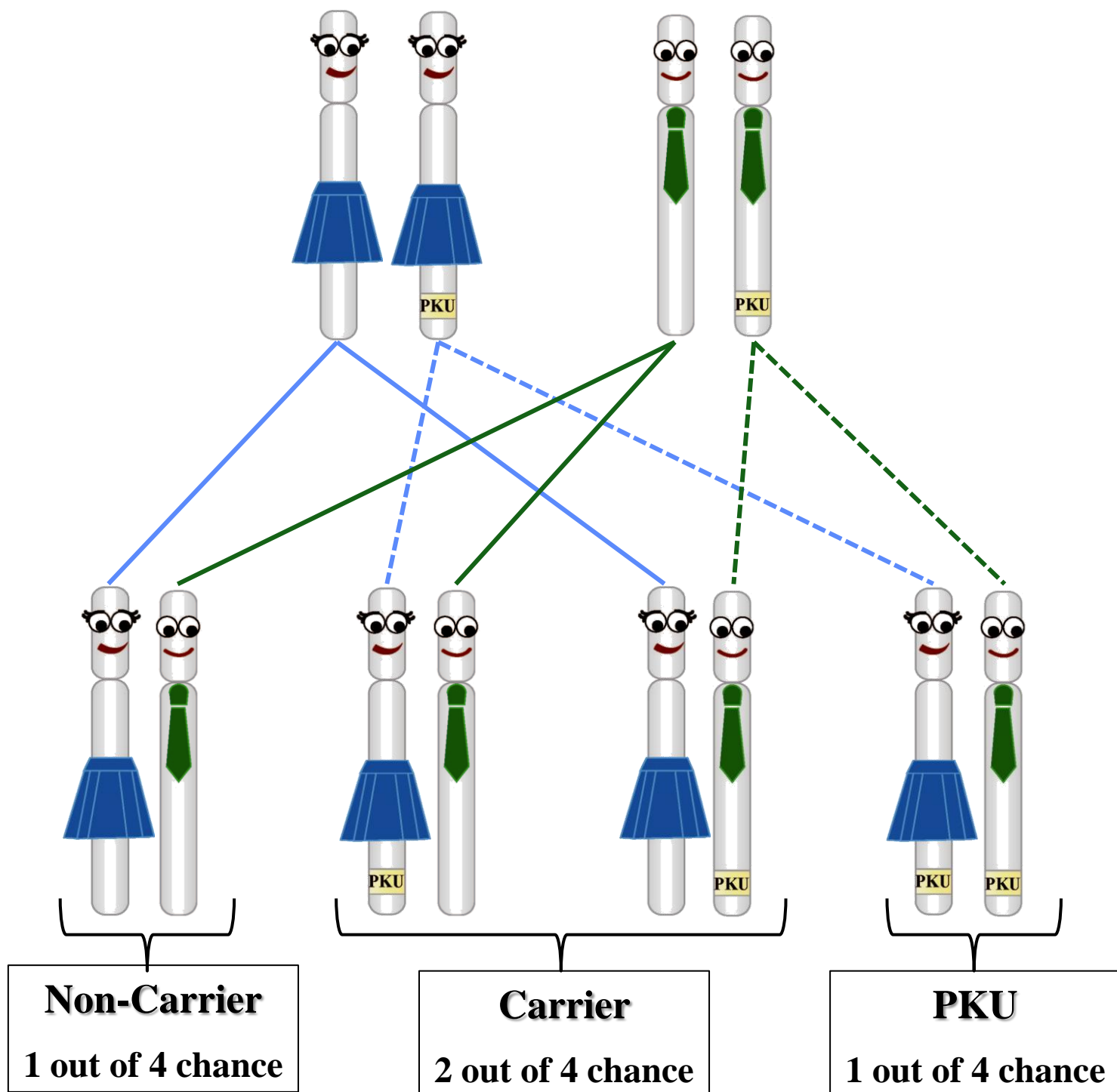
Chromosome #12 and the PKU Gene

Our genes are organized on our body on Chromosomes.

Mr. and Mrs. Chromosome are both carriers of PKU.

See the chances of having PKU based on Autosomal Recessive Inheritance.

Mrs. Chromosome Mr. Chromosome



AUTOSOMAL RECESSIVE INHERITANCE

Chromosome #12 and the PKU Gene

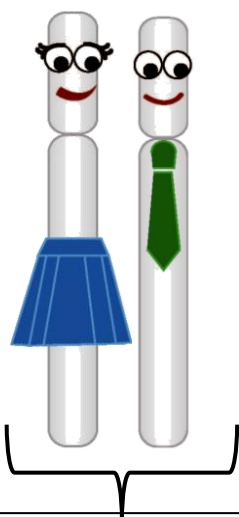
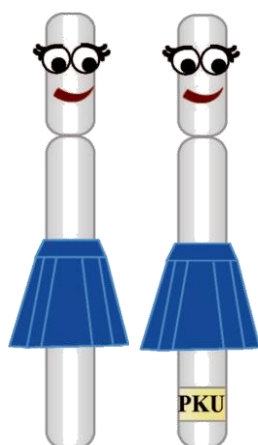
Our genes are organized on our body on Chromosomes.

Mr. and Mrs. Chromosome are both carriers of PKU.

See the chances of having PKU based on Autosomal Recessive Inheritance.

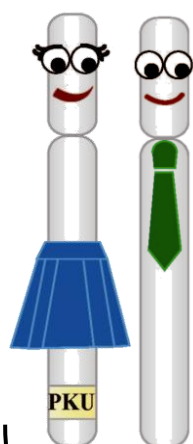
Mrs. Chromosome

Mr. Chromosome



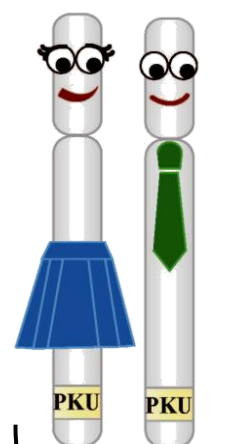
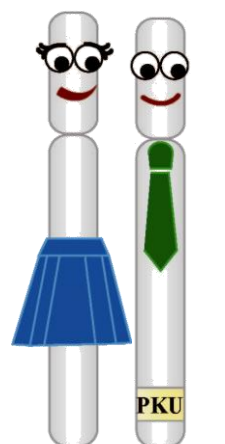
Non-Carrier

1 out of 4 chance



Carrier

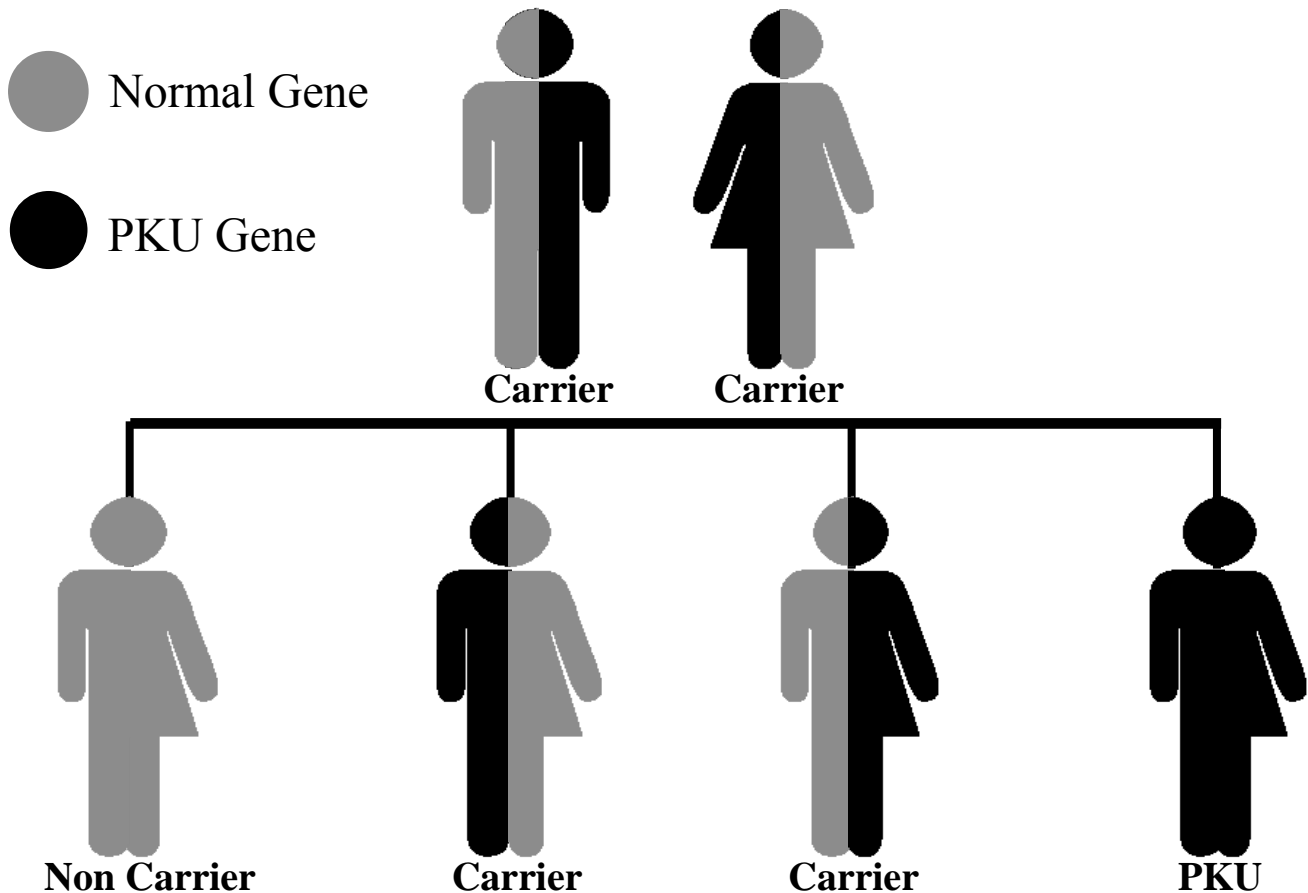
2 out of 4 chance



PKU

1 out of 4 chance

AUTOSOMAL RECESSIVE INHERITANCE



WHAT DOES AUTOSOMAL RECESSIVE INHERITANCE REALLY MEAN?

Autosomal recessive disorders are passed to a child through both parents' genes.

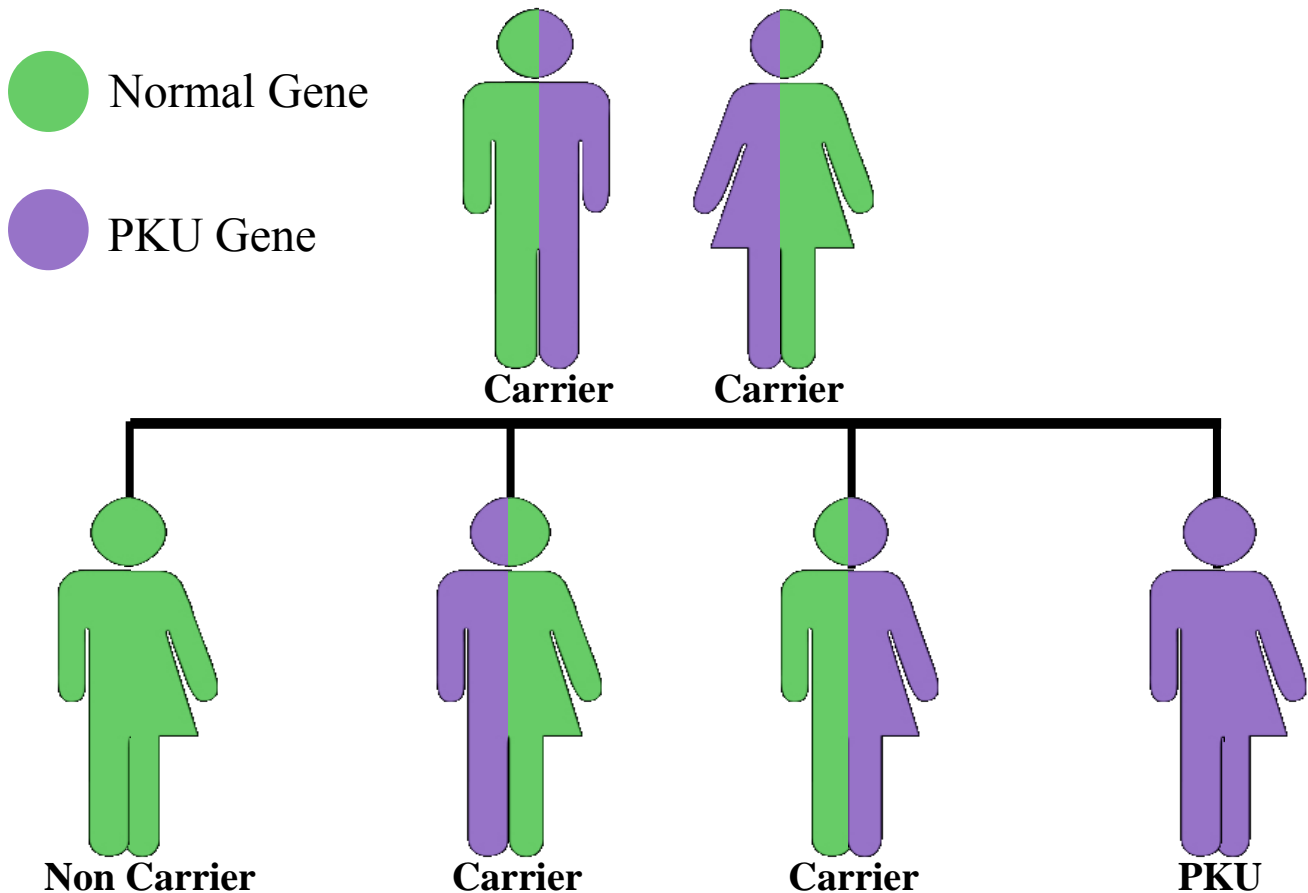
If both parents carry the PKU gene, there is a:

25% chance that the PKU gene will not be passed; the child will NOT be a carrier and will NOT have PKU.

50% chance that the PKU gene will be passed from one parent; the child will be a PKU carrier.

25% chance that the PKU gene will be passed from both parents, and the child will have PKU.

AUTOSOMAL RECESSIVE INHERITANCE



WHAT DOES AUTOSOMAL RECESSIVE INHERITANCE REALLY MEAN?

Autosomal recessive disorders are passed to a child through both parents' genes.

If both parents carry the PKU gene, there is a:

25% chance that the PKU gene will not be passed; the child will NOT be a carrier and will NOT have PKU.

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25% chance that the PKU gene will be passed from both parents, and the child will have PKU.

QUANTITATIVE TESTING FOR FUTURE SIBLINGS

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

[Date]

[Recipient's address]

RE:

DOB:

[Hospital]#:

Dear [PCP]:

We are writing regarding the expected child of Mr. & Mrs. [name]. As you know, Mr. & Mrs. [name] have a child with hyperphenylalaninemia (traditionally referred to as PKU). They therefore are presumed to be carriers for hyperphenylalaninemia and have a 25% risk with each pregnancy for having a child with hyperphenylalaninemia. Due to this increased risk, we do recommend quantitative testing on any newborn child they might have. We understand that [mother's name] is due [on or in date]; therefore, we are sending you and [mother's name] our recommendations for testing the baby.

We recommend that serum for quantitative phenylalanine and tyrosine levels be drawn at 72 hours of age and sent to [laboratory]. If these results are in the normal range, repeat testing should be done at two weeks of age in the same manner. **We want to stress that the quantitative testing is in addition to the regular newborn screen, as the newborn screen tests for other disorders in addition to hyperphenylalaninemia.**

The samples [detail specimen requirements, laboratory's address and shipping requirements]. We recommend the facility collecting and sending the specimen contact the [receiving laboratory] at [receiving laboratory telephone number] to confirm the above instructions. In addition, we suggest that you alert us to the baby's birth so that we can assist in the tracking of results, so that, in the event the baby is affected, diagnosis is made and treatment started promptly.

In the meantime while we are awaiting results of the quantitative phenylalanine and tyrosine levels are pending, the newborn should be on a normal, unrestricted diet. If the newborn is found to have elevated phenylalanine levels consistent with hyperphenylalaninemia, diet will be initiated at that time.

Please feel free to call us if you have any questions at [clinic telephone number].

Sincerely,

cc [mother's name]

OUTCOMES OF TREATMENT

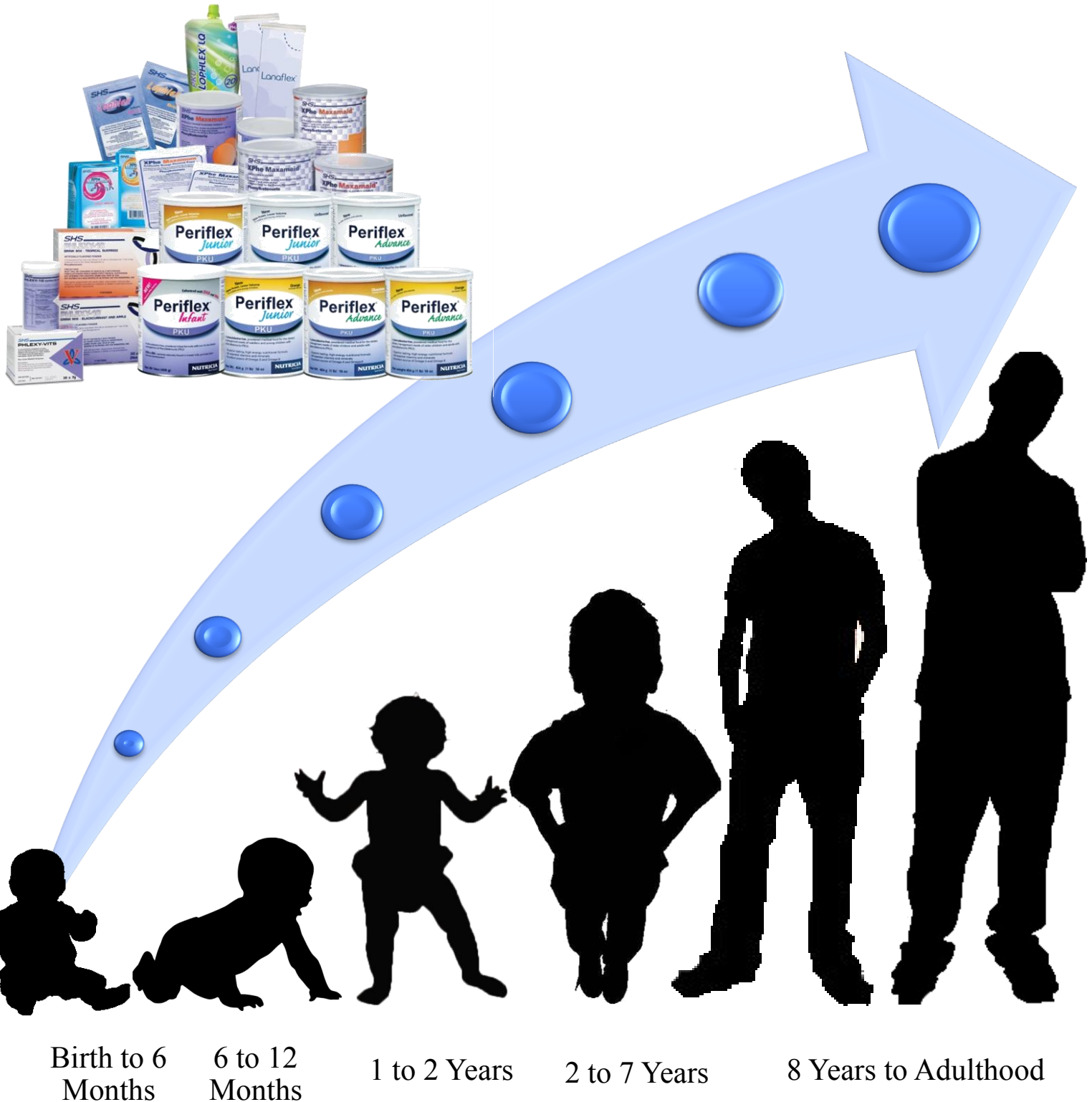


OUTCOMES OF TREATMENT



PRINCIPLES OF DIET PRESCRIPTION

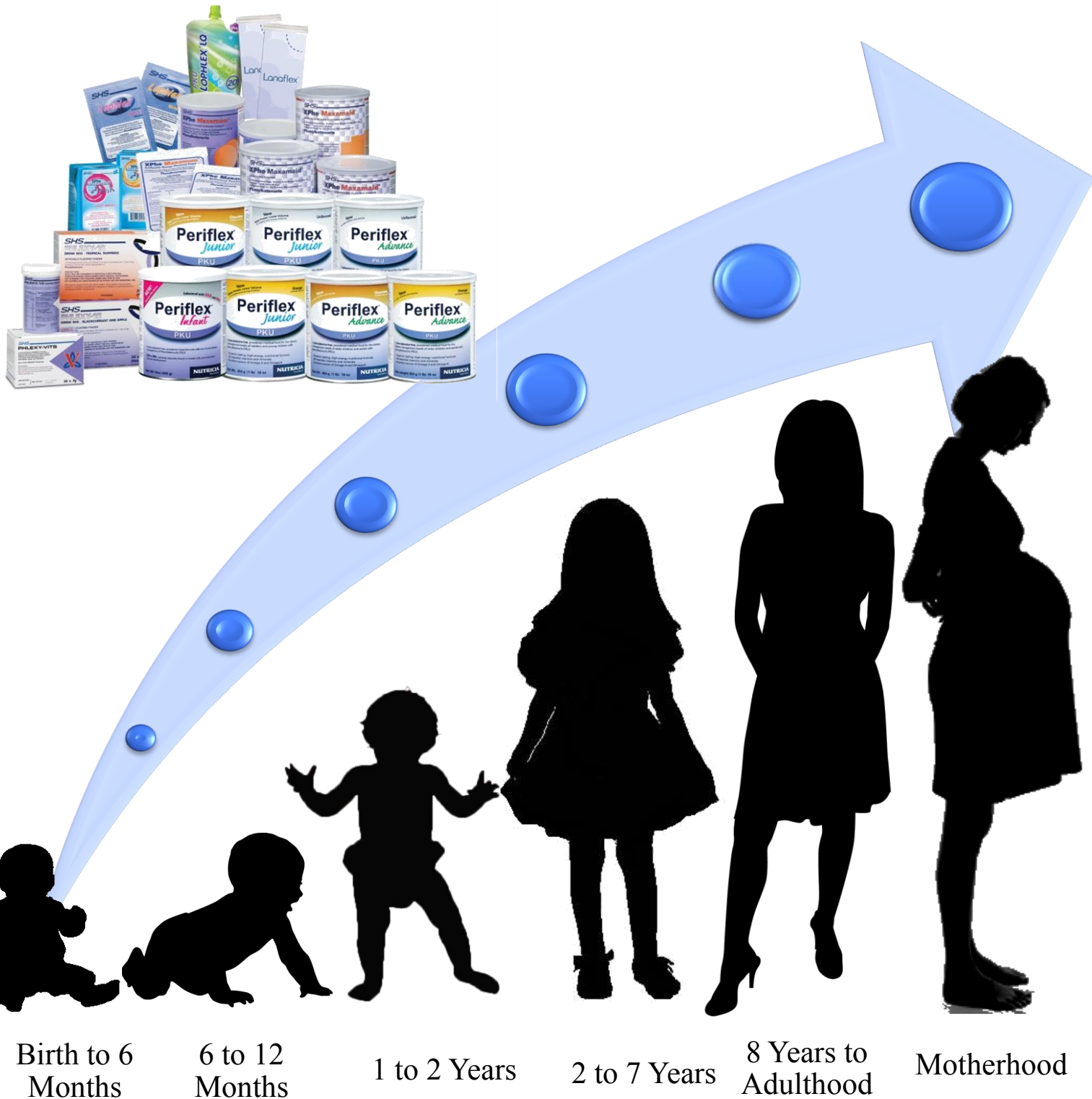
Throughout a Lifecycle



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

PRINCIPLES OF DIET PRESCRIPTION

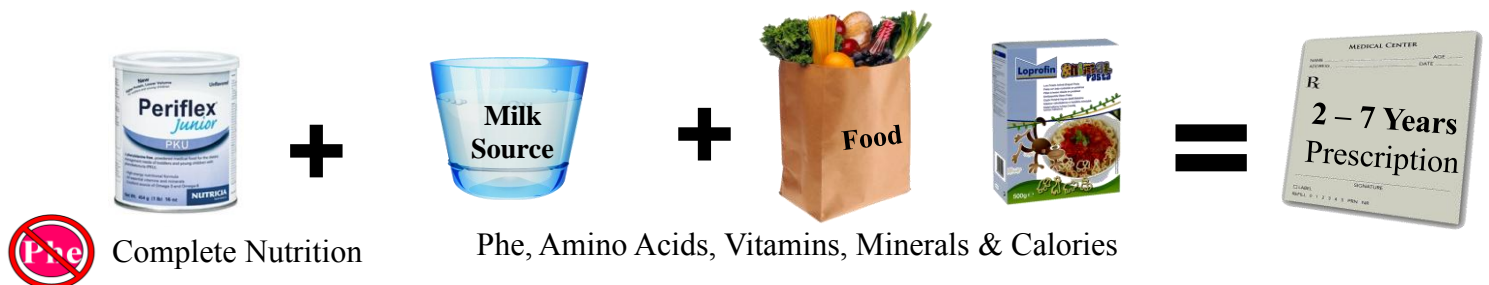
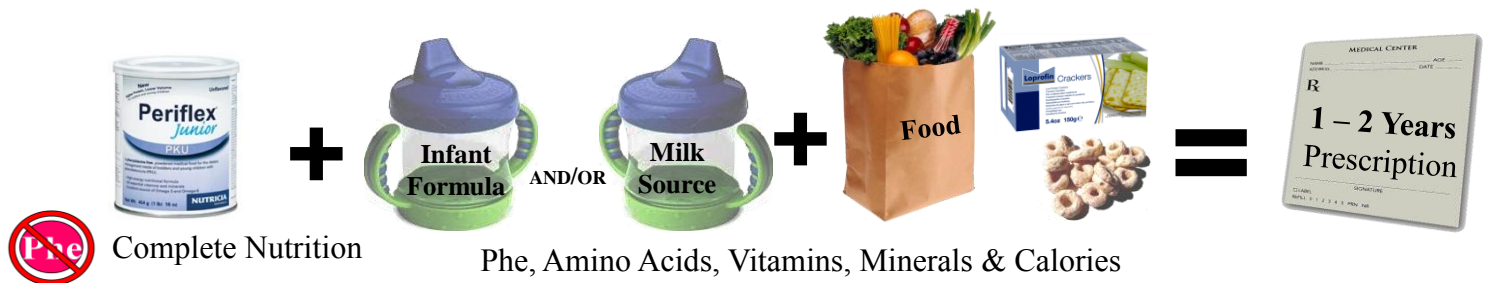
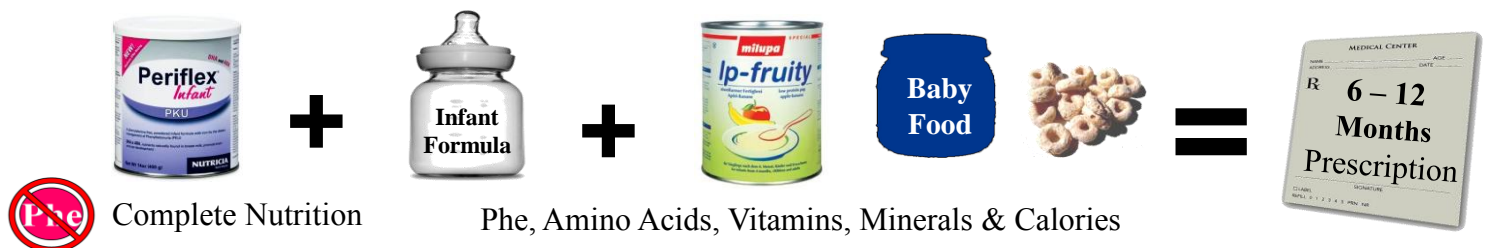
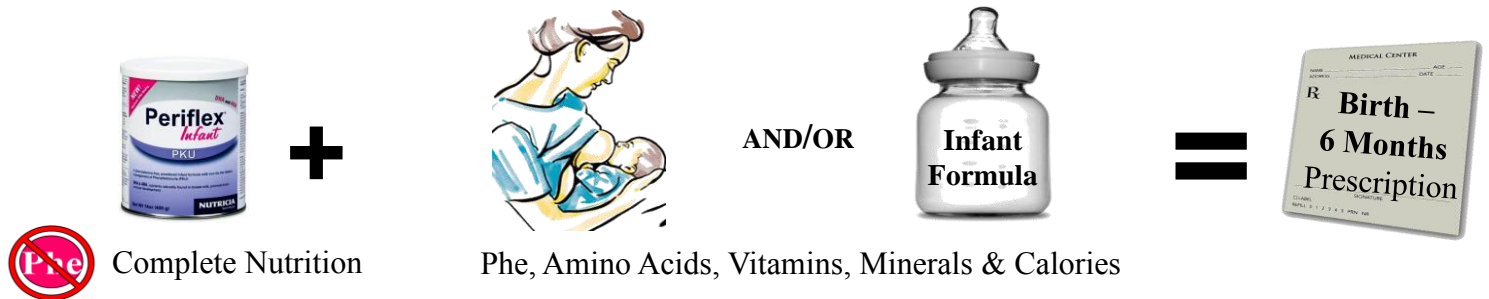
Throughout a Lifecycle



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

PRINCIPLES OF DIET PRESCRIPTION

Birth to 7 Years Old

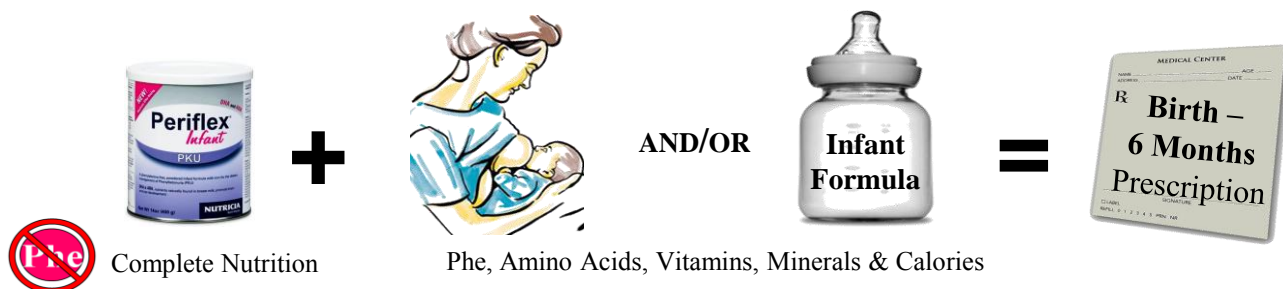


If an infant or child eats only what is allowed on a low phenylalanine diet without medical food, they would be malnourished in protein, calories, essential vitamins and minerals. The **medical food** provides most of the protein needs and daily requirements of essential vitamins and minerals.



DAILY DIET PRESCRIPTION

Birth to 6 Months



Name: _____ DOB: _____

Step 1: Add 1-2 ounces of water to hand shaker.

Step 2: Measure out _____ grams of _____, then add to hand shaker.
of grams Medical Food i.e.: Periflex

Step 3: Measure out _____ grams of _____, then add to hand shaker.
of grams Infant Formula or Milk Source

Step 4: Shake gently in hand shaker.

Step 5: Add water to make a total volume of _____ mL or _____ ounces.

Step 6: Shake vigorously for 10-15 seconds.

Step 7: Put formula into clean sterile formula bottles and refrigerate until use.

If Breastfeeding: Once medical food prescription has been completed, baby may breastfeed.

The above formula preparation is calculated for your child's 24 hour clock (a 24 hour period).
A new formula MUST be prepared at the start of each new 24 hour clock.

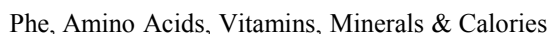
If the preparation is completed before the 24 hour clock is over and your child is still hungry, offer **extra** medical food as follows:

Measure out ____ grams of _____, add ____ ounces of water to make a total of ____ ounces.

Example: Measure out 10 grams of Periflex Infant, add 2 ounces of water to make a total of 2 ounces

Medical food may provide complete nutrition without any Phe.

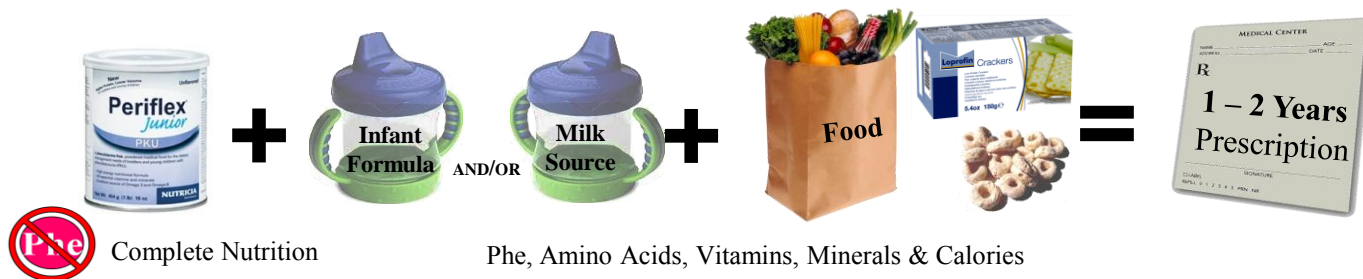
Breast milk and/or infant formula provide additional amino acids, vitamins, minerals, and calories.





DAILY DIET PRESCRIPTION

1 to 2 Years Old



Name: _____

DOB: _____

Medical Food/Formula:

Step 1: Measure Medical Food/Formula:

_____ grams of _____ . Add to hand shaker.
of grams Medical Food

_____ grams of _____ . Add to hand shaker.
of grams Medical Food

Step 2: Measure out _____ grams of _____. Add to hand shaker.
of grams Infant Formula or Milk Source

Step 3: Add water to make a total volume of _____ ounces or _____ ml

Step 4: Shake vigorously for 10-15 seconds.

Step 5: Put formula into a clean container and refrigerate until use.

Regular and Low Protein Food:

mg Phe gm Protein # Exchanges

Circle One

Medical food may provide complete nutrition without any Phe.

Formula with a milk source, cereal, baby food, finger foods, low protein and new solid foods provide additional amino acids, vitamins, minerals, and calories.

DAILY DIET PRESCRIPTION

2 to 7 Years Old



Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

Name: _____ DOB: _____

Medical Food/Formula:

Step 1: Measure Medical Food/Formula:

_____ grams of _____ . Add to hand shaker.
of grams Medical Food

_____ grams of _____ . Add to hand shaker.
of grams Medical Food

Step 2: Measure out _____ grams of _____ . Add to hand shaker.
of grams Milk Source

Step 3: Add water to make a total volume of _____ ounces or _____ ml

Step 4: Shake vigorously for 10-15 seconds.

Step 5: Put formula into a clean container and refrigerate until use.

Regular and Low Protein Food:

mg Phe gm Protein # Exchanges

Circle One

Medical food may provide complete nutrition without any Phe.
Formula with a milk source, in addition to low protein and other foods provide additional amino acids, vitamins, minerals, and calories.

FORMULA PREPARATION

DATE: _____

Name: _____ DOB: _____

Step 1: Add 1-2 ounces of water to hand shaker.

Step 2: Measure out _____ grams of _____, then add to hand shaker.
of grams Medical Food i.e.: Periflex

Step 3: Measure out _____ grams of _____, then add to hand shaker.
of grams Infant Formula or Milk Source

Step 4: Shake gently in hand shaker.

Step 5: Add water to make a total volume of _____ mL or _____ ounces.

Step 6: Shake vigorously for 10-15 seconds.

Step 7: Put formula into clean sterile formula bottles and refrigerate until use.

If Breastfeeding: Once medical food prescription has been completed, baby may breastfeed.

The above formula preparation is calculated for your child's 24 hour clock (a 24 hour period).
A new formula **MUST** be prepared at the start of each new 24 hour clock.

If the preparation is completed before the 24 hour clock is over and your child is still hungry, offer **extra** medical food as follows:

Measure out _____ grams of _____, add _____ ounces of water to make a total of _____ ounces.

Example: Measure out 10 grams of Periflex Infant, add 2 ounces of water to make a total of 2 ounces

Call Our Clinic So We Can Make A Diet Change If:

- If you are continually making extra formula because your child is hungry.
- If you are consistently dumping formula because your child can not finish their prescription within their 24 hour clock.

Clinic Contact Info: _____

24 HOUR DIET DIARY

Name: _____

Dates Covered: _____

Date of Birth: _____

Age: _____

Weight/Length: _____

Medical Food/Formula	Amount

Kuvan:
Tyrosine:
Multivitamin:
Other:

Add water to make _____ ml (_____ fl. oz.)

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)
Totals					

Patient's appetite today was: ___ Better than usual ___ Usual ___ Poor

Patient was ill today: ___ No ___ Yes, describe: _____

Medication Required? ___ No ___ Yes (Name and amount of prescription): _____

Did patient regurgitate food or formula? ___ Yes ___ No Did patient have diarrhea? ___ Yes ___ No

Additional Notes: _____

PKU MEDICAL FOOD/FORMULA

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

RE: _____

BD : _____

TCH: _____

To Whom It May Concern:

This 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, designed to provide just enough phenylalanine for growth, development, and physiologic needs; while keeping blood phenylalanine levels in a narrow acceptable range. Children with PKU who are not begun 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 age, and maintained and monitored over time.

Indefinite continuation of dietary management is recommended to all patients with PKU. We also recommend reinstitution of dietary restriction to all patients in whom diet was discontinued in childhood in the 1970's, when that was the usual practice. These recommendations are based on evidence (R Gassio, et al., *Pediatr Neurol* 2005;33:267-271, V Leuzzi et al, *Pediatr Neurol* 2005;33:267-271 and SE Waisbren et al., Phenylalanine blood levels and clinical outcomes in phenylketonuria:..., *Mol. Genet. Metab.* (2007), doi:10.1016/j.ymgme.2007.05.006), indicating that there is a decline in average IQ and executive functioning skills, which may lead to development of difficulties in school and job performance with diet discontinuation and/or poor diet adherence. Adult women with PKU must have dietary control prior to and during pregnancy to prevent adverse effects. It has been known since the 1950's that uncontrolled PKU in pregnancy causes severe mental retardation and birth defects (severe heart and intestinal defects are most common). The Maternal PKU Collaborative Study demonstrated that early control of phenylalanine levels coupled with attention to the multiple problems of nutrition in pregnancy on a phenylalanine-restricted diet may result in the birth of completely healthy children. The following studies detail the complications that may be seen with poor diet adherence but prevented if maintaining an appropriate diet during pregnancy: WB Hanley et al, *Eur J Pediatr* 1996;155 S:S169-72, B Rouse et al, *Am J Med Genet* 1997; 69(1):89-95 and SE Waisbren et al, *JAMA* 2000; 283(6) 756-62.

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 medical 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.

These medical foods are an artificial replacement of the normal protein-containing foods that we all require for growth and for 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.

PKU MEDICAL FOOD/FORMULA

Medical foods fall in 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.

Recognizing that these substances are the major source of essential amino acids necessary for life and constitute the principal medical treatment for children with PKU; several health insurance companies and health plans including Blue Cross/Blue Shield, CHAMPUS, AETNA, Prudential, Travelers, Kaiser Permanent of California and Colorado. The Department of Agriculture’s WIC program, and Fireman’s Fund American Life Insurance Company offer coverage for these items. Also, the effective cost-benefit ratio in the screening and nutritional management of these individuals has clearly been shown by any number of individuals and groups including the GAO.

We request that you approve coverage for medical food for management of PKU for _____. We would prescribe the amount of specific medical food and of phenylalanine from natural foods; and monitor _____’s clinical status using laboratory studies (phenylalanine nutrients) and clinical evaluations. The medical foods could be dispensed through The Children’s Hospital or any other pharmacy; laboratory studies would utilize The Children’s Hospital Laboratory for phenylalanine levels and The Children’s Hospital or other laboratory for other needed surveillance tests. If you have any questions regarding any of this information, please contact us at (303) 724.2338.

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 PKU**

Nutricia

- Add-Ins
- Lanaflex
- Lophlex
- Lophlex LQ
- Milupa PKU 2
- Milupa PKU 3
- Periflex Advance
- Periflex Infant
- Periflex Junior
- Phlexy 10 System
- XPhe Maxamaid
- XPhe Maxamum
- XPhe Maxamum Drink

Abbott Nutrition

- Phenex-1
- Phenex-2

Applied Nutrition

- PhenylAde 40
- PhenylAde 60
- PhenylAde AA Bar
- PhenylAde AA Blend
- PhenylAde Essential Drink Mix
- Phenylade MTE AA Blend

Cambrooke Foods

- Camino Pro
- Camino Sorbet Stix

Mead Johnson

- Phenyl Free 1
- Phenyl Free 2
- Phenyl Free 2 HP






Vitaflo

- PKU Coolers
- PKU Express
- PKU Gel

APPROPRIATE INFANT FEEDING PRACTICES

- ☑ Infants should be fed in a high chair. Another good position is to seat the baby in an upright position on the parent's lap. This helps to make the baby feel secure about this new feeding experience. The baby should always be checked to make sure that the food is being swallowed easily. The caregiver and infant should have good eye contact so that they can readily see each other.
- ☑ Solids should be fed from a spoon. Spoon feeding plays an important part in the development of the ability to self-feed. It also promotes the proper development of tongue muscles that are important for speech and allows the infant experience the taste and texture of foods.
- ☑ Each new food should be introduced one at a time with approximately 3-5 days in between each new item. This will allow the infant to become accustomed to new foods and provide an opportunity for parents to identify any one food that may cause an adverse reaction. If an adverse reaction occurs (rash, hives, vomiting, diarrhea) this food should be eliminated from the diet until a later date.
- ☑ Baby food jars should be washed before opening. Jar lids should make a popping sound when opened. If the "bubble" on top of the jar has already popped up, DO NOT feed that the food in that jar to the baby.
- ☑ New foods that are rejected by an infant should be offered at another time. Try offering your baby the same food another day or at another meal time.
- ☑ Acceptance of new foods can be encouraged by a positive attitude.
- ☑ It is important for the parent or caregiver to allow the baby to set the pace for a feeding by waiting until the baby indicates s/he is ready for another spoonful.
- ☑ It is not necessary for an infant to finish a bottle or solids. The baby is usually the best judge of how much to eat. An infant with hyperphenylalaninemia must finish all the metabolic prescription so the clinic relies on you to help us individualize the prescription in order to ensure that your infant can finish their formula and solids without overfeeding or force-feeding.

INAPPROPRIATE INFANT FEEDING PRACTICES

-  Solid foods should not be fed from a bottle.
-  Infants do not need salt, sweeteners, and seasonings, added to their bottles/food. Plain foods allow the infant to experience the individual tastes of foods. Do not offer honey or corn syrup; they may contain botulinum spores which may cause botulism (food poisoning) in infants.
-  Near the age of 6 months, babies begin to hold their own bottles. Babies should not be put to bed with a bottle because this will start a habit which may be difficult to break and which can lead to baby bottle tooth decay.
-  Do not feed the baby directly from the baby food jar. Instead food should be placed in a clean dish and total weight in grams recorded. The reason for this is two-fold. If the baby is fed directly from the jar the baby's saliva will enter the food, which can cause the food to spoil. In addition, food must be weighed before and after a meal in order to accurately calculate how much phenylalanine the baby is getting from food.
-  Do not use the microwave to warm foods as this can cause uneven heating. It is safer to heat baby's food using the conventional methods or serve it at room temperature.

SUGGESTED MEAL PATTERNS

6 - 12 MONTHS OLD

Months of Age	Early Morning	Mid-Morning	Noon	Mid-Afternoon	Evening	Bedtime
6 - 7	Formula ¹	Cereal ²	Formula	Formula	Cereal	Formula
7 - 8	Formula	Cereal Vegetables	Formula Vegetables	Formula	Cereal Vegetables Fruit	Formula
8 - 9	Formula	Cereal Fruit	Formula Vegetables Fruit	Formula	Combination Dinners ³ Vegetables Fruit	Formula
Introduce Finger Foods						
9 - 10	Formula	Cereal Fruit	Formula Combination Dinners Vegetables/Fruit	Formula	Cereal Vegetables Fruit	Formula
Introduce Low Protein Foods⁴						
10-12	Formula	Cereal Fruit	Formula Low Protein Food Vegetables/Fruit	Formula	Low Protein Food Vegetables Fruit	Formula

¹ **Formula:** medical food and infant formula (if mother is breastfeeding, schedule will need to be individualized) prepared according to formula prescription.

² **Infant cereals:** must be mixed with a medical food (Periflex Infant, 10gms of Periflex Infant, add water to make 2 oz.). As the baby gets used to eating the cereal, you can offer the cereal mixed with the metabolic formula prescription to reduce the drinking volume within the 24 hour clock.

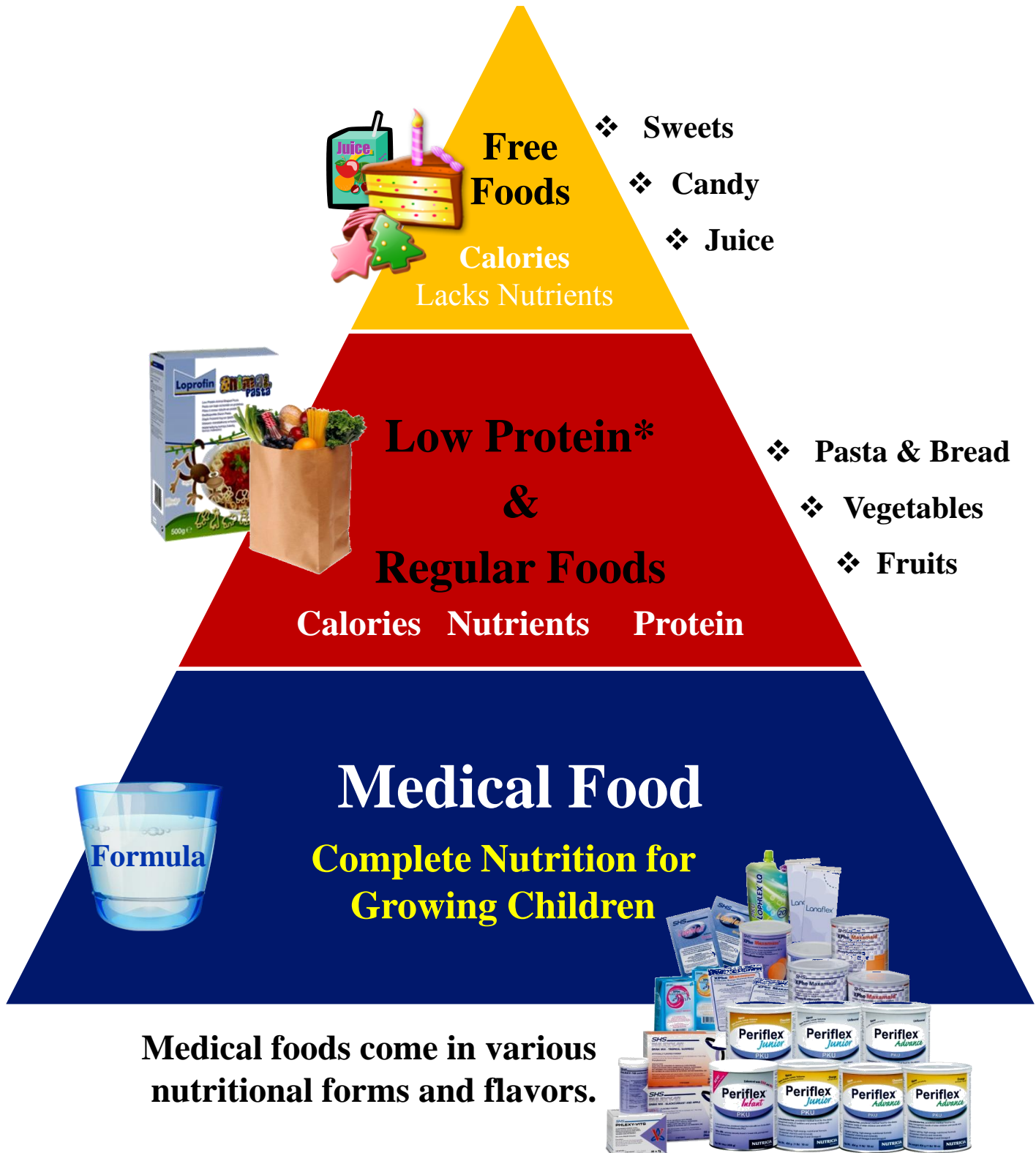
³ **Combination dinners:** commercially prepared 2nd and 3rd foods for growing babies are encouraged because of their low protein content and added texture.

⁴ **Low Protein Food:** low protein foods are introduced for satiety, texture and palette acceptance, and long term dietary compliance (pasta, crackers, cheese, bread and baking mix options).

Near the age of one year infants become interested in holding utensils and feedings themselves. They enjoy playing with spoons during meal or playtime. Babies gradually learn to get food on the spoon and the spoon to their mouth, although food is often spilled before it gets into their mouth. This mess is OK.

AVOID “Phenueurosis” (a fear regarding total phenylalanine intake from food). Despite our best efforts, we do help nurture this fear, but relax, we will make sure your baby gets all the Phe he/she needs.

CHOOSE YOUR FORMULA FIRST



* Nutricia North America -- Low Protein Products - www.shsna.com/pages/loprofin.htm

CHOOSE YOUR FOODS

Finger Foods For Infants With Metabolic Disorders



Finger foods should be firm enough to pick up, yet soft enough to chew, swallow, and digest.



Avoid small pieces of hard foods that may cause choking or gagging.



Use fresh, frozen, or canned foods but avoid foods high in salt or seasoning.



If you introduce a food and the baby does not like it, try serving it another day or at another mealtime.

CHOOSE YOUR FOODS

Babies 6 to 8 Months Old

When Do You Introduce Solid Foods?

The American Academy of Pediatrics (AAP) currently recommends gradually introducing solid foods when a baby is about 6 months old.

Is Your Baby Ready to Eat Solids?

- **Is your baby's tongue-thrust reflex gone or diminished?**

This reflex prevents infants from choking on foreign objects, but also causes them to push food out of their mouths. Ask your pediatrician.

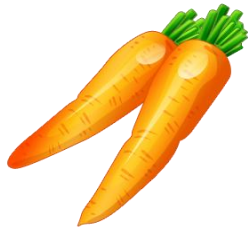
- **Can your baby support his/her own head?**

To eat solid food, an infant needs good head and neck control and should be able to sit up in a high chair.

- **Is your baby interested in food?**

A 6-month-old baby who stares and grabs at your food at dinnertime is clearly ready for some variety in the food department.

Remember “Yes” and “No” Foods.



- | | |
|-----------------------------------|-------------------------------|
| ❖ Applesauce | ❖ Milupa Ip cereal* /Cheerios |
| ❖ Very Small Pieces of Fruit | ❖ Soft Cooked Vegetables |
| ❖ Banana Slices | ❖ Carrot Slices |
| ❖ Grapes: peeled and cut quarters | ❖ Squash |
| | ❖ Peas: pop skins |

Always discuss the addition of new foods with your metabolic dietitian.

* Nutricia North America -- Low Protein Products - www.shsna.com/pages/loprofin.htm

CHOOSE YOUR FOODS

9 to 12 Months Old

Fruits

Selection & Preparation

Fresh:

- Wash thoroughly.
- Remove skins and seeds.
- Cut into small, bite size pieces or thin sticks (i.e. quarter grapes)
- At one year, include unpeeled fruits, berries, pears, and nectarines.

Frozen/Canned:

- Look for fruits packed in their own juices.
- Avoid fruits canned in heavy syrup.
- Frozen fruits are soothing to teething gums.

Dried:

- Buy pitted or seedless prunes, apples, apricots, peaches, and dates.
- Avoid raisins, which can cause choking.



❖ Apple

❖ Apricot

❖ Avocado

❖ Banana

❖ Berry

❖ Grape

❖ Kiwi

❖ Mango

❖ Melon

❖ Nectarine

❖ Orange

❖ Papaya

❖ Peach

❖ Pear

❖ Plum

Some babies may have an allergic reaction to highly acidic fruits.

Always discuss the addition of new foods with your metabolic dietitian.

CHOOSE YOUR FOODS

9 to 12 Months Old

Vegetables

Selection & Preparation

Vegetables can be served hot or cold, but all will need to be cooked.

Fresh:

- Wash thoroughly.
- Raw vegetables are difficult to chew, swallow, and digest.
- Cook until tender and easily pierced with a fork.
- Cut into small pieces, long thin strips, or grate.

Canned or Jarred:

- Be careful of the amount of sodium, choose low sodium if possible.
- Rinse the can or jar before opening.
- Can be served directly from the can, cut to appropriate size.

Frozen:

- Must be cooked until tender and cut to appropriate size.



❖ Asparagus

❖ Beets

❖ Broccoli

❖ Carrot

❖ Corn

❖ Green bean

❖ Green Pea

❖ Pumpkin

❖ Squash

❖ Sweet peppers

❖ Sweet Potato

❖ Zucchini

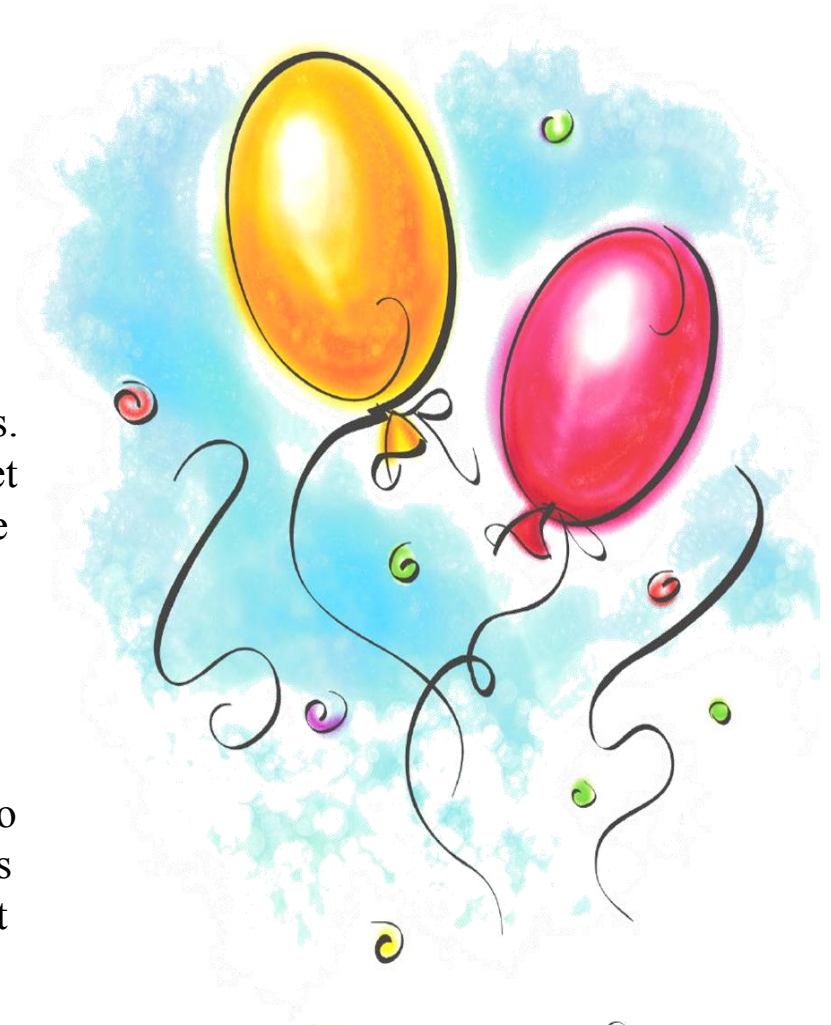
Some vegetables should be added to the diet closer to one year of age.

Always discuss the addition of new foods with your metabolic dietitian.

PARTY TIME TIPS

A Friend's House

- ❖ Tell the hostess what foods your child can have.
- ❖ Offer to stay and help with the party.
- ❖ Avoid buffet lunches where children can help themselves. Ask your hostess if the buffet lunch can be served from the kitchen so you can keep an eye on what your child is eating.
- ❖ Provide your own food, but drop it off before the party so as not to make too much fuss over your child's special diet in front of the other kids.
- ❖ If you don't stay, give the hostess clear instructions (depending on the child's age); leave a number where you can be reached.



At School

- ❖ Try to always have several appropriate low Phe treats in the teacher's closet or the school's refrigerator or freezer.



PARTY TIME RECIPES

ICE CREAM

Yields: 5-8 Servings

Phe: FREE

Ingredients:

3 cups	Rich's Whip Topping
1 cup	Water
cup	Sugar
2-3 tsp	Vanilla
2-3 tsp	Yellow Food Dye



Directions:

1. Mix all ingredients together and stir until the sugar is dissolved.
2. Mix with a mixer for 1 minutes.
3. Freeze in the metal cylinder container of an ice cream machine.

CHOCOLATE CAKE

Yields: 13 slices

Phe: 12 mg per slice (30 mg Phe/100g)

Ingredients:

1 box	Loprofin Chocolate Cake Mix*
cup	Vegetable Oil
1 cup	Sparkling Water
1 can	Frosting



Directions:

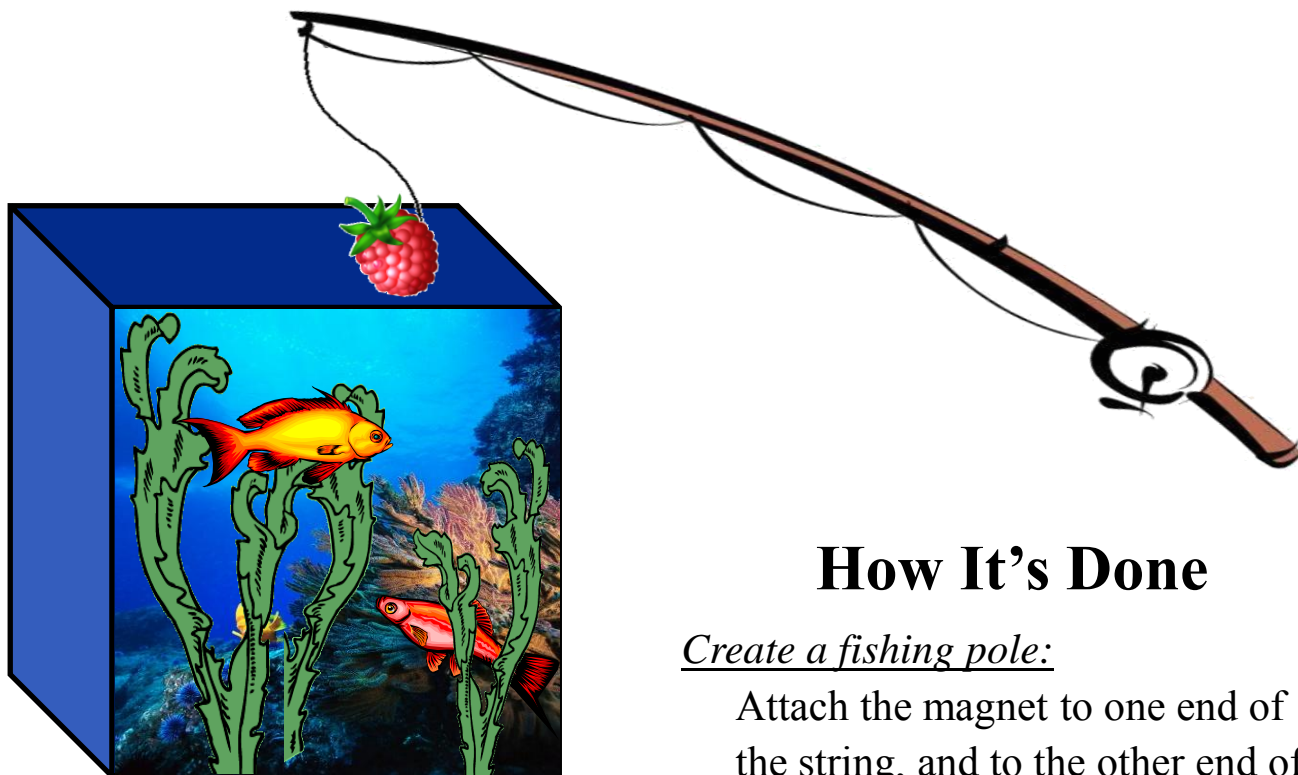
1. Pour cake mix into mixing bowl and add oil.
2. Add sparkling water and mix well.
3. Pour mixture into greased cake tin and let stand for 15 minutes.
4. Bake in an oven at 350 for 45-55 minutes.
5. When cake has cooled completely, frost with your favorite Phe free frosting.

* Nutricia North America -- Low Protein Products - www.shsna.com/pages/loprofin.htm

TEACHING AID

Fishing For Phe

Objective: To learn the names of a variety of foods and to recognize them as “Yes” and “No” foods.



How It's Done

Create a fishing pole:

Attach the magnet to one end of the string, and to the other end of the string, to the stick.

Create an ocean or lake:

- ❖ Cover a cardboard box with blue paper.
- ❖ Draw fish, seaweed, or anything you find in the sea or a lake on the paper.
- ❖ Fill the box with paper food models and attach a paper clip to each model.

Materials Needed

- ❖ Stick or dowel
~ about two feet long
- ❖ One small magnet
- ❖ String, 3 feet long
- ❖ Card board box
- ❖ Paper food models
- ❖ Paper clips
~ one for each food model

Go Fishing!

Adapted from C. Trahms et. al. Games That Teach

TEACHING AID

Clinic Supermarket

Objective: To learn the names of a variety of foods and to recognize them as “Yes” and “No” foods.



How It's Done

Create a sign for your store:

Develop a sign that clearly states “Clinic Supermarket” and decorate the sign with pictures of food. Coupons and magazine clippings are an easy, inexpensive, and colorful approach to decorating.

Create a shopping environment:

- ❖ Place food models on a table and allow each child to shop for “yes” foods.
- ❖ The container for shopping can be a paper bag, shopping basket, or a mini shopping cart.
- ❖ This concept can be expanded upon based on the age of the audience. You can include looking up the Phe content of the foods purchased, buying foods for a recipe, and calculating the Phe in the recipe.

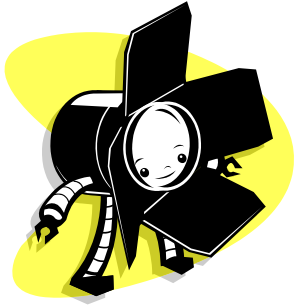
Materials Needed

- ❖ Posterboard
- ❖ Markers, crayons, or paints
- ❖ Magazines and newspapers
- ❖ Paper or plastic food models
- ❖ Basket or paper bag

TEACHING AID

Role Playing

Objective: To rehearse when and how to say “Yes” or “No” when confronted with food choices.



Lights...

Camera...



Action



Materials Needed

- ❖ Paper or plastic food models

How It's Done

Create an interactive environment:

- ❖ Using food models, have the clinic professional or parent offer a “yes” or a “no” food to the child.
- ❖ Have the child identify the food either as a “yes” or a “no” food and then ask the child if they would like a little taste.
- ❖ Reinforce appropriate answers and modify inappropriate responses.

TEACHING AID

Red Light!! Green Light!!

Objective: To reinforce “Yes” and “No” food choices.

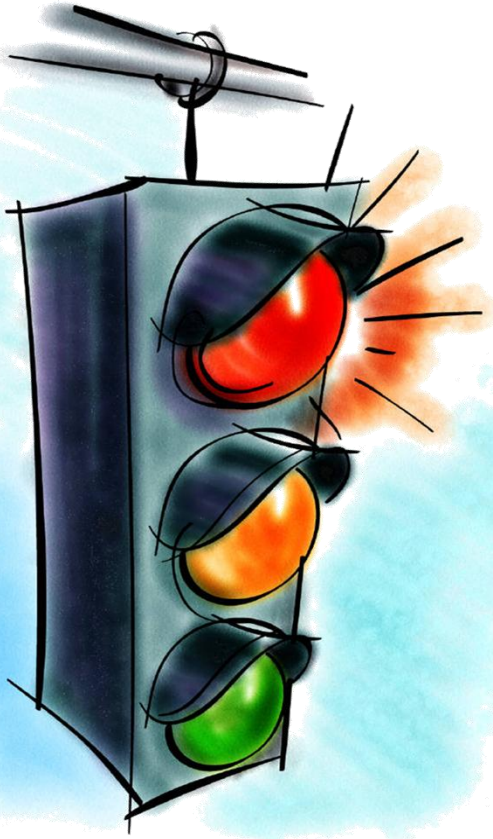
How It's Done

Create a board:

- ❖ Make a felt board that is in the shape of a traffic light.
- ❖ Attach food models to separate pieces of felt to use as game pieces.

Create a shopping environment:

- ❖ Use green for “yes” foods and red for “no” foods.
- ❖ Develop more game pieces as the group matures (i.e.: yellow is for foods that are “yes” but in very limited quantities).
- ❖ Use felt backed food models and let the children place them in the appropriate section.
- ❖ A positive reward system is used with all of these programs.
 - ❖ Stickers
 - ❖ Buttons
 - ❖ Applause
 - ❖ Verbal affirmation
 - ❖ Low protein treats



Materials Needed

- ❖ Posterboard
- ❖ Felt for board and game pieces
- ❖ Paper food models
- ❖ Glue or staples

TEACHING AID

Ways We Are Alike & Ways We Are Different

Objective: To introduce the concept of genetic variability and to achieve an understanding that variability is what makes each individual unique and special.



The Long Term Goal is to Achieve Acceptance of Hyperphenylalaninemia as an Inherited Trait!

How It's Done

- ❖ Discuss the terms “alike” and “different” and use these terms in relation to physical characteristics.
- ❖ Compare your physical traits to those of others in the room, pointing out ways you are alike and ways you are different.
- ❖ Identify a feature in yourself that is different than that seen in other individuals in the room. Point out how this makes you unique and special.
- ❖ Compare physical characteristics of other individuals present.
- ❖ Have all individuals with PKU in the room raise their hand. Acknowledge this as a trait that is either shared with other people in the room or as a trait that is unique and special.

No Materials Needed

TEACHING AID

We Are Alike & Different

Objective: To engage preschool and early elementary children in discussion about how everyone has ways they are alike and ways they are different, and how that is good.



How It's Done

Begin a discussion:

- ❖ Use the handout “We Are Alike & Different” to introduce the terms “alike” and “different.”
- ❖ Once all the children have completed the instructions on the handout, have them discuss their own traits and characteristics.
- ❖ Ask all participants with brown hair to raise their hand. Count the number of hands, write the number on the board. Repeat this using other colors of hair and traits.
- ❖ Discuss that there are some traits that we can not see, give examples (PKU).
- ❖ Have all individuals with PKU raise their hand. Make is “cool” to have PKU.
- ❖ Emphasize that both differences and similarities are good.

Additional Activity 1: Read the book We're Different, We're The Same.

Additional Activity 2: Ask the participants why they think they have a nose, hair, and other traits discussed. Introduce the concept of a gene being a recipes or a set of instructions to make something. Discuss that there are many recipes or genes inside our body (that we can not see). For example, a recipes or instructions to make our eyes, ears, hair color. Introduce the concept of a gene or recipe for PKU. Ask the participants if they can list any other recipes they have.

Handouts Needed
We Are Alike & Different

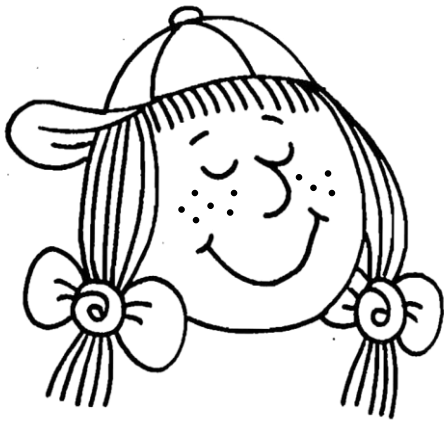
Adapted from March of Dimes Activity Master 2

HANDOUT: We Are Alike & Different

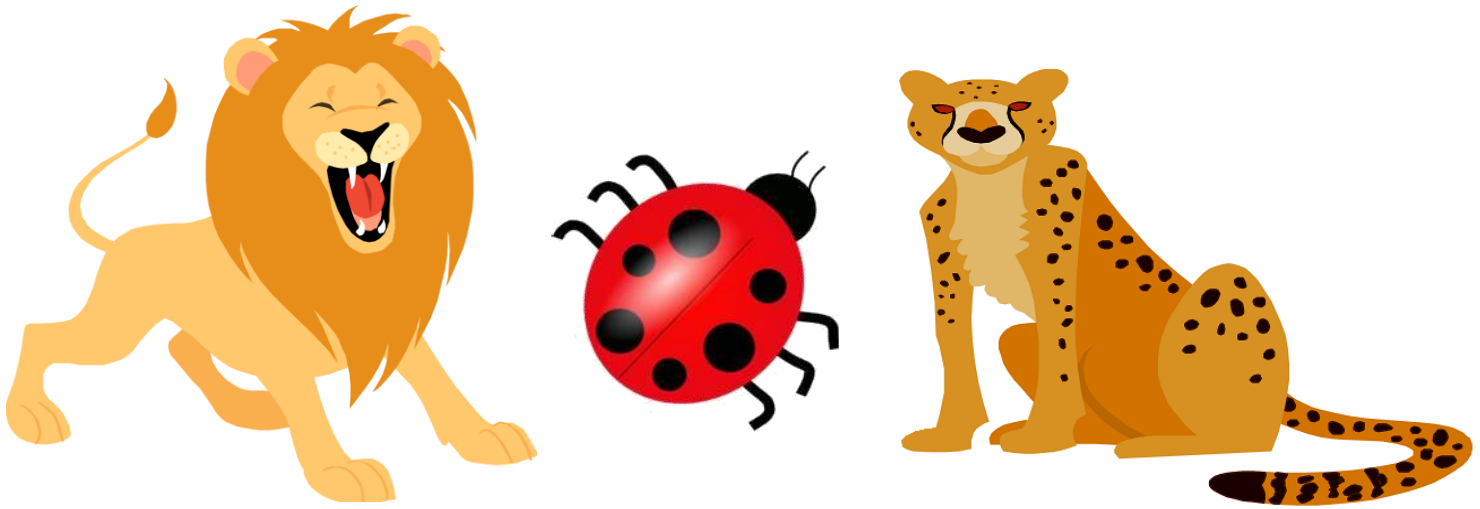
Draw a circle around all the children who have curly hair.

Draw a square around all the children who have rosy cheeks.

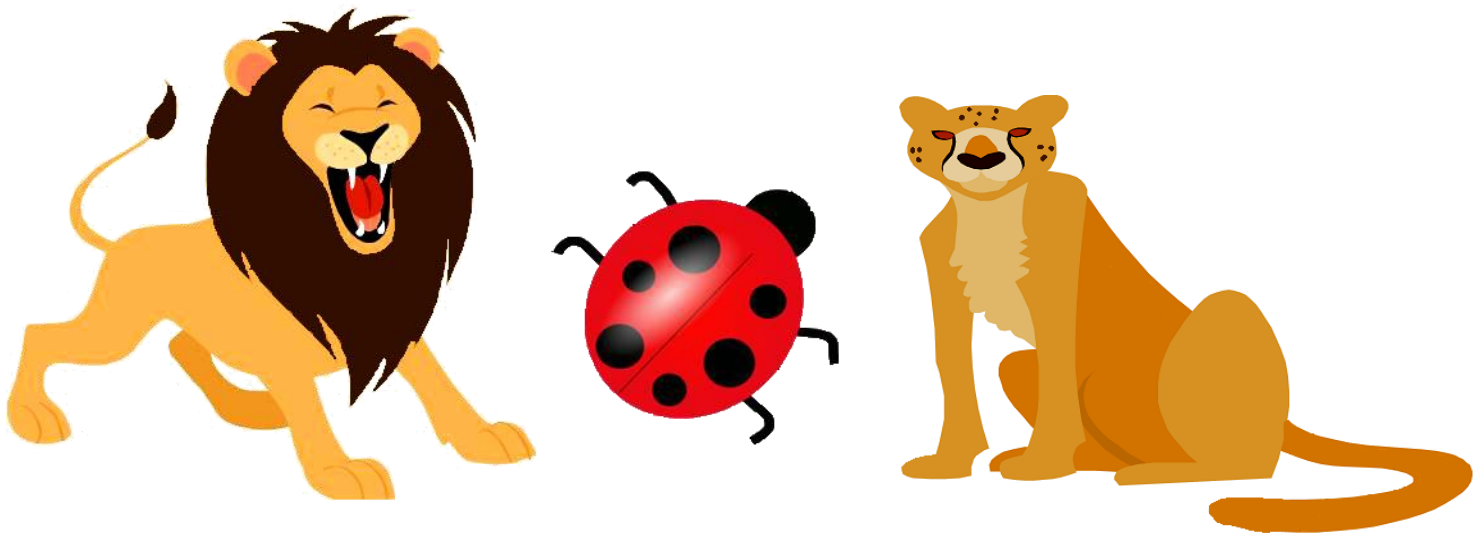
Draw a triangle around all the children who have freckles.



HANDOUT: Alike & Different



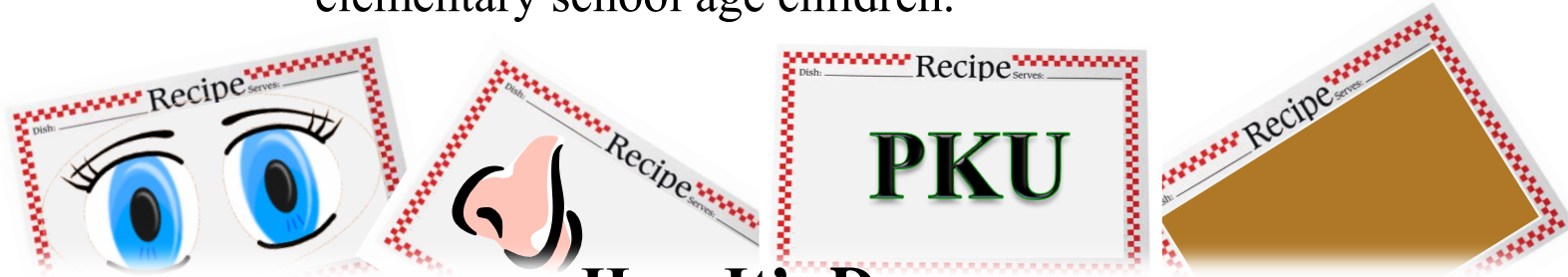
How are these animals alike?
How are they different?



TEACHING AID

My Genetic Recipe Book

Objective: Introduce the concept of a gene to kindergarten and early elementary school age children.



How It's Done

Introduce (or review) the concept of a gene being a recipe (i.e. a set of instructions to make something).

- ❖ Discuss the term “recipe” and how it is a set of instructions to make something. Introduce the term “gene” and discuss how it is like a recipe.
- ❖ Dependent upon participants writing skills, have participant’s write the word “gene” on a piece of paper or white board. Discuss that inside our bodies, there are thousands of genes or recipes.

For example: There are genes (i.e. recipes) to make our eyes and genes (i.e. recipes) to make our fingers. There are genes for hair color; individuals with brown hair have a gene to make their hair brown. There are genes that result in PKU.

Use the My Genetic Recipe Book Handout.

- ❖ Have participants write their names on the top of the handout. Ask them to feel their hair and determine if it is curly or straight. If their hair is curly, they should circle the “curly recipe gene”; if straight, they should circle the “straight recipe gene”. Next talk about hair color. Have them circle the “hair color recipe gene” that is appropriate for them. Note, on the hand out sheet, you will need to add colors to the blank boxes (such as back, brown, blond and red) as these were left blank due to the high likelihood that a color printer or copier would not be used when generating the hand out.
- ❖ Ask participants if they have eyes. Since they do, they need to circle the “recipe gene for eyes”. Repeat this for all traits. Finally ask the participants if they have PKU or HFI. Describe HFI, noting that people with this disorder cannot eat fruit. Have them circle the appropriate recipe gene.
- ❖ End by celebrating that they have just made a recipe gene book specific for them!! How cool is that!!

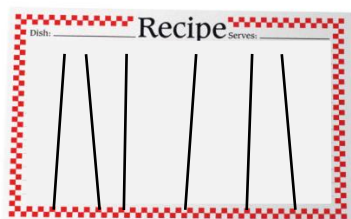
Handout Needed
My Recipe Gene Book

HANDOUT: My Genetic Recipe Book

Name _____

Each box represents a gene or recipe to make important parts of your body. Circle the genes (or recipes) that you have. For example, if you have curly hair, circle the curly hair gene. If you have straight hair, circle the straight hair gene. *Note for instructor:* The hair color genes need to be colored in (such as black, brown, blond, and red).

Gene For Hair Texture



Gene For Hair Color



Gene For Eyes



Gene For Nose



Gene For PKU



Gene For HFI



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RESOURCES



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<<http://www.dshs.state.tx.us/kids/colorbook/denny/denny1.shtm>>

Everyone Has Something. Margaret Domnick. Bloomington, Indiana: Author House, 2004. Print. ISBN: 1420800507

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Mile High, Low Protein Cookbook. Available through the Low Protein Food Store, IMD Clinic, The Children's Hospital, Aurora, CO. 303-724-2338

More Phe, More Choices: Think Healthy! Laurie Bernstein, Sommer Meyers, Casey Burns, Kathryn Bloxsom, Janine Gessner, and Catherine Long. IMD Clinic, The Children's Hospital, Aurora, CO. 303-724-2338

More Phe, More Choices: Think Healthy! Early Childhood. Laurie Bernstein, Casey Burns, and Kelly Parker. IMD Clinic, The Children's Hospital, Aurora, CO. 303-724-2338

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