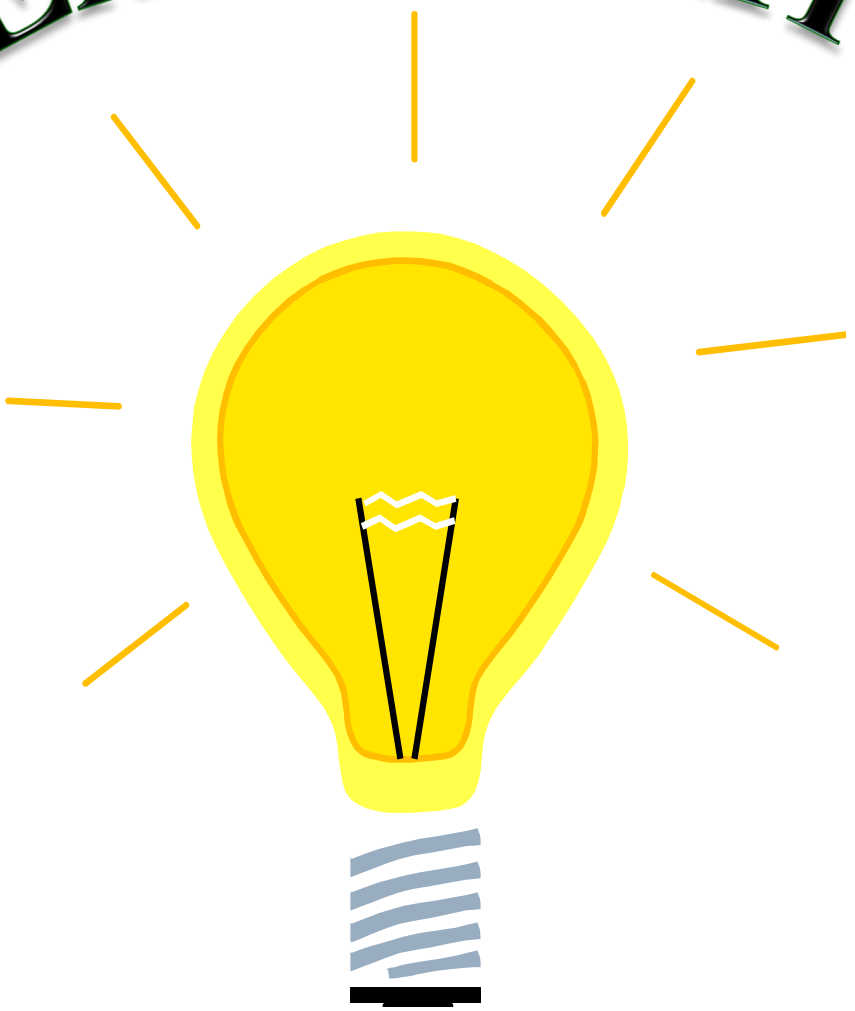
EAT RIGHT



STAY BRIGHT™

Guide For Hyperphenylalaninemia

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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 positive[1]
- ❑ 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 Issues[2]

- ❑ 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
- ❑ Siblings[3]
  - "Why Can't I Eat That?", *pg.* 113-127
- ❑ Parents plans to return to work or school
- ❑ Child care[4]
- ❑ 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
  - Biopterin 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 communication[5]
- ❑ 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 school[7]
- ❑ 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 mess[8]
- ❑ 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 levels[9]
- ❑ 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 diagnosis[10] 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, friends[11]
- ❑ 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 Issues[13]

- ❑ 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 autonomy[14]

CHECKLIST: One to Three Years Old

Parenting Skills and Chronic Disease

- ❑ Self-concept[15]
- ❑ 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 reading[17] (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 Issues[19]

- ❑ 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/~kommal/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 hyperphe, 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 that 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

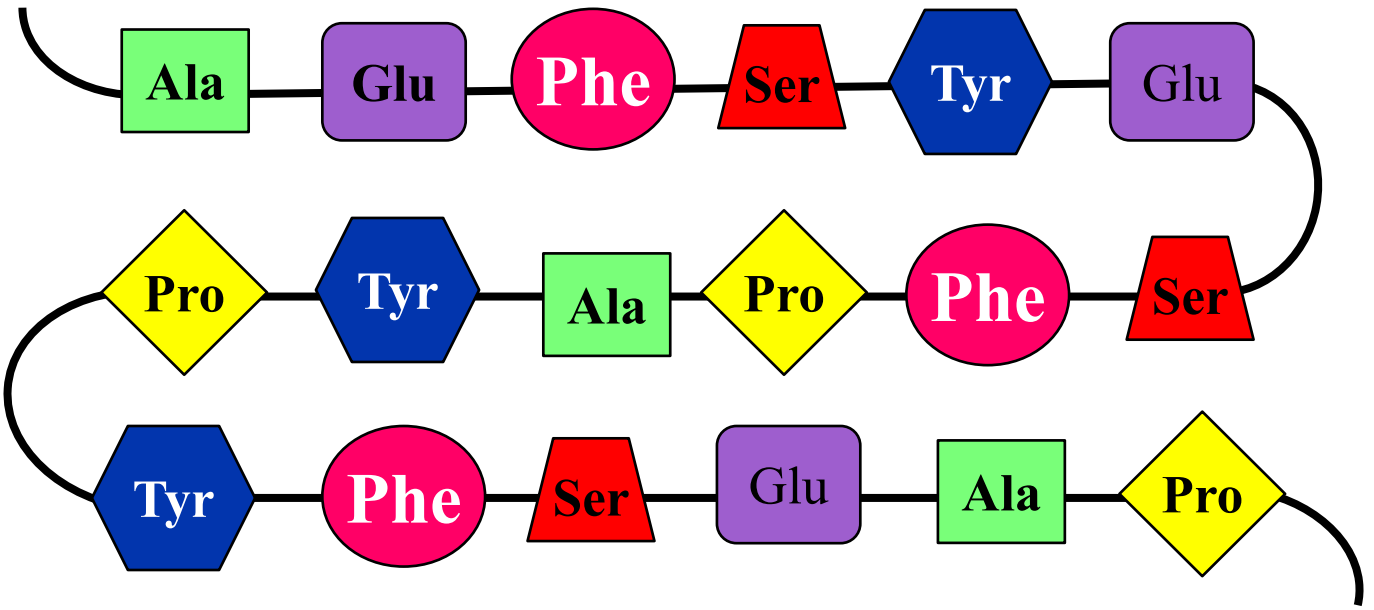
Secondary phenylalanine level is also high.
Confirmation that baby has PKU.
Baby should be seen at the metabolic clinic.

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.

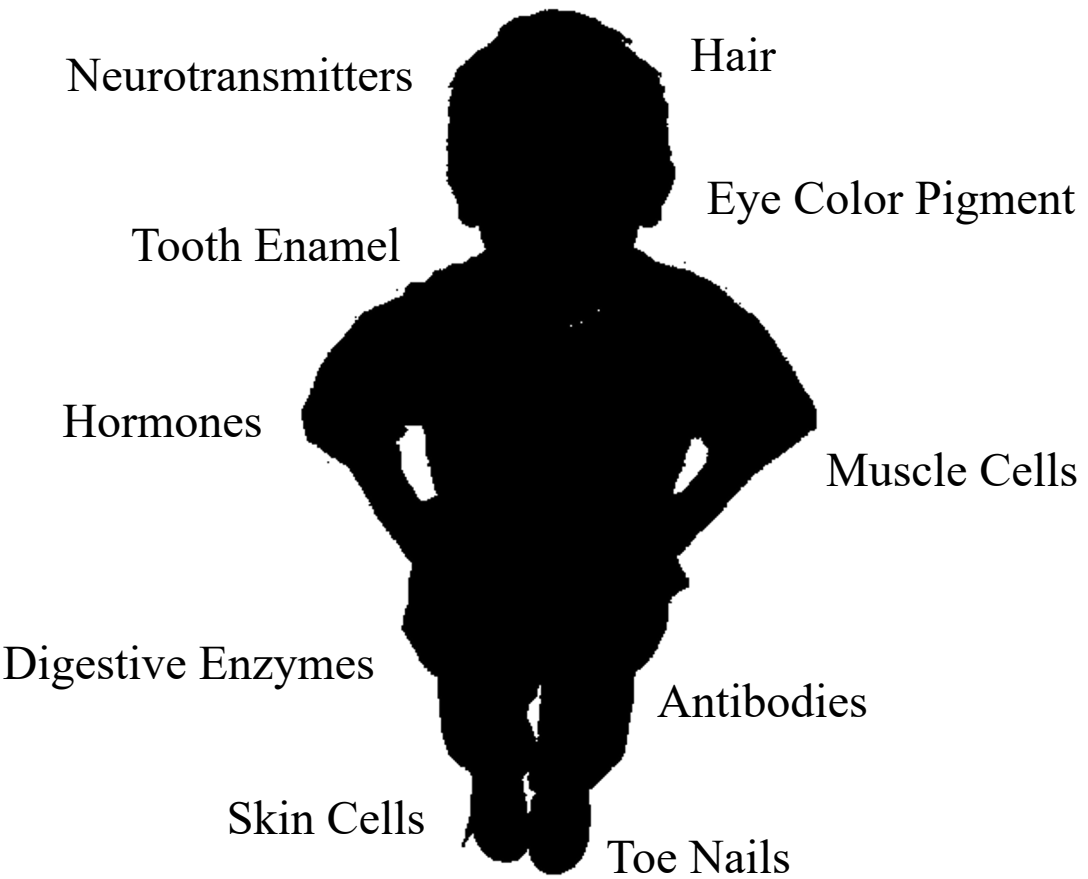
*We often shorten the word "*Phenylalanine*" to "*Phe.*"

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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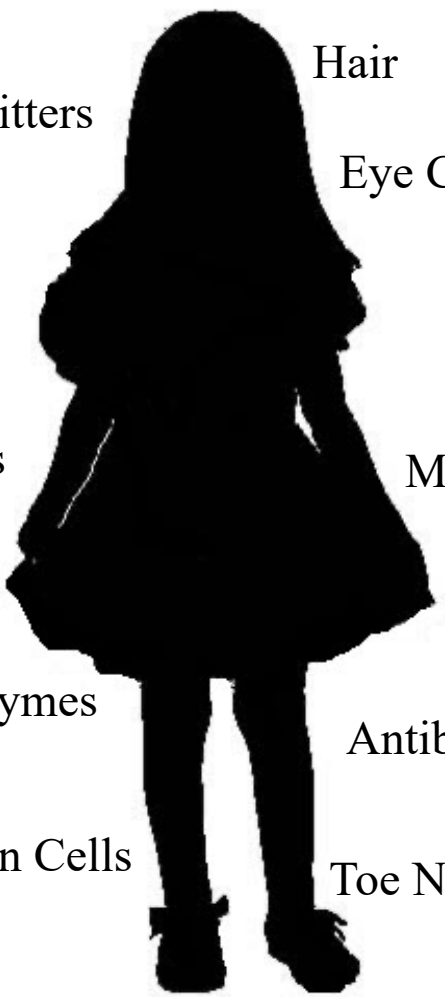


Hair

Neurotransmitters

Eye Color Pigment

Tooth Enamel

Hormones

Muscle Cells

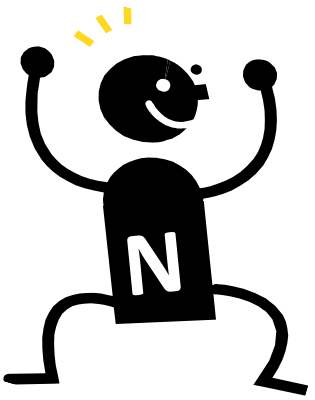
Digestive Enzymes

Antibodies

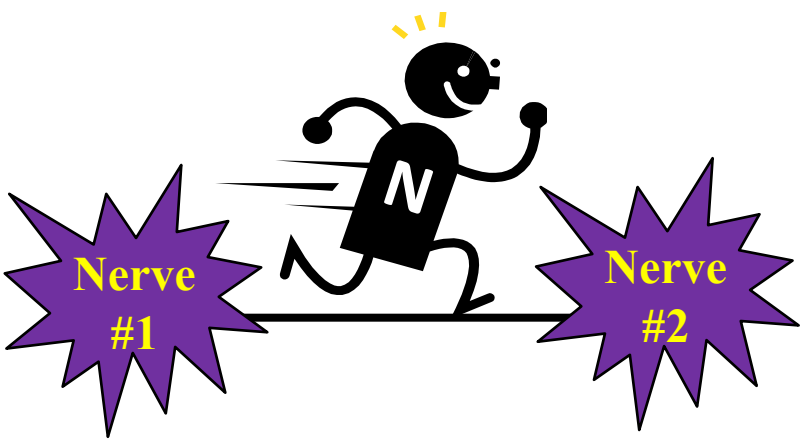
Skin Cells

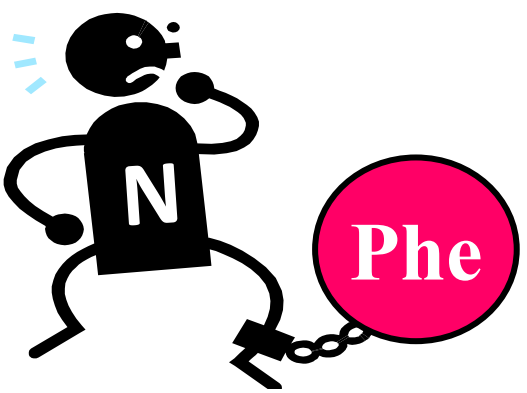
Toe Nails

Mr. Neurotransmitter



Mr. Neurotransmitter





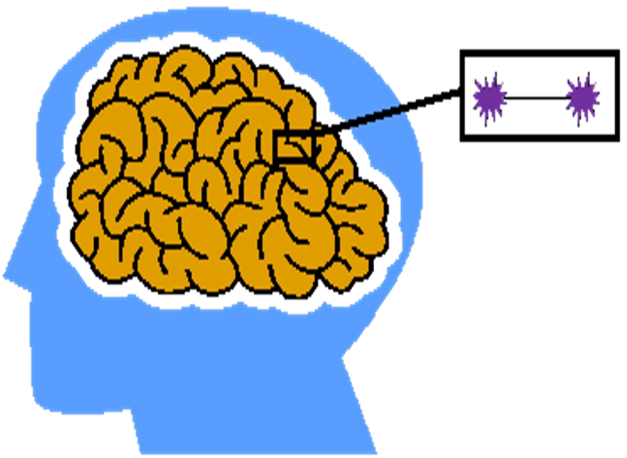
The cells in our brain make chemicals called neurotransmitters.

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

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

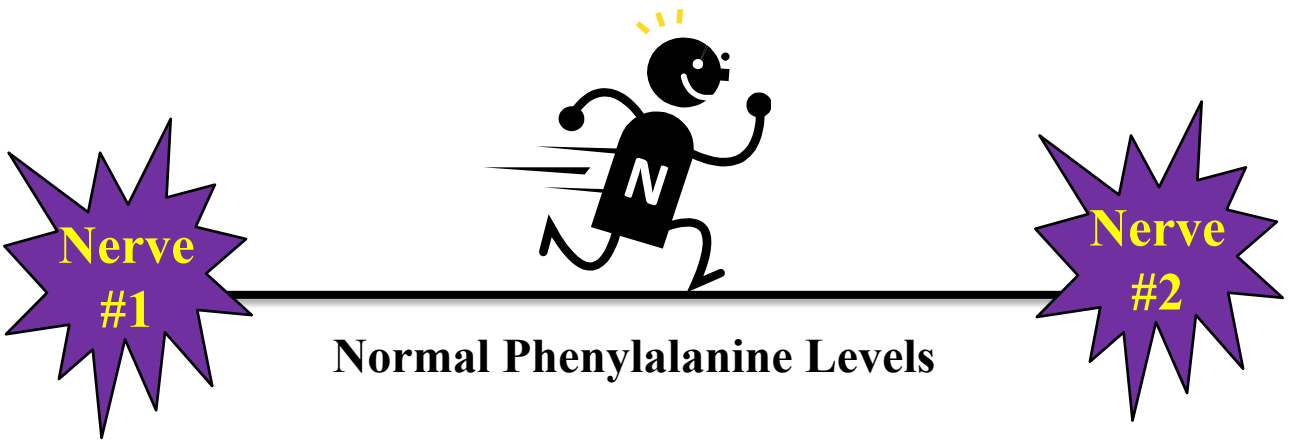
Too much phenylalanine and not enough tyrosine interferes with the formation and performance of neurotransmitters.

ALTERED NEUROCHEMISTRY

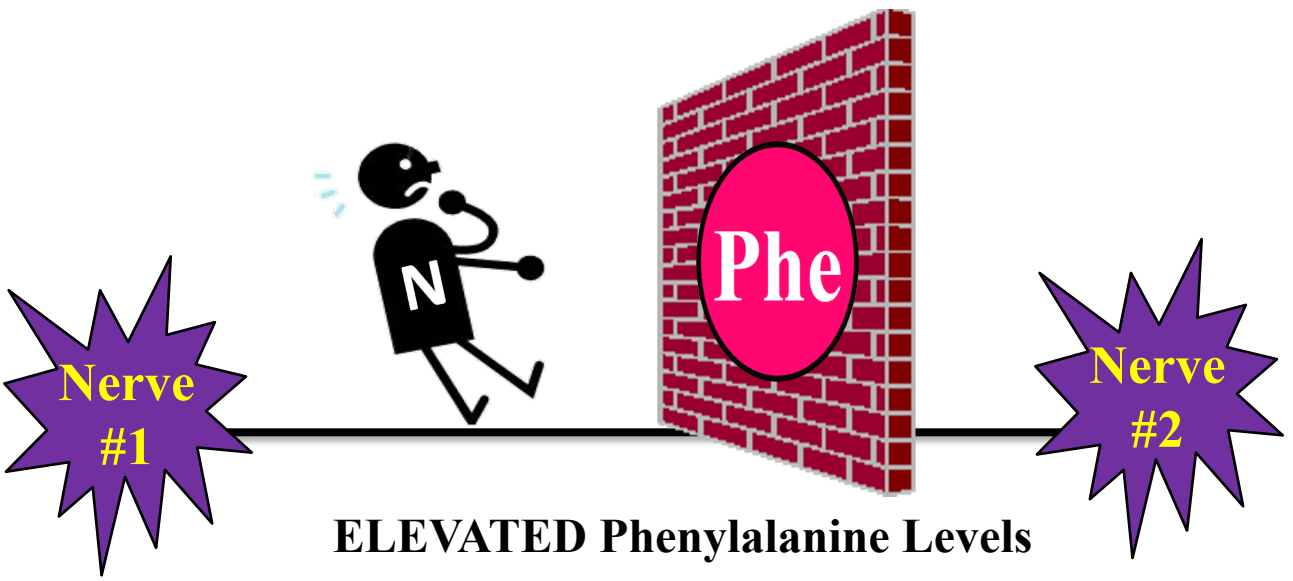


When phenylalanine levels are in treatment range, neurotransmitters can deliver clear messages.

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

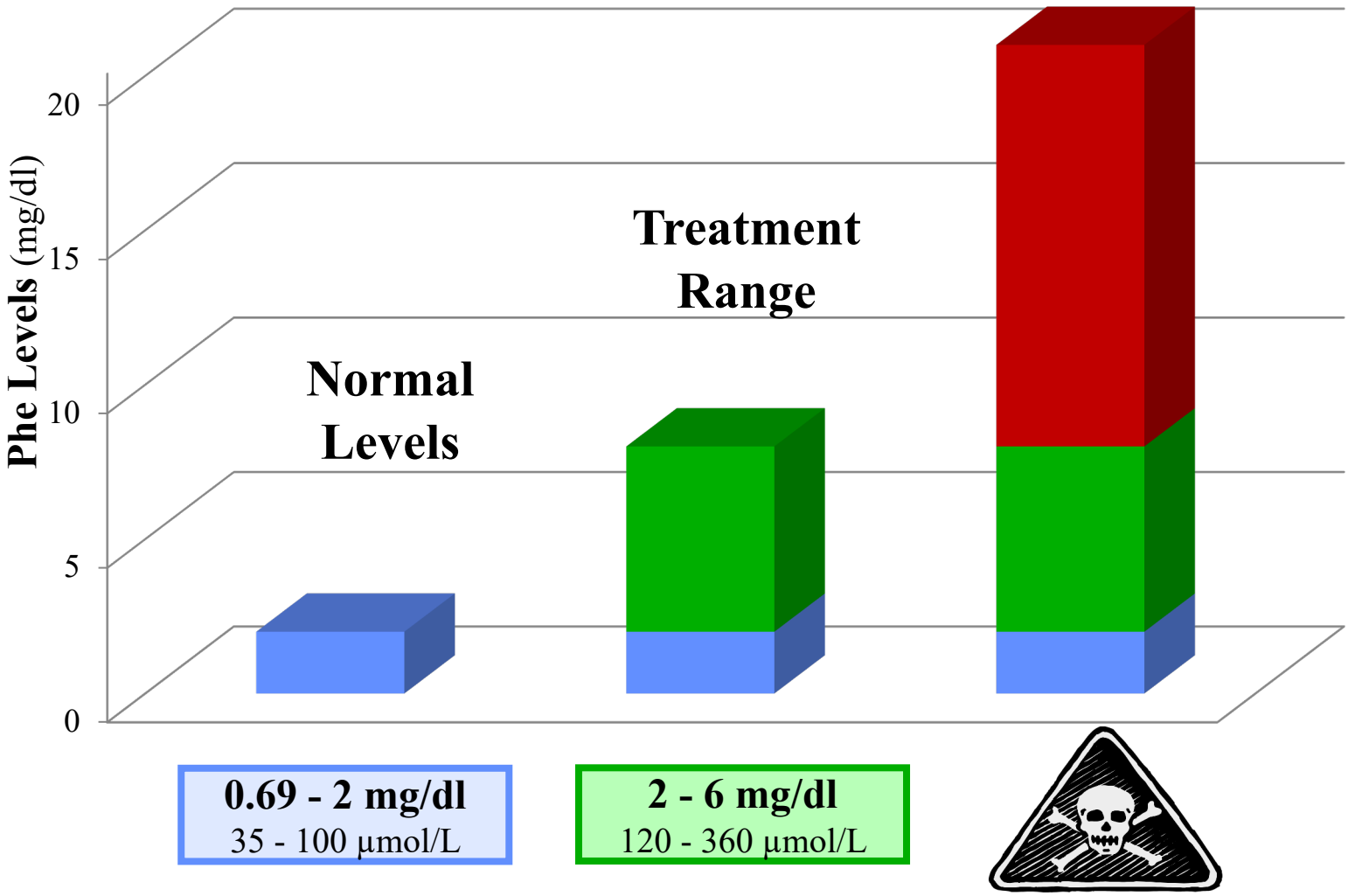


Normal Phenylalanine Levels



ELEVATED Phenylalanine Levels

Phenylalanine (Phe) Levels



0.69 - 2 mg/dl
35 - 100 µmol/L

2 - 6 mg/dl
120 - 360 µmol/L

High Phe Levels:

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

Phenylalanine (Phe) Levels

Treatment Range
120 - 360 µmol/L (2 – 6 mg/dl)

Low	Normal	Non- PKU HPA	Mild PKU	Classic PKU
< 35 µmol/L (< 0.5 mg/dl)	35 - 120 µmol/L (0.5 - 2 mg/dl)	120 - 600µmol/L (2 - 10 mg/dl)	600 - 1200 µmol/L (10 -20 mg/dl)	> 1200 µmol/L (> 20 mg/dl)

Hyperphenylalaninemia (HPA)

[------hyper-----][-----phenylalanin-----][------emia-----]

high levels of → *phenylalanine* → *in the blood*

Phenylketonuria (PKU)

[------phenylketon-----][------uria-----]

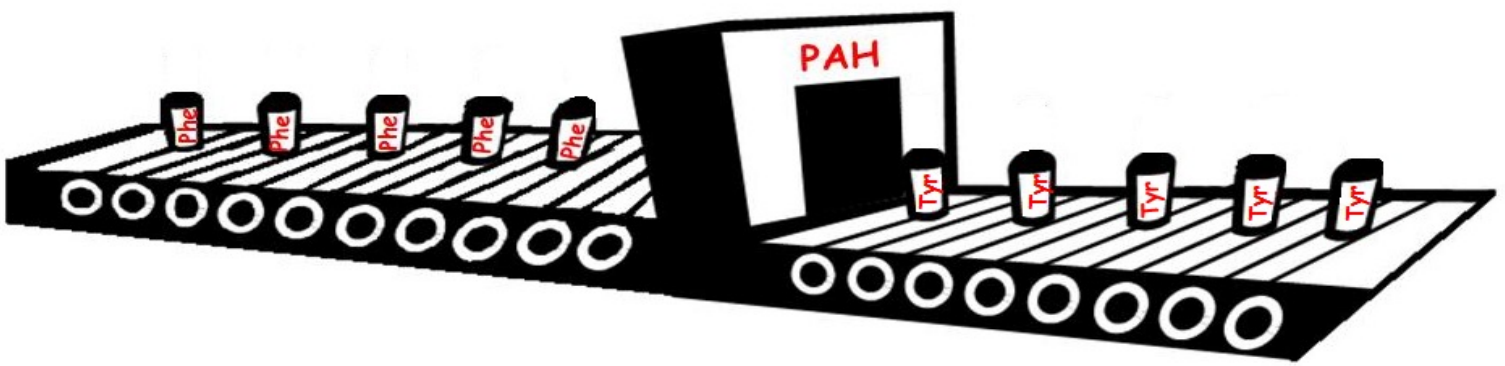
phenylketone → *in the urine*

Information taken from Nutrition Management of Patients with Inherited Metabolic Disorders by Phiyllis Acosta

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.**

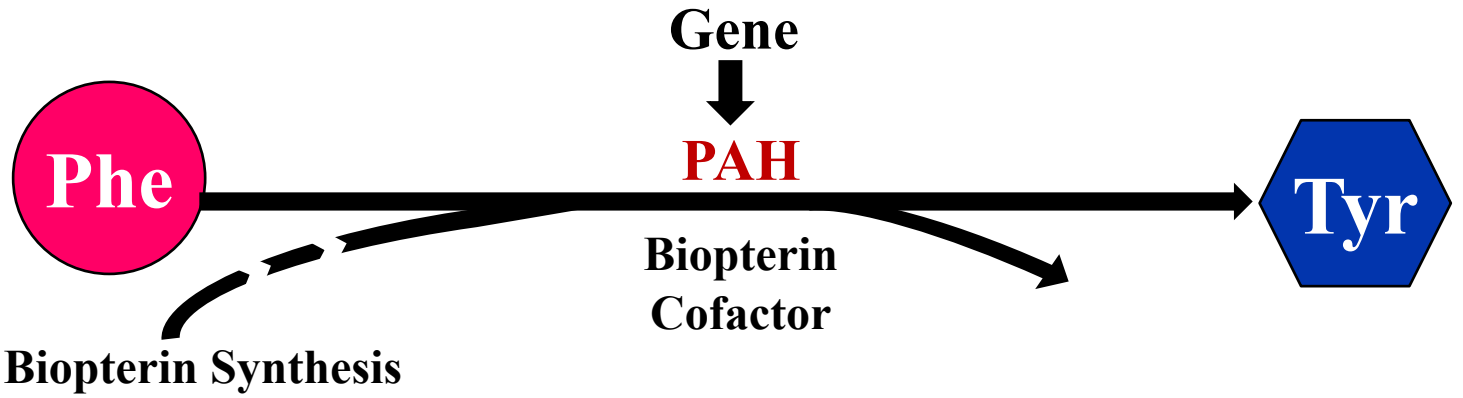
The Result: Too much **Phe** and not enough **Tyr**.

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

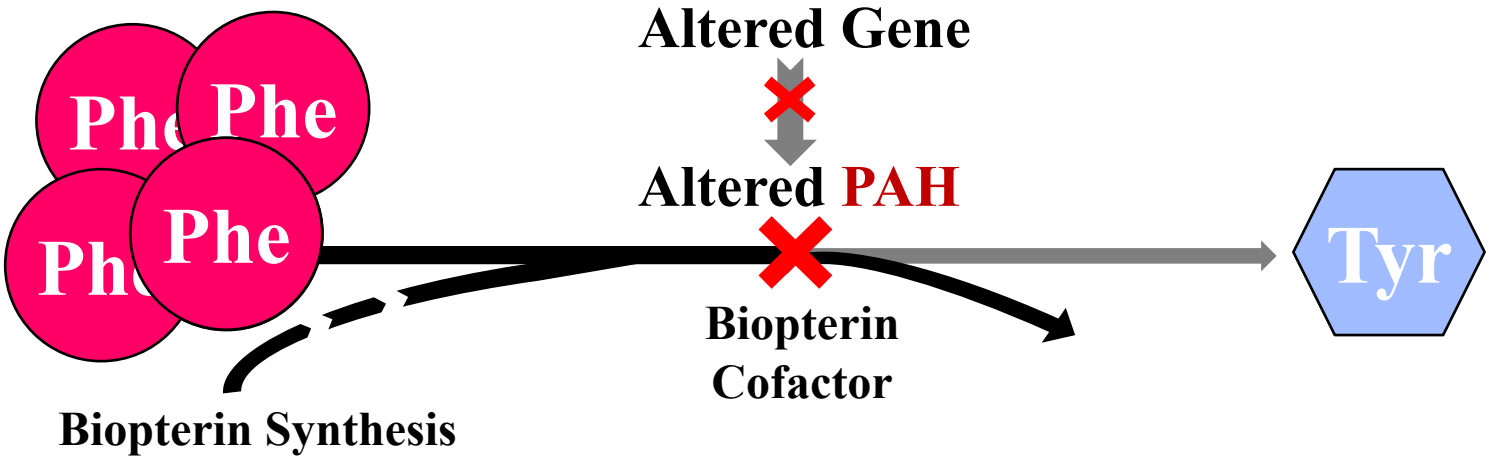
Pathways

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

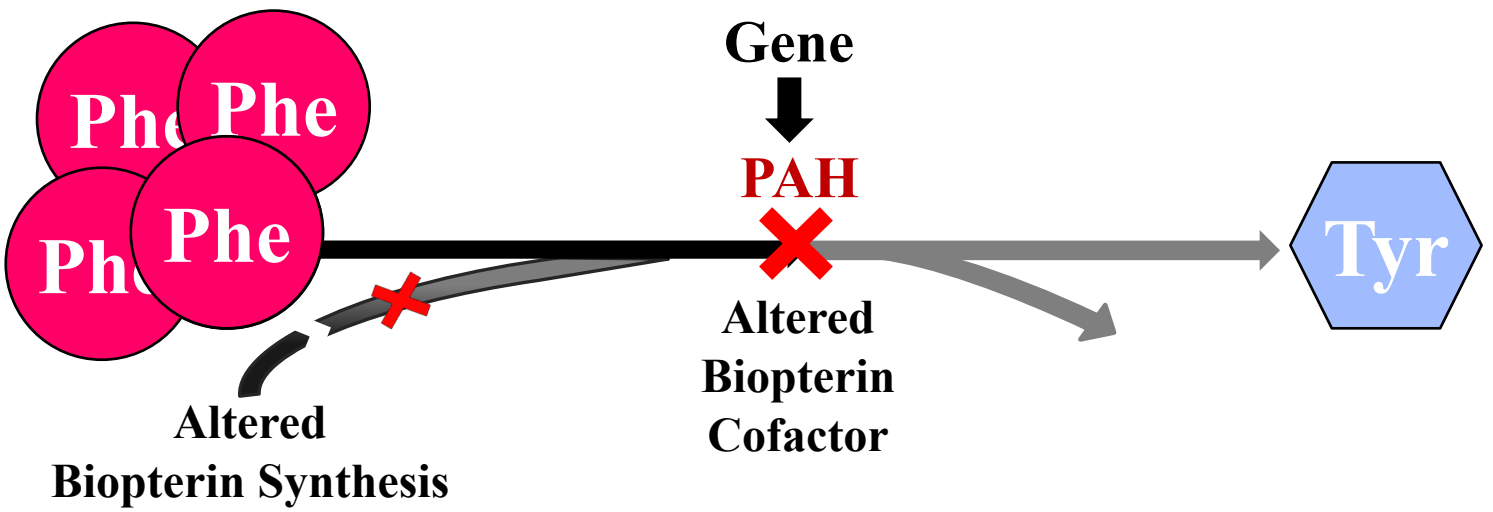
Normal Phenylalanine Pathway



Classic PKU Pathway



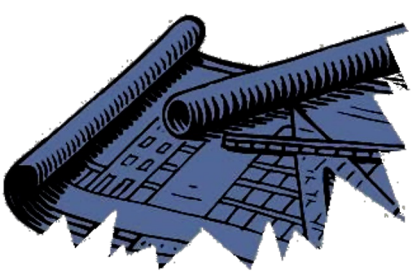
Biopterin Cofactor Defect Pathway



DNA BLUEPRINT

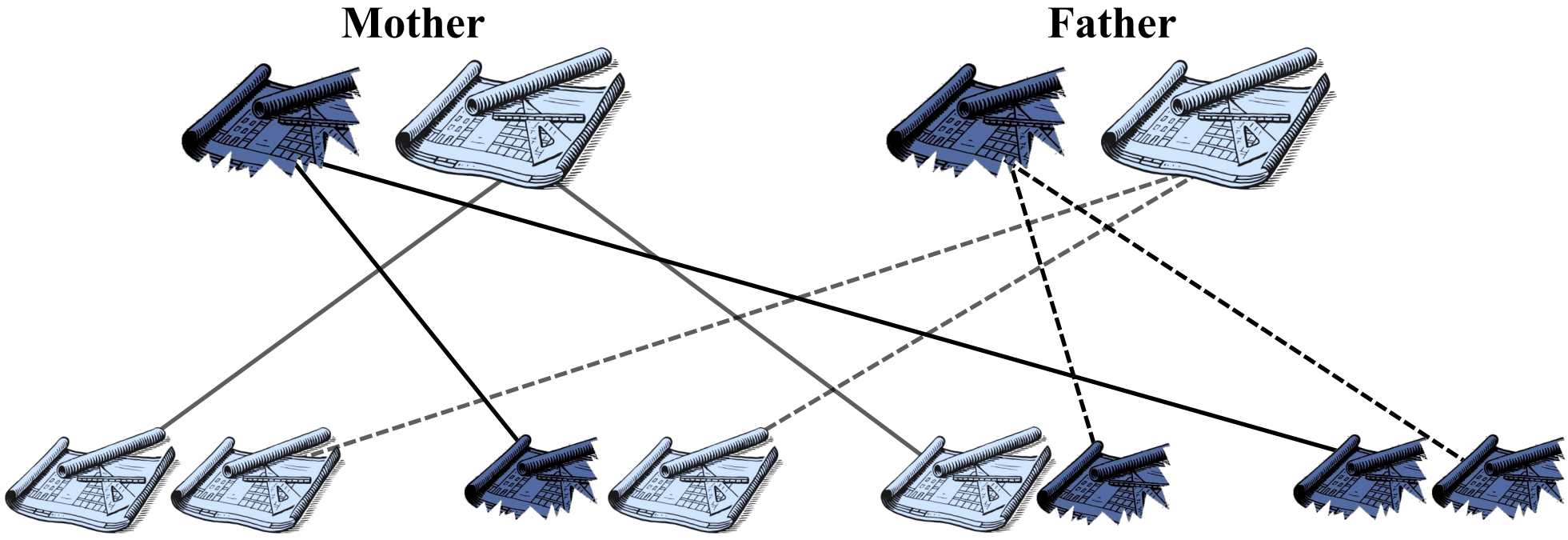
Instructions are normal. Phenylalanine hydroxylase (PAH) is made. Works efficiently.

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

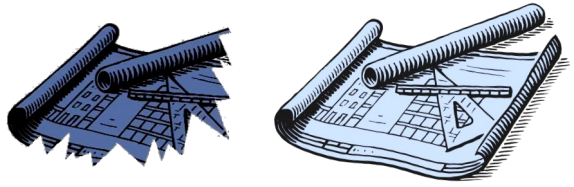


Gene for normal or full phenylalanine hydroxylase (PAH) activity

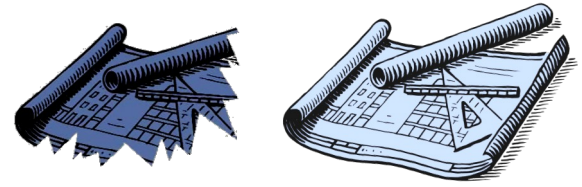
Gene for altered phenylalanine hydroxylase (PAH) activity

AUTOSOMAL RECESSIVE INHERITANCE

Mother



Father



Gene for normal or full phenylalanine hydroxylase (PAH) activity

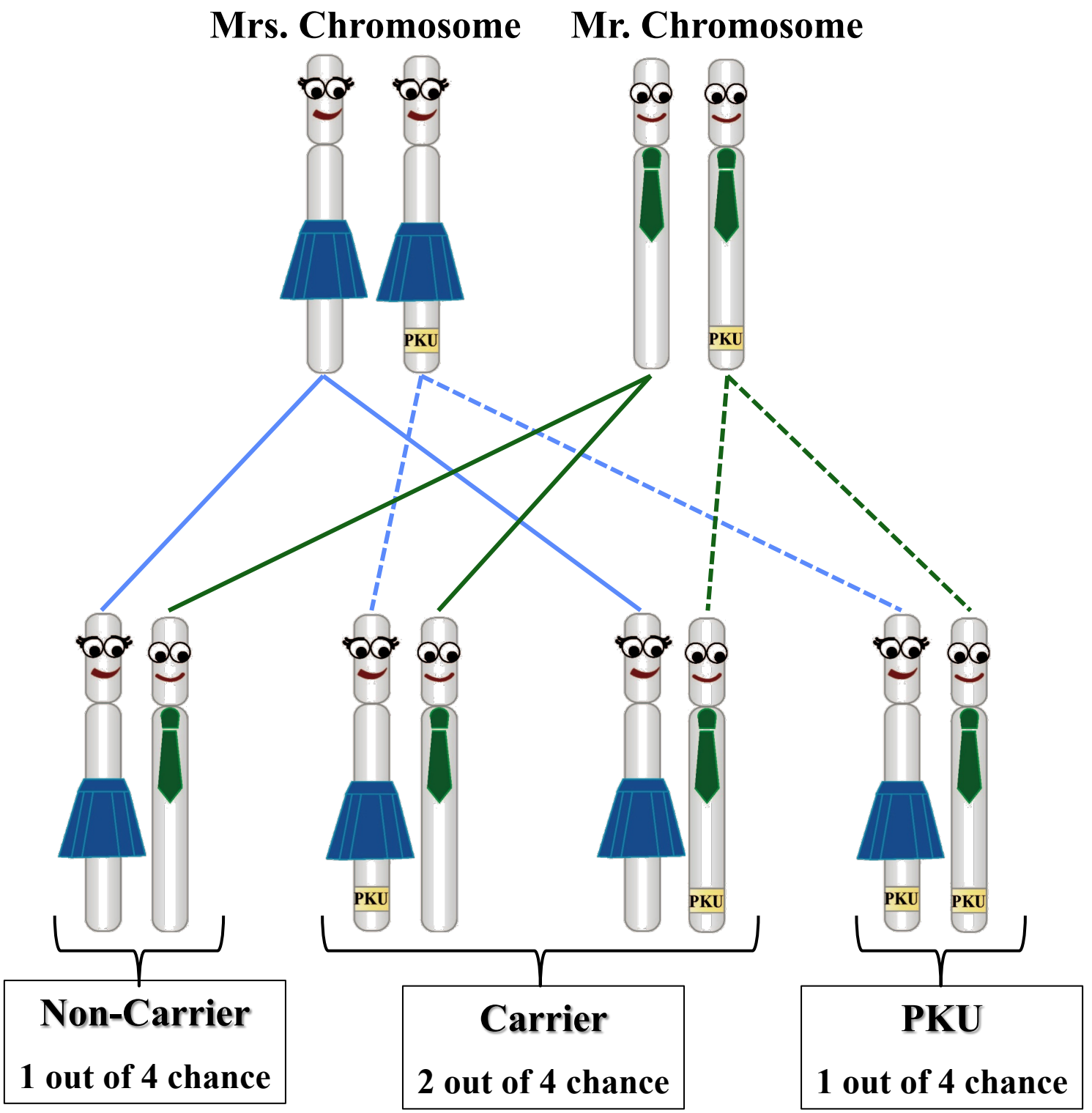
Gene for altered phenylalanine hydroxylase (PAH) activity

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.



Images by Janine M. Gessner

AUTOSOMAL RECESSIVE INHERITANCE

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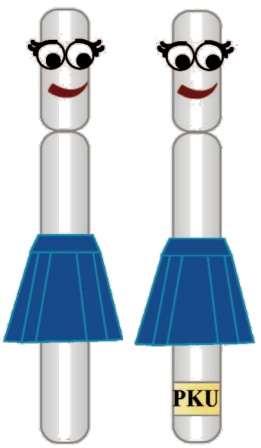
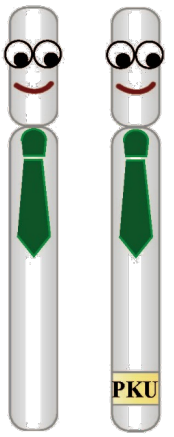
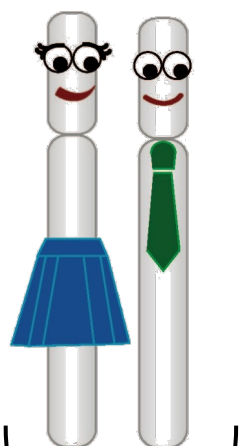
Mrs. Chromosome **Mr. Chromosome**

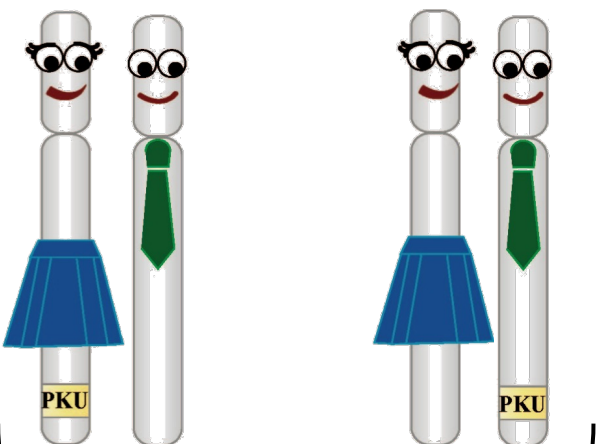






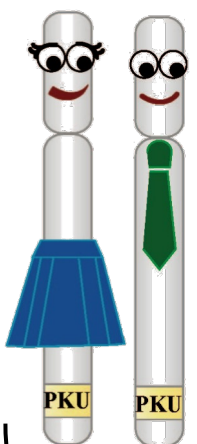
Non-Carrier

1 out of 4 chance



Carrier

2 out of 4 chance

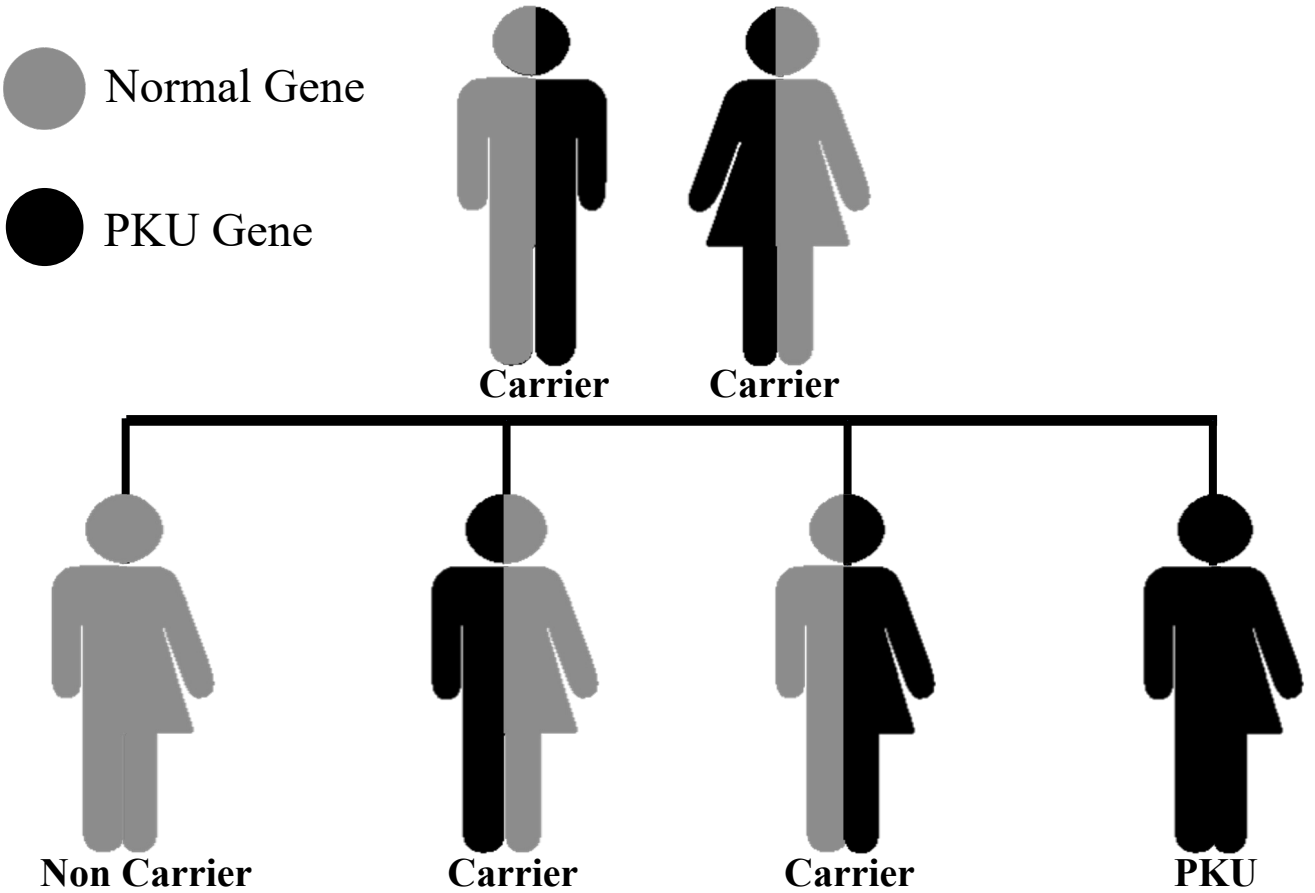


PKU

1 out of 4 chance

Images by Janine M. Gessner

AUTOSOMAL RECESSIVE INHERITANCE



WHAT DOES AUTOSOMAL RECESSIVE INHERITANCE REALLY MEAN?

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

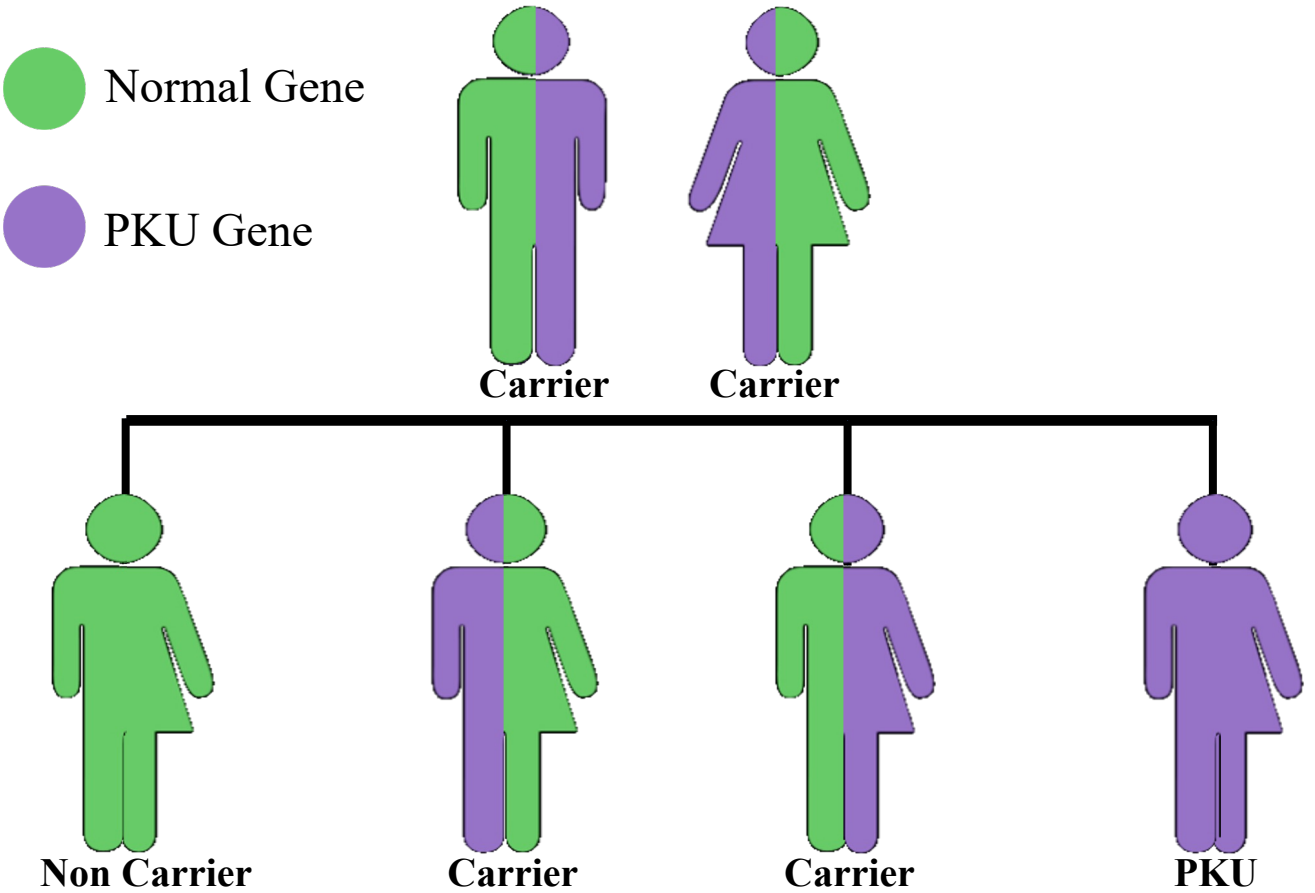
If both parents carry the PKU gene, there is a:

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



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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 phenyalanine 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

Birth to 6 Months | 6 to 12 Months | 1 to 2 Years | 2 to 7 Years | 8 Years to Adulthood

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

Principles of Diet Prescription

Throughout a Lifecycle

Birth to 6 Months | 6 to 12 Months | 1 to 2 Years | 2 to 7 Years | 8 Years to Adulthood | Motherhood

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

PRINCIPLES OF DIET PRESCRIPTION

Birth to 7 Years Old

+

AND/OR

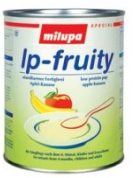
=

Phe Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

+

+



=

Phe Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

+

AND/OR

+

=

Phe Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

+

+

=

Phe Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

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

+

AND/OR

=

Phe

Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

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.

<u>If Breastfeeding</u>: 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 <u>10</u> grams of <u>Periflex Infant</u>, add <u>2</u> ounces of water to make a total of <u>2</u> 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

6 to 12 Months

+

+

=

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 **Infant Formula**

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.

Solid and Low Protein Food:

_____

mg Phe **gm Protein** **# Exchanges**

Circle One

Medical food may provide complete nutrition without any Phe.
Infant formula with the addition of cereal, baby food, and finger foods
(at ~ 9 months) provide additional amino acids, vitamins, minerals, and calories.

DAILY DIET PRESCRIPTION

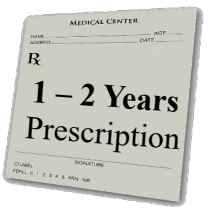
1 to 2 Years Old

+

AND/OR

+

=



Phe 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 **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

Phe 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.

<u>If Breastfeeding</u>: 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

<u>Call Our Clinic So We Can Make A Diet Change If:</u>

- 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 Foods[4]			
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 "Pheneurosis" (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.

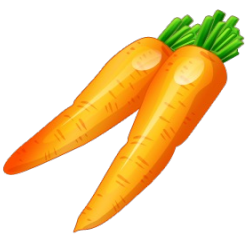
- **Can your baby support his/her own head?**

  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
- ❖ Very Small Pieces of Fruit
  - ❖ Banana Slices
  - ❖ Grapes: peeled and cut quarters
- ❖ Milupa lp cereal* /Cheerios
- ❖ Soft Cooked Vegetables
  - ❖ Carrot Slices
  - ❖ 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.

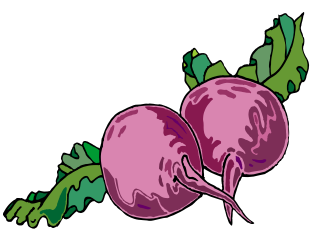
Canned or Jarred:

- 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
¾ 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 1½ 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*
½ cup	Vegetable Oil
1 cup	Sparkling Water
1 can	Frosting

Directions:

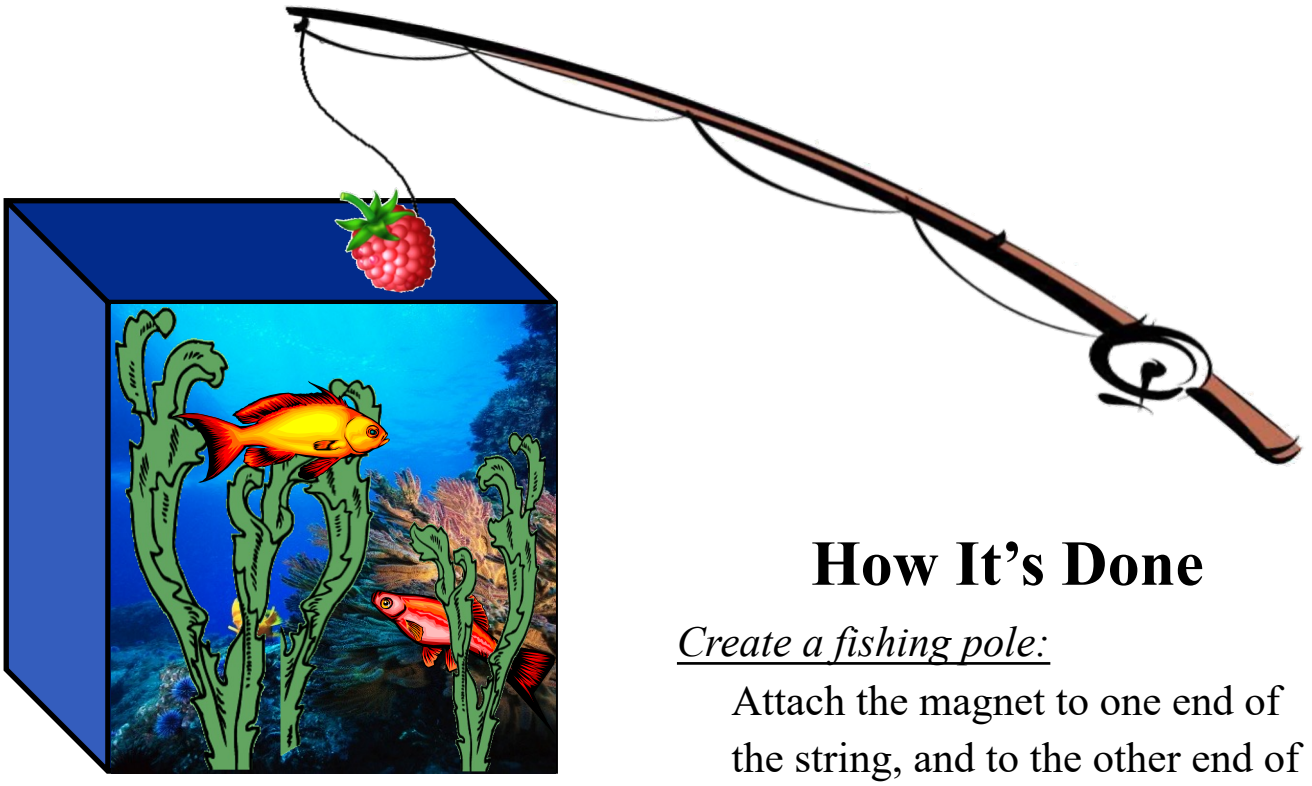
1. Pour cake mix into mixing bowl and add oil.
2. Add sparkling water and mix well.
3. Pour mixture into greased cake tin and let stand for 15 minutes.
4. Bake in an oven at 350° for 45-55 minutes.
5. When cake has cooled completely, frost with your favorite Phe free frosting.

* 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 "Y*es*" and "*No*" foods.



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

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.

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.

Materials Needed

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

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.

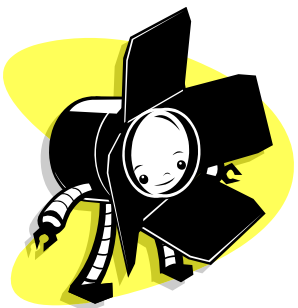
Create a shopping environment:

- 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.

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

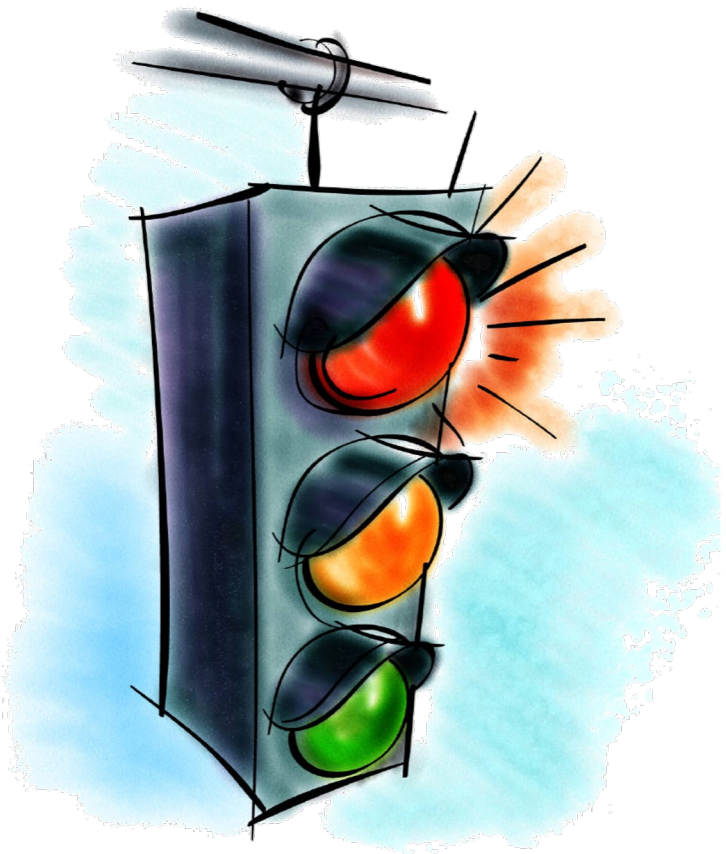
Create an interactive environment:

- 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.



Materials Needed

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

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

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

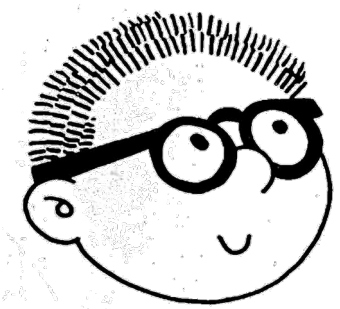
- Discuss the terms "alike" and "different" and use these terms in relation to physical characteristics.
- Compare your physical traits to those of others in the room, pointing out ways you are alike and ways you are different.
- Identify a feature in yourself that is different than that seen in other 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.

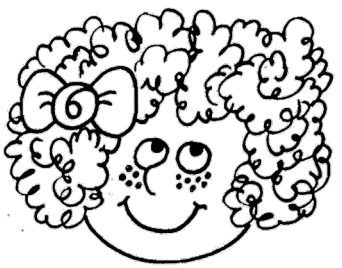
No Materials Needed

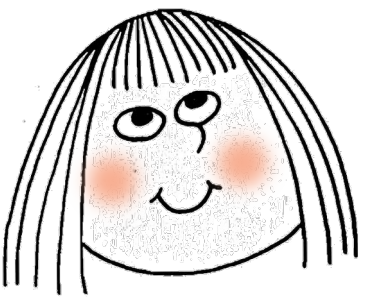
TEACHING AID

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

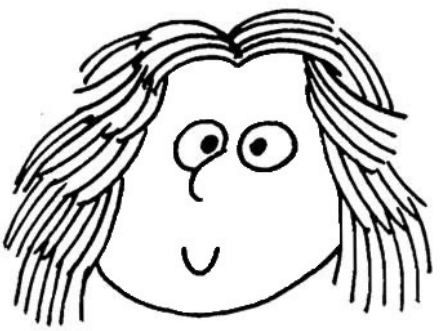
Adapted from March of Dimes Activity Master 2

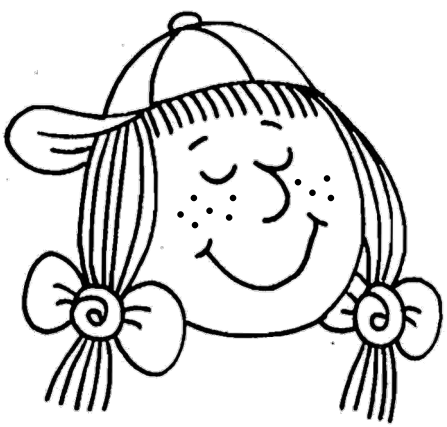
HANDOUT: We Are Alike & Different

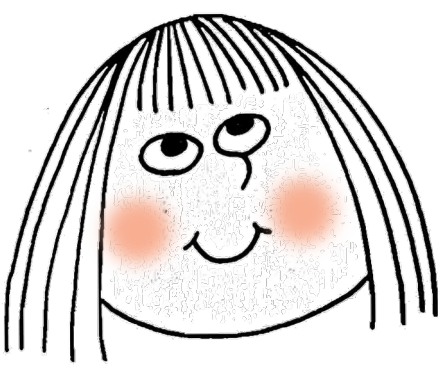
Draw a circle around all the children who have curly hair.

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

Draw a triangle around all the children who have freckles.

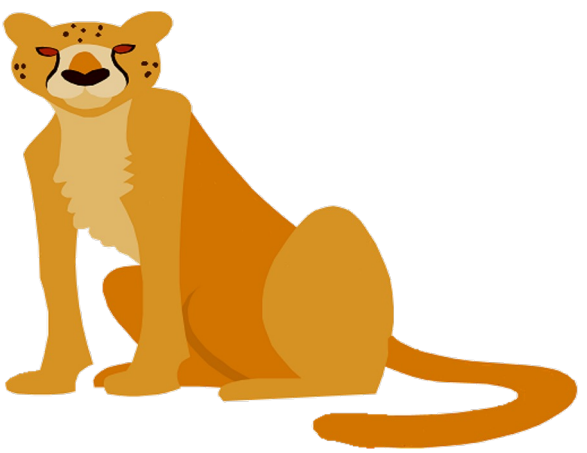





HANDOUT: Alike & Different

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!!

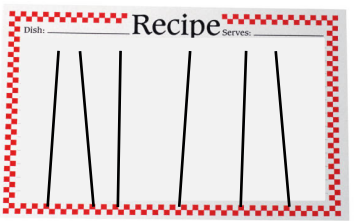
Handout Needed
My Recipe Gene Book

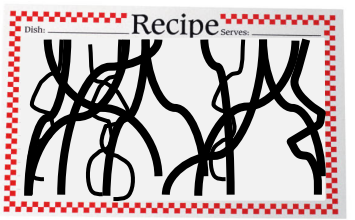
HANDOUT: My Genetic Recipe Book

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).

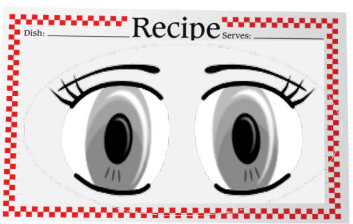
Gene For Hair Texture



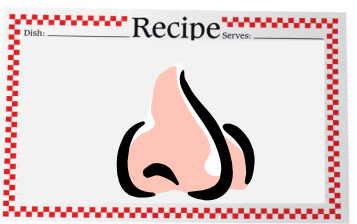


Gene For Hair Color

Gene For Eyes



Gene For Nose



Gene For PKU

Gene For HFI

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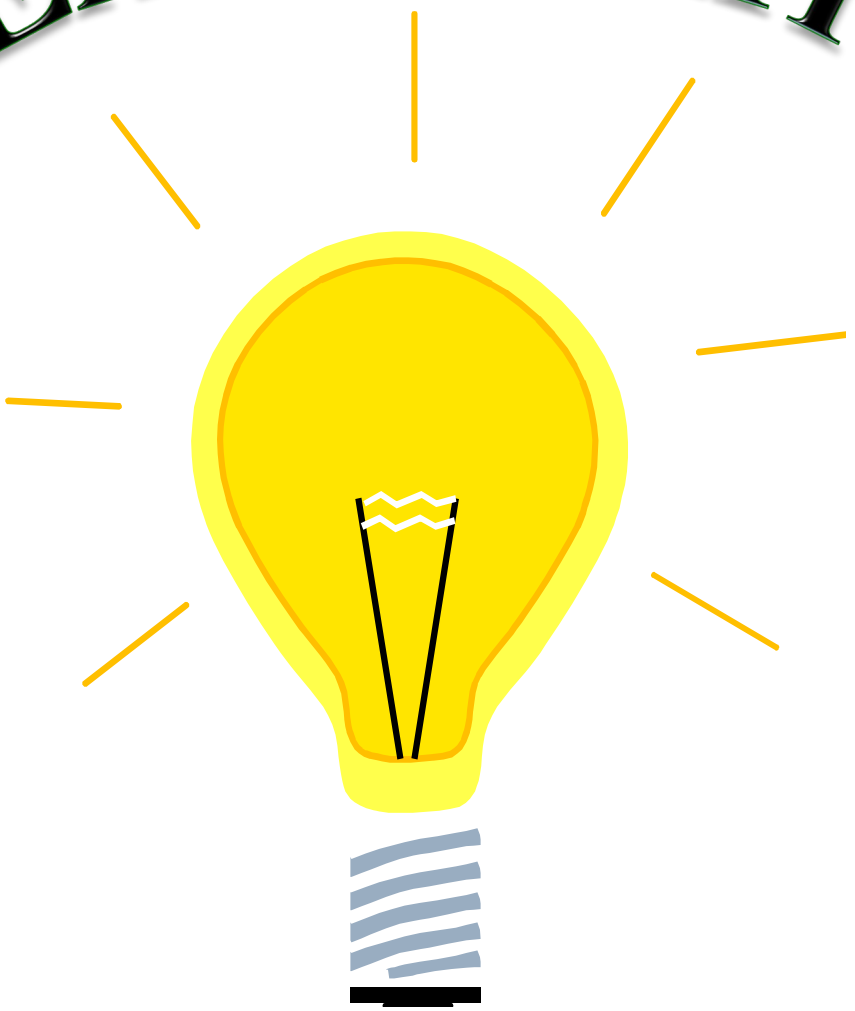
EAT RIGHT



STAY BRIGHT™

Guide For Hyperphenylalaninemia

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CHAPTER TWO

The Elementary School Years

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: Introduction

Children of school age should be active participants in their health care. To successfully involve the children, strategies should be oriented to their physical, cognitive, emotional, social and psychological development. In each of these areas there is progression during the elementary school years, and a solid base of knowledge about these parameters is useful. This is a critical time for children with metabolic disorders to adapt successfully to their disorder. Children who feel good about themselves are better equipped to withstand peer pressure.

Children need both the freedom of personal expression and the structure of expectations and guidelines so that they can accept and begin to participate responsibly in the management of their disorder. Developing an understanding of their metabolic disease can be facilitated by acquiring an age appropriate knowledge in areas of anatomy, physiology, biochemistry and genetics, as they relate to hyperphenylalaninemia. We continuously work to promote age appropriate independence by encouraging developmentally appropriate decision-making. We hope that some of the following suggestions help health professionals, families and the child to work together to foster their emerging independence.

We have created three separate checklists that address the unique issues of each role:

- **Parents**

  Parents remain the front line providers of care during the elementary school years.

- **Children**

  With the child's increasing age and cognitive abilities, he also becomes an integral player in management, and the health professional should speak directly with the child.

- **Professionals**

  A professional checklist has been developed to remind us that the care of chronic diseases requires a sensitive and creative approach.

CHECKLIST: Parents

Planning For The Future

- ☐ Genetics
  - Future pregnancies
- ☐ Carrier testing for extended family
- ☐ Diet for life
- ☐ Adverse effects of elevated levels
  - MRI findings
  - Neurological findings
  - Personality changes
  - School and social performance
- ☐ Maternal PKU

Phe Levels, Growth Charts, Interim History

- ☐ Interim levels
  - Phenylalanine (Phe)
  - Tyrosine (Tyr)
- ☐ Intercurrent illness
- ☐ Growth

Daily Living Routine[1]

- ☐ Weighing, measuring, and preparing formula
- ☐ Cooking/recipes
- ☐ Diet records[2] (see teaching aids/handout)
- ☐ Blood draws (including time of draw in relation to food/formula intake)
- ☐ Family integration

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

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CHECKLIST: Parents

Psychosocial Issues

- ❑ School
- ❑ Peers
- ❑ Siblings
- ❑ Manipulation/power struggles
- ❑ Over-commitment/martyrdom
- ❑ Overprotection
- ❑ Family communication
- ❑ Parental attitudes
- ❑ Self-esteem
- ❑ Finances[3]
- ❑ Impact of diet on family life[4]
- ❑ Realistic expectations

Nutrition Intervention

- ❑ Use of the following items
  - Low protein foods
  - Low protein recipe books (V. Schuett)
  - Gram scale, bread machine, etc.
  - Low protein food lists
- ❑ Childcare/school/aftercare (see teaching aids/handouts)
  - Yearly education of school staff and childcare providers
  - Variety at home and school
  - Formula intake schedule at home and school[5]

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3

CHECKLIST: Children [6]

Age Appropriate Continuing Education

- ☐ Anatomy
- ☐ Biology
- ☐ Biochemistry
- ☐ Genetics

Phe Levels, Growth Charts

- ☐ Phenylalanine levels
- ☐ Tyrosine levels
- ☐ Heights and weights

Daily Living Routine

- ☐ Weighing, measuring, and preparing formula[7]
- ☐ Cooking/recipes[8]
- ☐ Diet records[9] (see teaching aids/handout)

Psychosocial Issues (see teaching aids/handout)

- ☐ Self esteem
- ☐ Siblings
- ☐ School[10]
- ☐ Peers
- ☐ Family dynamics and communication[11]
- ☐ Power struggles
- ☐ Sports/hobbies

Diet For Life

- ☐ Adverse effects of elevated levels (see teaching aids/handout)
- ☐ Attitude

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CHECKLIST: Professionals

Sound Educational Strategies [12]

- ❑ Passive - handouts, posters, videos, etc.
  - One on one and/or peer supported group
- ❑ Interactive - activity sheets, role play
  - One on one and/or peer supported group

Sound counseling [13]

- ❑ Empathy
- ❑ Genuineness
- ❑ Unconditional positive regard

Positive focus

- ❑ Feedback[14]

Empower parents [15]

Experience & Thoughts

1. As the child grows and matures, the daily living routines should reflect increased participation in and responsibility for management of PKU.

2. Delineation of roles becomes an important factor for good compliance. Structure facilitates responsibility and accountability. As early as Kindergarten, the parent can encourage the child to participate in diet record keeping.

3. Depending on the state and on the insurance provider, the cost of formula, low protein foods, and laboratory tests may impact the family dynamics.

4. Has the family returned to or achieved its full potential? Has there been traveling, camping, parental trips without children? Are all family members' needs being met; not just the child who is on the diet?

5. It has been our experience that children often finish the formula in the morning "to get it over with" or, conversely, delay intake until the evening. Without questioning the specific formula schedule this pattern would not be identified.

6. To achieve free flowing dialogue between staff and children, use age appropriate terminology. The use of fun phrases can break the ice and the generation gap.

7. How much is the child participating in weighing, measuring, and preparing formula? As age increases so should the expectations (i.e.: add the ingredients, pushing the blender button).

8. Safety in the kitchen is a major consideration. Level of participation is age and ability dependent. A good starting point is with reading the recipes and measuring the ingredients. Then move on to greater responsibility in the kitchen.

9. Diet records are very important in understanding the diet prescription. A child's participation in record keeping should start as early as Kindergarten. Encourage finding a middle ground. For example during the school year, let them be responsible for record keeping on Saturdays, Sundays and holidays. During the summer months, responsibility could be shifted to 2 days a week. These responsibilities should increase with age.

10. Begin and maintain discussions about peer pressure, feeling different, and self esteem.

11. The family is the child's primary support system. Always encourage communication. Invite all people involved to clinic.

12. Using a variety of approaches to teaching biochemistry, genetics and nutrition are vital to achieving success.

13. The psychologist, Carl Rogers, lists these three qualities in the effective counselor.

14. Provide the family with feedback relating to observations made during clinic appointments. This feedback can be based on your expectations for the child's age. Solicit feedback from family regarding clinic experience as well.

15. Empower parents by keeping them informed on resources that are available to their child. Ensuring their understanding in all areas of their child's disease creates an independence and makes interaction with the clinic more valuable. This places responsibility back into the hands of the family.

PRINCIPLES OF DIET PRESCRIPTION

Throughout a Lifecycle

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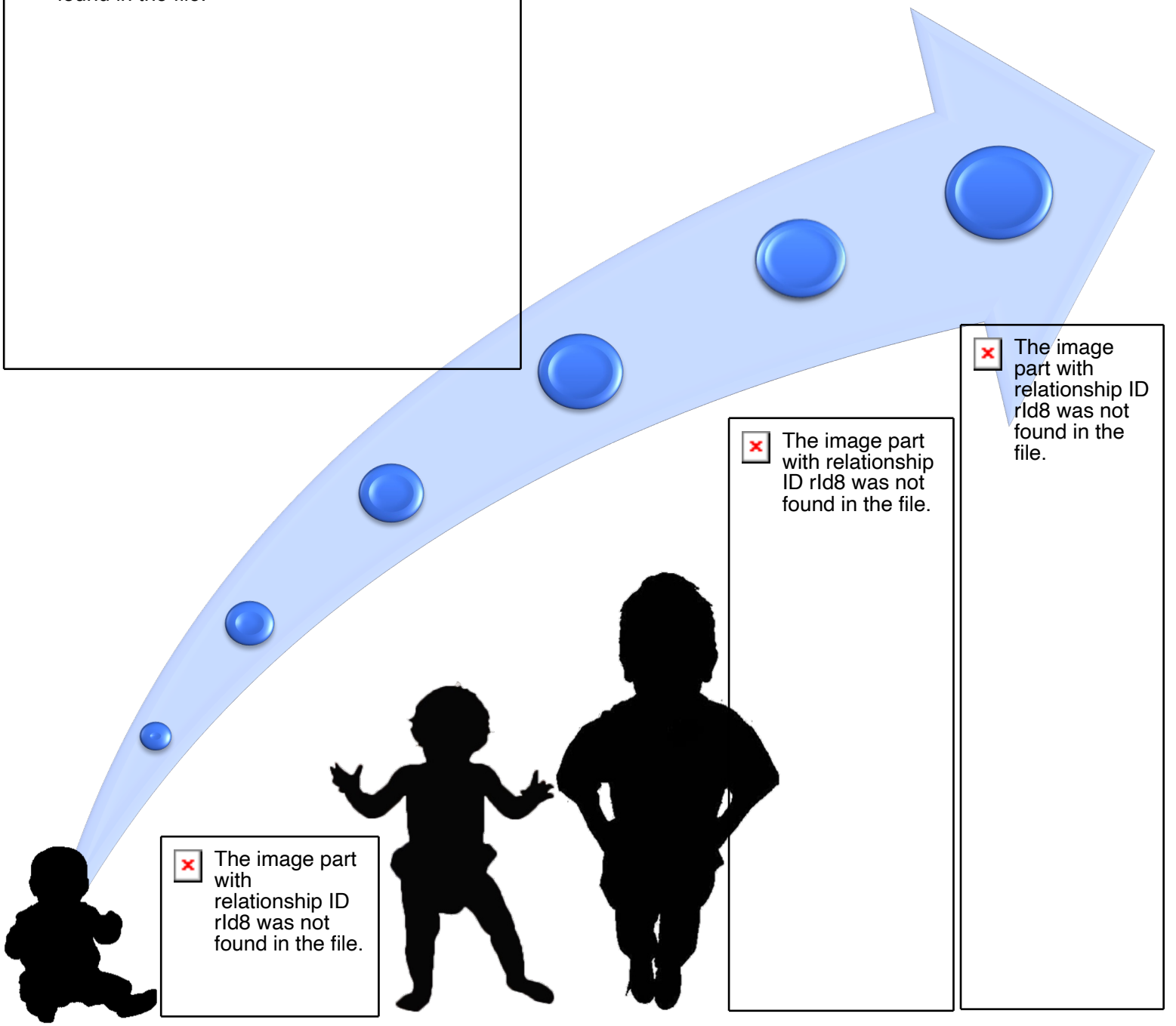

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Birth to 6 Months | 6 to 12 Months | 1 to 2 Years | 2 to 7 Years | 8 Years to Adulthood

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

PRINCIPLES OF DIET PRESCRIPTION

Throughout a Lifecycle

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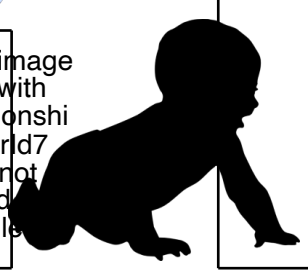


Birth to 6 Months

6 to 12 Months

1 to 2 Years

2 to 7 Years

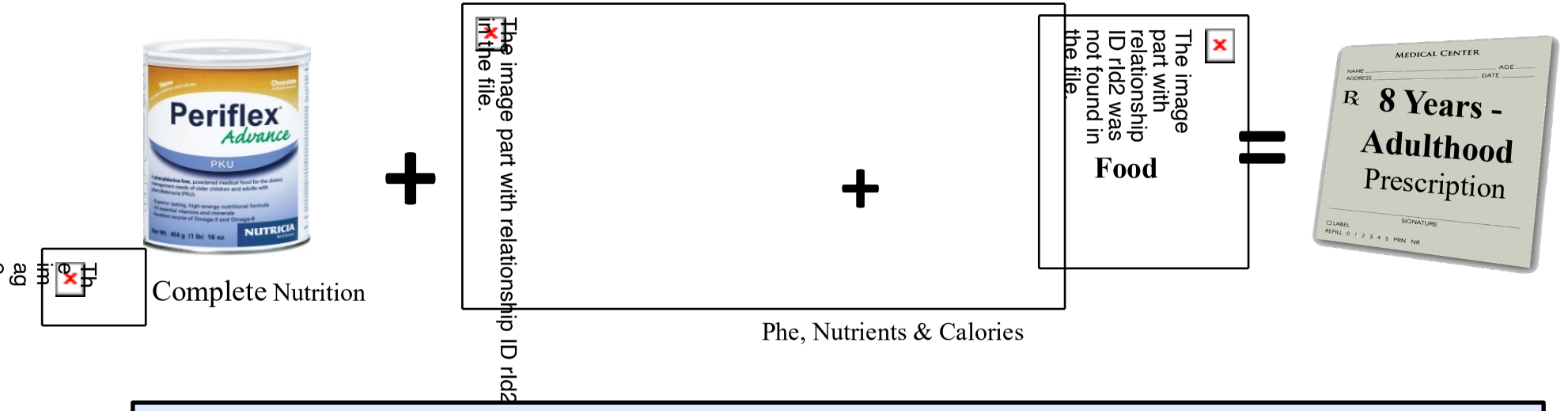
8 Years to Adulthood

Motherhood

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

PRINCIPLES OF DIET PRESCRIPTION

Elementary School Years



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

2 to 7 Years Old

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Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

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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: 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.
Formula with a milk source, in addition to low protein and other solid foods provide additional amino acids, vitamins, minerals, and calories.

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DAILY DIET PRESCRIPTION

8 Years Old Through Adulthood

+

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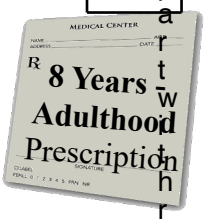

Phe Complete Nutrition

Phe, Amino Acids, Vitamins, Minerals & Calories

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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: 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/Height:**

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

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

Additional Notes:

_____

_____

TEACHING AID

Modified Food Pyramid

Objective: To provide individuals on diet with an understanding of how their formula plays a role in their diet.

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Materials Needed

- Crayons, Colored Pencils, or Markers.

Handouts:

My Pyramid For Kids
PKU Pyramid Coloring Page

How It's Done

Present the standard food pyramid

- Discuss the importance of various food groups.
- Incorporate formula as an essential nutritional component.
- Discuss how their formula becomes a large portion of their pyramid, replacing high protein foods such as the traditional milk and meat sections on the pyramid (light blue and purple).

Create a pyramid:

Have the children fill in their own pyramid, drawing their favorite foods in each food group and their formula in the medical food section.

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My PKU Pyramid

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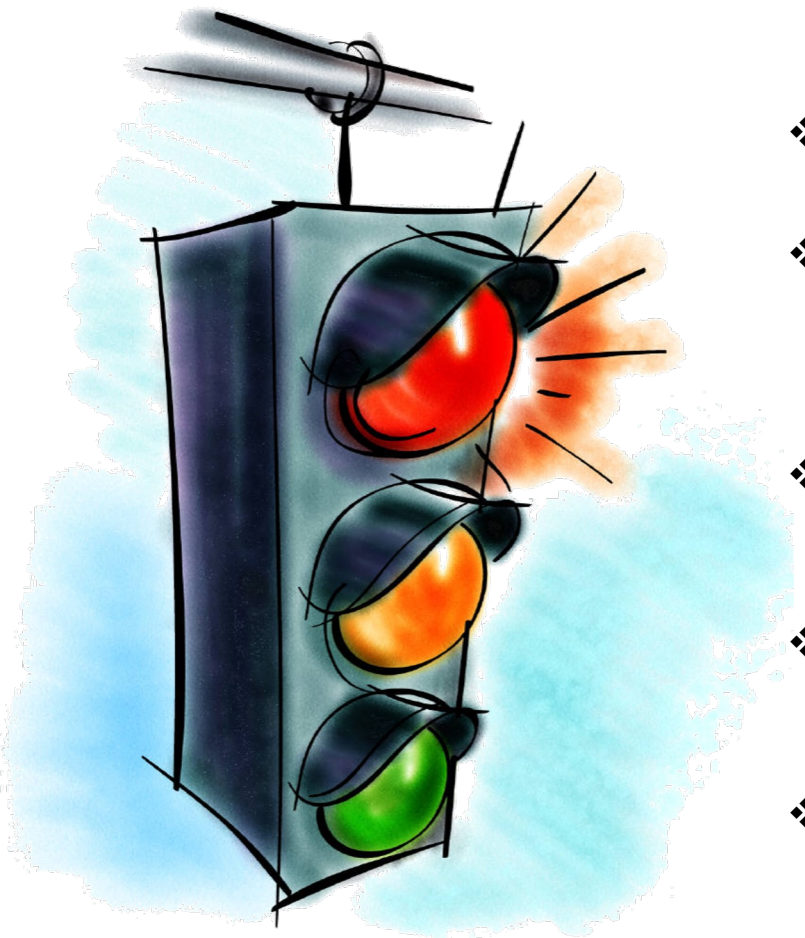
TEACHING AID

Red Light!!! Yellow Light!! Green Light!

Objective: To reinforce "*Yes*", "*Sometimes*", and "*No*" foods.



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, yellow for "Sometimes" foods, and red for "No" foods.
- Use felt backed food models and let the children place them in the appropriate section.
- This activity can also be reversed by having individuals identify foods already placed within the red, yellow or green circles as being misplaced or appropriate.
- A positive reward system is used with all of these programs:
  - Stickers
  - Buttons
  - Verbal affirmation
  - Low protein treats

Materials Needed

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

TEACHING AID

Clinic Supermarket

Objective: To increase the child's involvement in their diet.

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Materials Needed

- "Clinic Supermarket" sign
- Pens or pencils
- Calculators (optional)

Handout:
Supermarket Shopping List

How It's Done

Create a supermarket environment:

- Set up supermarket sign and food models like a grocery store.
- Each child will write a shopping list for a single meal (breakfast, lunch, or dinner) or a one day diet.
- Help each child to look up the Phe content of the foods they selected. Provide pen, paper, and calculator (optional) for Phe and protein calculations.
- Compare this information with their diet prescription and discuss the results with the child.
- This concept can be expanded upon based on age to include buying foods for a recipe, etc.

Supermarket Shopping List

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What meal are you making? *Circle One*

Breakfast Lunch Dinner Snack

What is on the menu?

_____

What ingredients do you need? How much do you need?

Example: Low Protein Cheese 1 slice 19 gm 20 mg Phe 0.4 gm Pro.

_____ gm _____ mg Phe _____gm Pro.

_____ gm _____ mg Phe _____gm Pro.

_____ gm _____ mg Phe _____gm Pro.

_____ gm _____ mg Phe _____gm Pro.

_____ gm _____ mg Phe _____gm Pro.

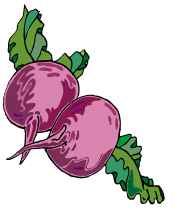
_____ gm _____ mg Phe _____gm Pro.

_____ gm _____ mg Phe _____gm Pro.

_____ gm _____ mg Phe _____gm Pro.

_____ gm _____ mg Phe _____gm Pro.

Total: _____ mg Phe _____gm Pro.



Does this menu fit your diet prescription?

YES or **NO**

TEACHING AID

Dramatic Play: Restaurant Setting

Objective: To increase the child's involvement in their diet.

Chef's Low Protein Specials

- **Wel-Plan Tortilla Chips****
- **Dietary Specialties Nacho Cheese Dip**
- **Low Protein Pasta with Alfredo Sauce**
- **Crispy Apple Coleslaw with Add-Ins***
- **Chocolate Cake***

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Materials Needed

- Paper or plastic food models
- Pens or pencils
- Order/note pad for waiter
- Calculator
- Apron/chef's hat

Handouts:

Menu Template
Menu Ideas
Food Models

How It's Done

Set up a restaurant-like situation

- Have all participating individuals play a role: customer, waiter or waitress, chef or cashier.
- All individuals should have the opportunity to play various roles

  The Customer orders the food from a menu on which the Phe content is listed.

  The Waiter or Waitress takes the order and serves the food.

  The Chef prepares the order by selecting models of the food ordered and placing them on a plate or tray.

  The Cashier rings up the amount of Phe and Protein ordered.

- Use this opportunity to begin a discussion about the Phe content of the foods, ordering in a restaurant, and monitoring daily intake of Phe .

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

** **Mile High, Low Protein Cookbook.** Low Protein Food Store, IMD Clinic, The Children's Hospital, Aurora, CO.

Menu Ideas

	Low Phe	High Phe
Breakfast	Fruit LP Cereals LP Pancakes LP Toast Juice Formula Coffee/Tea	Egg Dishes Yogurt Milk
Lunch	Salads LP Soups LP Sandwiches Juice Soda Formula Coffee/Tea	Salads Soups Sandwiches Milk
Dinner	Salad French Fries LP Pasta LP Tacos Juice Formula Coffee/Tea	Salads Pizza Burger Milk
Dessert	Fruit LP Cookies LP Cake LP Ice Cream	Ice Cream Pie Cake

LP = Low Protein

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Chef's Low Protein Menu

TEACHING AID

Role Playing

Objective: To increase the child's involvement in their diet.

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Lights...

Camera...

Action

Materials Needed

- Paper or plastic food models

How It's Done

Set up a school lunchroom situation

- Have each child line up, walk along side the table, and receive/choose a lunch.
- Continue the role play by sitting at a table together while "eating lunch".
- This activity may be modified by having the children bring their lunch box or providing them with a paper bag to fill with models of food representing a typical lunch they might bring to school.
- Facilitate discussion about their food choices and how it relates to their diet prescription.
- Also address the idea of trading food with their classmates.

TEACHING AID

Energy Sources

Objective: To introduce food and formula as essential forms of energy for use by the body.

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How It's Done

Use the handout "Energy Source Matching"

- The children are presented with two groups of pictures:
  - One group of pictures representing different kinds of fuel.
  - The second group representing users of the fuel.

For example:

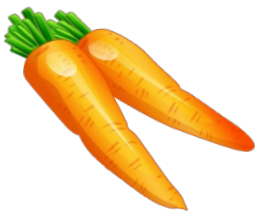
- Gasoline for a car
- Carrots for a rabbit
- Battery for flashlight
- **Formula for children with PKU**

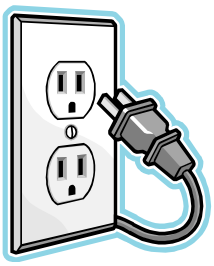
- Ask them to match the fuel with the user and discuss why and how each uses the source for energy.

Handout Needed
Energy Source Matching

Energy Source Matching

Draw a Line to Connect the Energy Source to its User





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Formula

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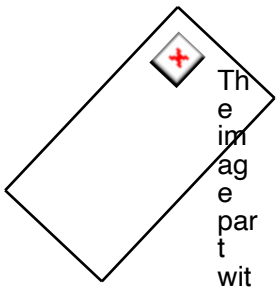

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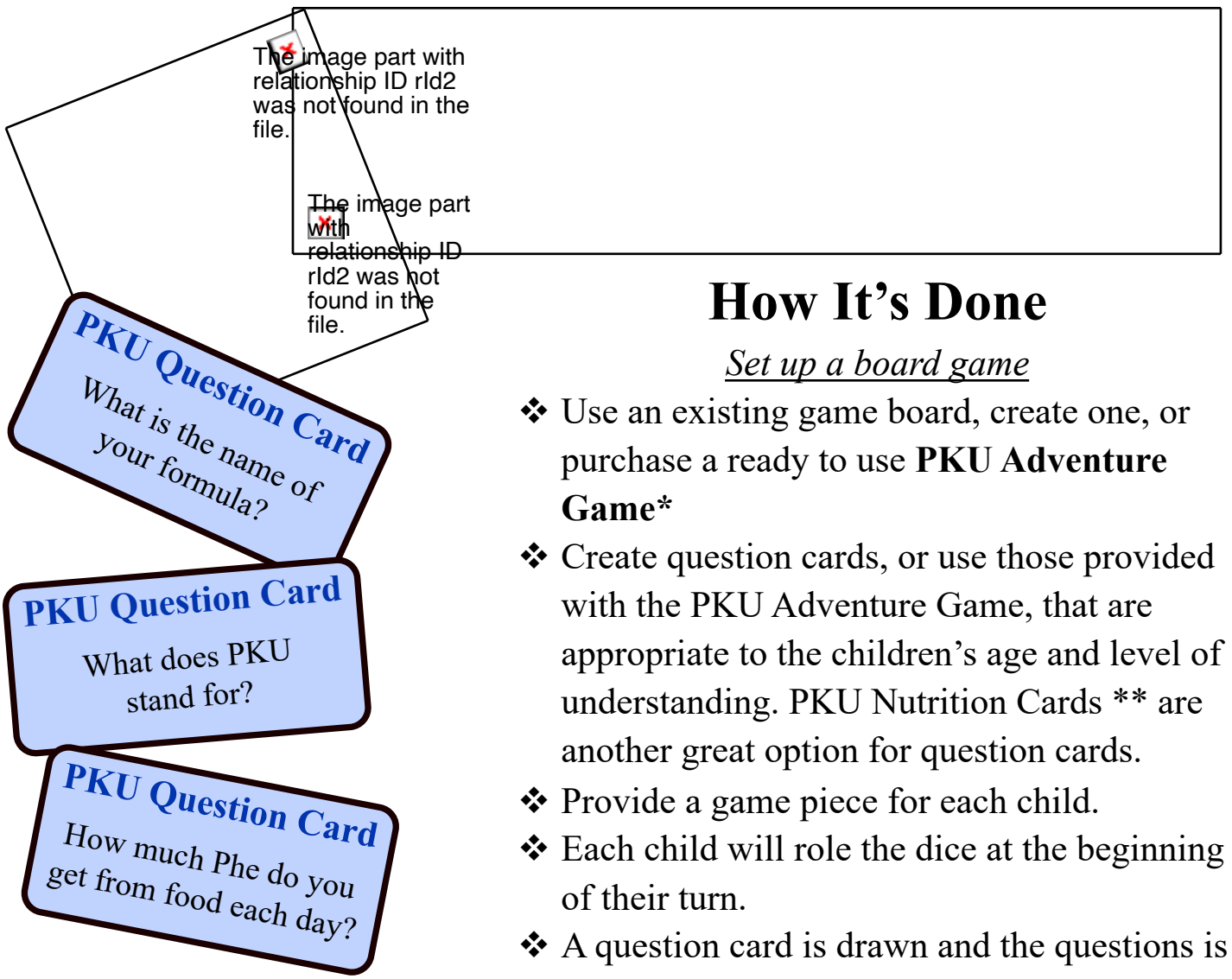
TEACHING AID

PKU Adventure Game

Objective: To assess the Hyperphenylalaninemia knowledge base while providing a fun learning environment.



How It's Done

Set up a board game

- Use an existing game board, create one, or purchase a ready to use **PKU Adventure Game***
- Create question cards, or use those provided with the PKU Adventure Game, that are appropriate to the children's age and level of understanding. PKU Nutrition Cards ** are another great option for question cards.
- Provide a game piece for each child.
- Each child will role the dice at the beginning of their turn.
- A question card is drawn and the questions is asked.
  - A correct answer allows the player to move their game piece ahead the number of spaces determined by the number they rolled on the dice.
  - An incorrect answer results in no movement of the game piece.

Materials Needed

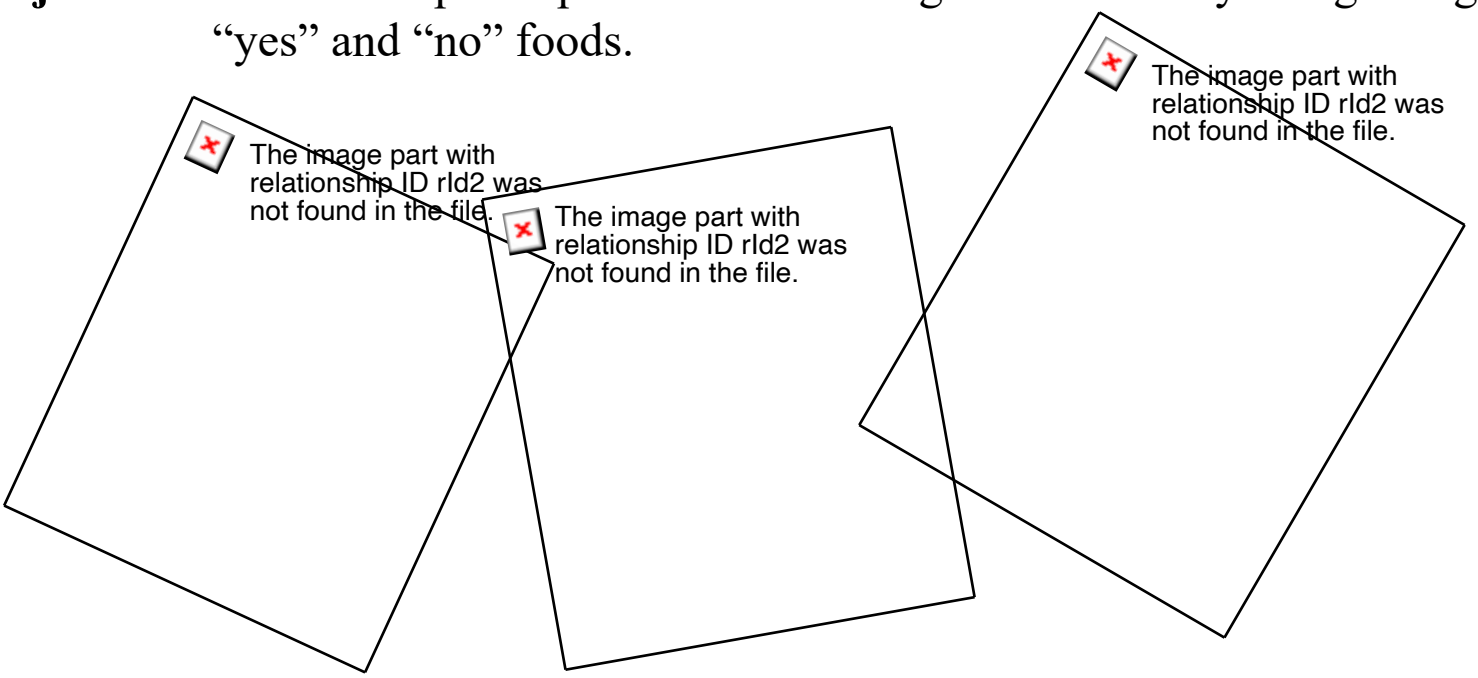
- PKU Adventure Game
- PKU Question Cards
- Dice
- Playing pieces

*** PKU Adventure Game.** Available through the Low Protein Food Store, IMD Clinic, The Children's Hospital, Aurora, CO.
**** PKU Nutrition Cards**. Available through Nutricia North America Web. <http://www.shsna.com/>.

TEACHING AIDS

"Yes" "No" BINGO

Objective: To increase participants understanding of their diet by recognizing "yes" and "no" foods.



How It's Done

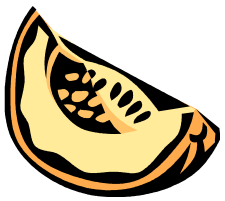
This game can be as simple or complex as you decide to make it. For younger participants, limit discussion to "yes", "no", and "sometimes" foods. If participants are older, lead discussion to include topics such as serving size, cooking method, etc.

- Using the cut out game pieces mixed in an envelope, draw one square and call the name of the food out load. Have participants mark the appropriate square with a piece of candy, and have the participants say if it is a yes, no, or sometimes food (for older participant, begin to discuss what a typical serving size is and how much Phe is in one serving).
- Once a participant has five in a row (in any direction) they should shout "BINGO". To prove they have a BINGO they will need to read off the foods that make up their BINGO, and say if the food is a yes, no, or sometimes food.
- Game boards can be shuffled or traded between participants and all game pieces put back into the envelope , this game can be played until all the foods have been discussed.

Materials Needed
- Bingo Game Pieces
- Bingo Game Boards
- Candy (as markers)

BINGO GAME PIECES

Cut out these pictures, mix them in an envelope.

	The image part with relationship ID rId3 was not found in the file.	The image part with relationship ID rId3 was not found in the file.		The image part with relationship ID rId3 was not found in the file.
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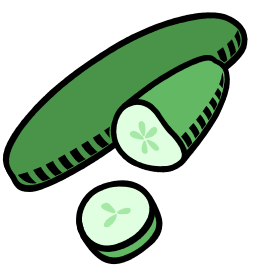
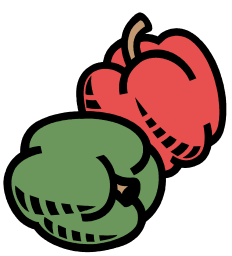
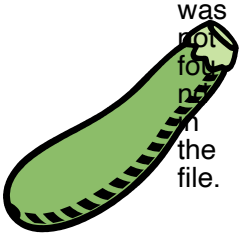
The image part with relationship ID rId3 was not found in the file.			The image part with relationship ID rId3 was not found in the file.	The image part with relationship ID rId3 was not found in the file.
The image part with relationship ID rId3 was not found in the file.		The image part with relationship ID rId3 was not found in the file.	The image part with relationship ID rId3 was not found in the file.	
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BINGO GAME BOARD

Place candy on the appropriate square and discuss each food until you get five in a row.

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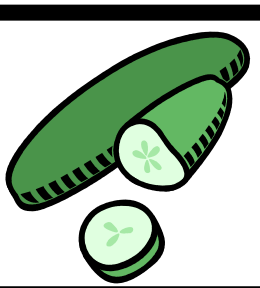
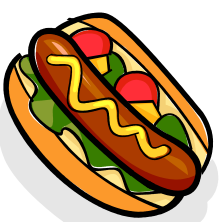
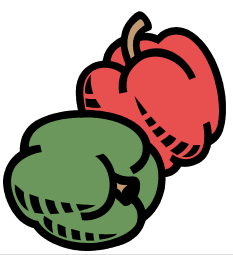
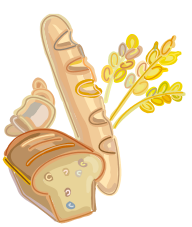
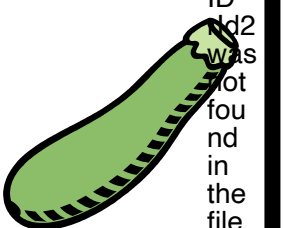
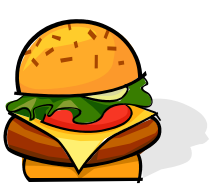
The image part with relationship ID rId2 was not found in the file.		**FREE SPACE**		The image part with relationship ID rId2 was not found in the file.
		The image part with relationship ID rId2 was not found in the file.		
The image part with relationship ID rId2 was not found in the file.		The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.

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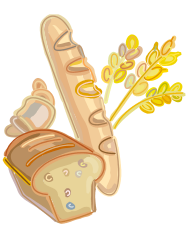
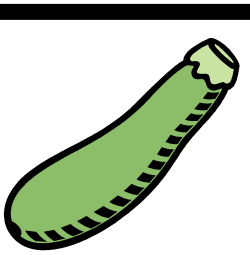
The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.		The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.
The image part with relationship ID rId2 was not found in the file.				

BINGO GAME BOARD

Place candy on the appropriate square and discuss each food until you get five in a row.

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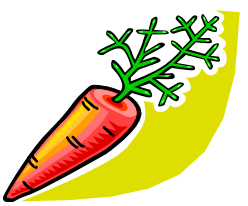
The image part with relationship ID rId4 was not found in the file.	The image part with relationship ID rId4 was not found in the file.			The image part with relationship ID rId4 was not found in the file.
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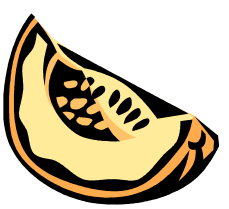
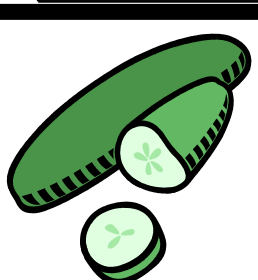
The image part with relationship ID rId4 was not found in the file.	The image part with relationship ID rId4 was not found in the file.	The image part with relationship ID rId4 was not found in the file.		
	The image part with relationship ID rId4 was not found in the file.	The image part with relationship ID rId4 was not found in the file.	The image part with relationship ID rId4 was not found in the file.	The image part with relationship ID rId4 was not found in the file.

BINGO GAME BOARD

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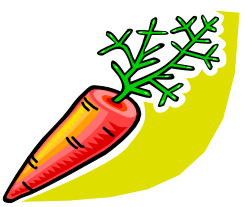
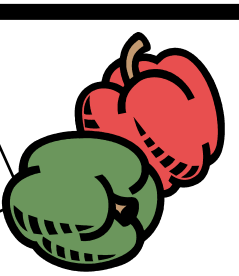
		The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.	
The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.	

BINGO GAME BOARD

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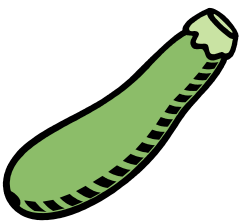
The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.		The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.
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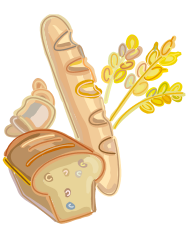
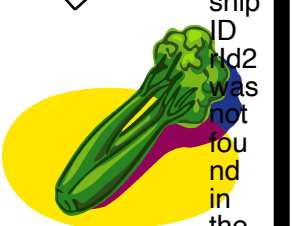
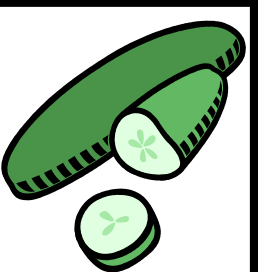
The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.		The image part with relationship ID rId2 was not found in the file.	The image part with relationship ID rId2 was not found in the file.
The image part with relationship ID rId2 was not found in the file.		The image part with relationship ID rId2 was not found in the file.		The image part with relationship ID rId2 was not found in the file.

TEACHING AID

Recipe Preparation

Objective: To introduce the use of recipes and increase the child's level of responsibility.

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Materials Needed

Recipes:

Snacks that Slither
Spooky Snacks

Handouts:

It's A Mix-up
Mix and Match
Grocery Shopping Is A Must!
Answer Keys

How It's Done

Each handout is designed to increase the level of responsibility

- **"It's a Mix-up"**
  Discuss the importance of each step and the obvious missing step of weighing the ingredients.
- **"Mix and Match"**
  Help the child to become familiar with the ingredients and identify what it takes to create a meal.
- **"Grocery Shopping is a Must!"**
  Participants identify "Yes" foods beginning with a specific letter that can be used for meals, recipes or as snacks. Enhance this exercise by visiting the kitchen in your hospital or clinic.
- **"Have Snacks that Slither!"**
  Actually make the recipe! Identify and gather ingredients, weigh/measure, follow directions, count Phe and enjoying the finished product.
- This can be done using recipes from the **Mile High Low Protein Cookbook©**

Mile High, Low Protein Cookbook. Low Protein Food Store, IMD Clinic, The Children's Hospital, Aurora, CO.

It's A Mix-up

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Cut out the images
&
organize them into
the correct order.

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It's A Mix-up

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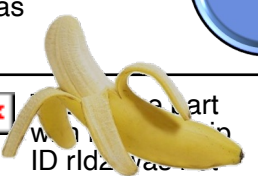

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ANSWER KEY: It's A Mix-up

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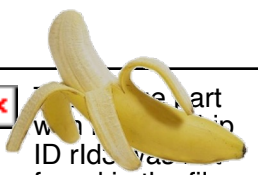

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Mix & Match

What should the Chef serve for dinner?

Draw a line between each dish and the ingredients needed to make it.

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LP Cheese
LP Lasagna Noodles
Tomatoes
Mushrooms

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LP Pizza Dough
LP Cheese
Mushrooms
Tomato Sauce

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LP Taco
Taco Shell
Lettuce
Tomato
LP Cheese

Grocery Shopping's A Must

The Chef was in a hurry to get to the store. He only wrote down the first letter of each food item, and now he can't remember what to buy!

Help by listing several foods allowed in your diet that begins with the letter provided.

Pizza Toppings

B_____

P _____

M _____

G _____

S_____

Colorful Fruits

B_____

P _____

M _____

G _____

S_____

Breakfast Foods

B_____

P_____

M_____

F_____

C_____

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Pizza Toppings

Color Fruits

Breakfast Foods

ANSWER KEY: Grocery Shopping's A Must

This is not an all inclusive list, just suggestions for foods that could work as answers. Encourage the children to think of their own favorite foods.

Pizza Toppings

Bell peppers, broccoli, black olives

Pineapple, peppers

Mushrooms, mozzarella*

Garlic, green olives, green peppers

Sauce, spinach

Colorful Fruits

Bananas, berries, blackberries, blueberries

Peaches, pineapple, pears, plums, prunes

Mango, melon

Grapefruit, grapes

Star fruit, strawberries

Breakfast Foods

Banana, bagels*

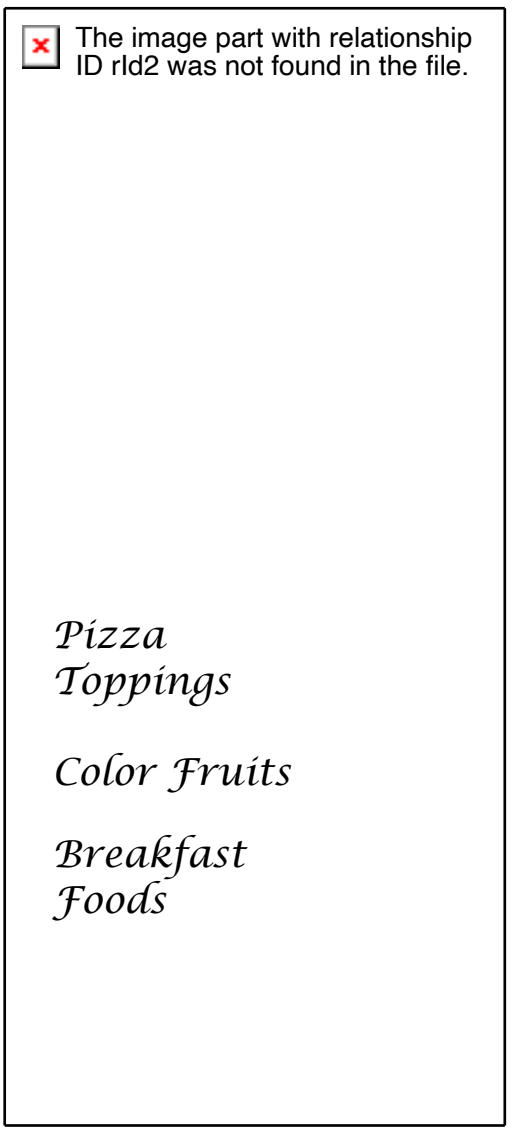
Peaches, pineapple, pancakes*

Melon, muffins*

Formula

Cereal*

*** Indicates a LOW PROTEIN food choice**



RECIPES: Snacks That Slither

A SUPER SILLY SNAKE SANDWICH

Yields: 1 Serving
Phe: 28mg **Protein:** 0.56 g

Ingredients:

19g (1 slice)	Low Protein Cheese
31g (5 each)	Loprofin Crackers*
64g (5 slices)	Apple slices
5g (2 each)	Grapes

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Directions:

1. Cut the cheese into 4 pieces.
2. Lay down a cracker, a piece of cheese and an apple slices
3. Repeat the pattern until you use up all the cheese, crackers and apples.
4. Add a grape for the head and the tail.

DYNAMITE DIRT PUDDING

Yield: 1 Serving
Phe: 23 mg **Protein:** 0.6g

Ingredients:

½ cup	Butterscotch Pudding
57g	Banana, *sliced*
2 each (31g)	Loprofin Chocolate Wafers*
2 or 3	Gummy Worms

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Directions:

1. Put pudding into a cup.
2. Add banana slices.
3. Fill with more pudding.
4. Put a few "Gummy Worms" into the pudding.
5. Top with cracker crumbs.

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

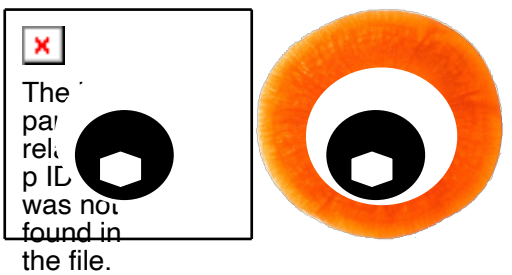
RECIPES: Spooky Snacks

SCARY EYEBALLS

Yields: 1 Serving
Phe: 34 mg **Protein:** 0.8 g

Ingredients:

1 medium	Carrot
2 Tbsp	Low protein cream cheese
3 each	Black olives, pitted



Directions:

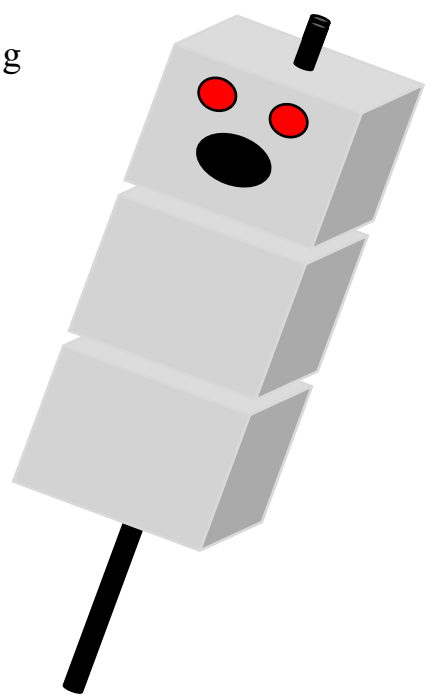
1. Chop carrots into bit size chunks.
2. Top each with a blob of cream cheese.
3. Place halved olives on top of cream.

SPOOKY GHOST

Yields: 1 Serving
Phe: 20 mg **Protein:** 0.49 g

Ingredients:

3 each	Marshmallows
2 each	Cinnamon “Red Hots”
1square	White almond bark
1 tub	Black decorating gel
1 each	Wooden skewer



Directions:

1. Place marshmallows on skewers.
2. Melt almond bark and cover
3. Place Red Hots as eyes.
4. Make a mouth with decorating gel.

TEACHING AID

ABC's

Objective: To be able to identify and spell the name if their metabolic disease.

Materials Needed

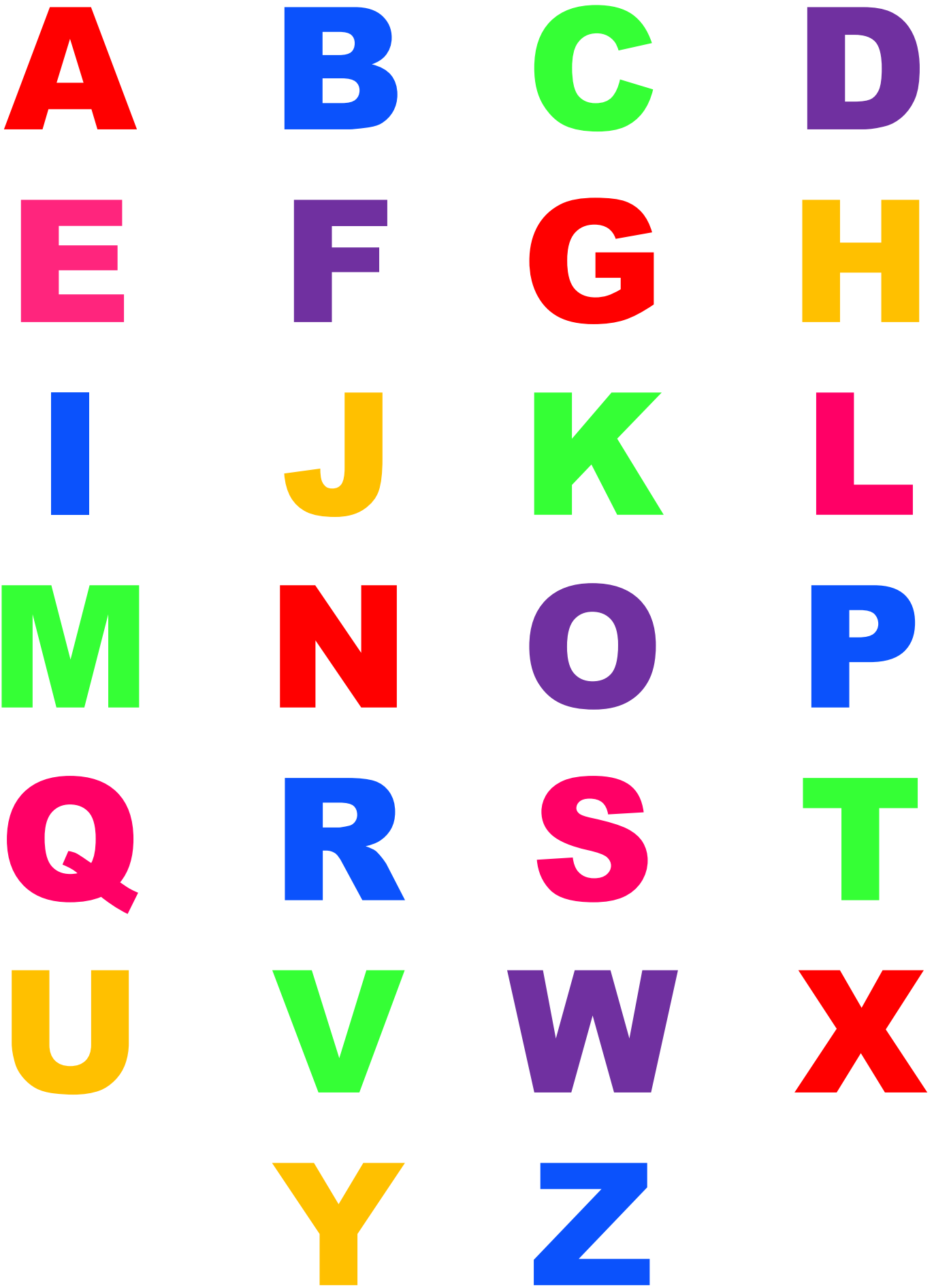
- List of metabolic disorders
- Cut-out letters

How It's Done

Use the "ABC Cutouts" Handout

- Have the children pick out the name of their metabolic disorder from a large list.
- You may choose to point out other metabolic diseases that are managed by diet.
- Using jumbled cut-out letters, ask each child to spell their disorder. This can be done as a group on a felt or black board or individually.
- Use a word find activity including the name of the disease and low Phe foods. Practice writing the disease name.

ABC Cutouts



PKU Word Find

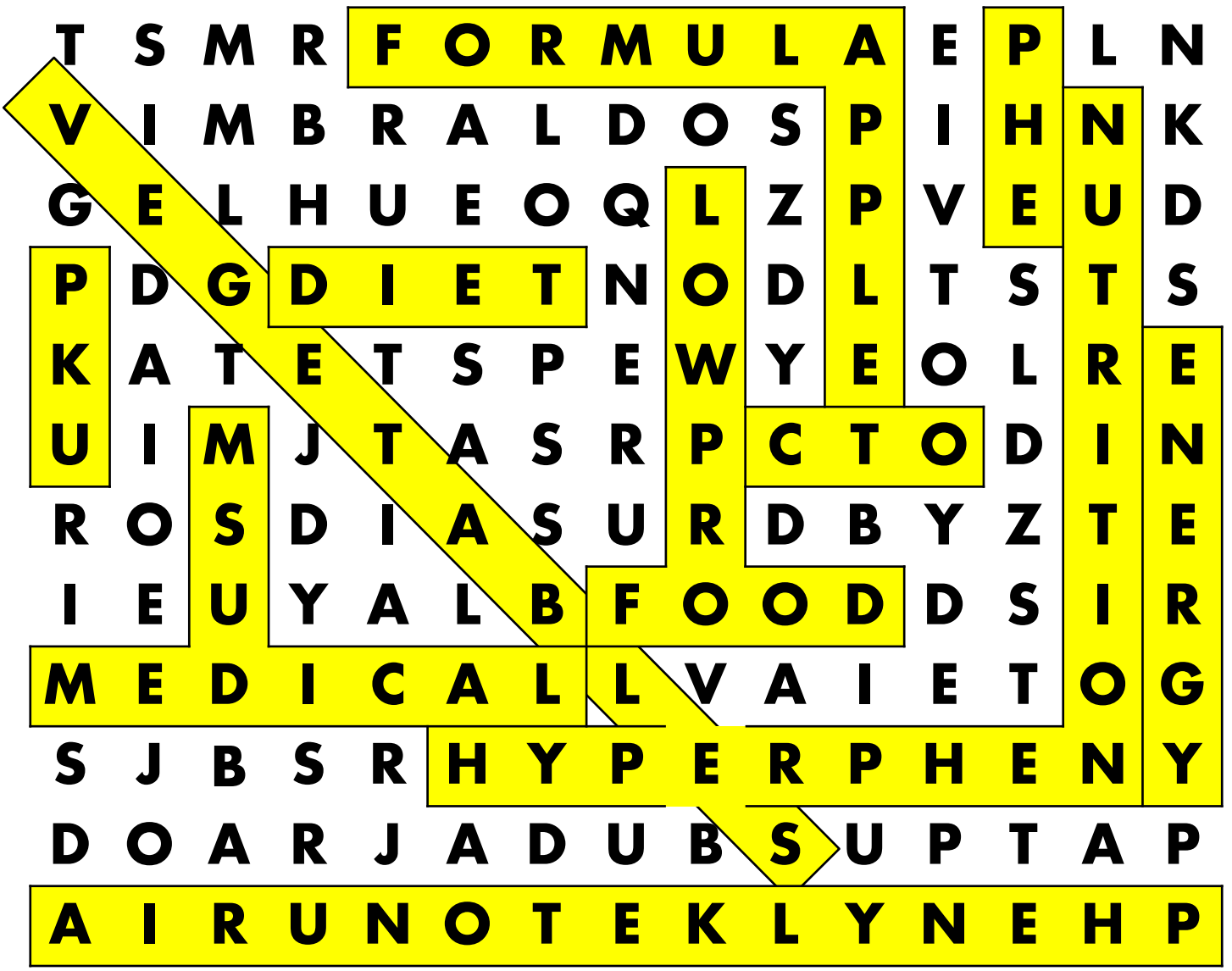
Find the words below.

Words can be horizontal, vertical, diagonal, and even backwards!

T S M R F O R M U L A E P L N
V I M B R A L D O S P I H N K
G E L H U E O Q L Z P V E U D
P D G D I E T N O D L T S T S
K A T E T S P E W Y E O L R E
U I M J T A S R P C T O D I N
R O S D I A S U R D B Y Z T E
I E U Y A L B F O O D D S I R
M E D I C A L L V A I E T O G
S J B S R H Y P E R P H E N Y
D O A R J A D U B S U P T A P
A I R U N O T E K L Y N E H P

APPLE
DIET
ENERGY
FOOD
FORMULA
FRUIT
HYPERPHE
LOWPRO
MEDICAL
MSUD
NUTRITION
OTC
PHE
PHENYLKETONURIA
PKU
VEGETABLES

ANSWER KEY: PKU Word Find



APPLE
DIET
ENERGY
FOOD
FORMULA
FRUIT
HYPERPHE
LOWPRO
MEDICAL
MSUD
NUTRITION
OTC
PHE
PHENYLKETONURIA
PKU
VEGETABLES

TEACHING AID

Herman The Human Pathway

Objective: To introduce the idea of a metabolic pathway.

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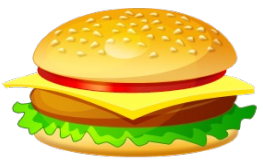

Low Phe vs. High Phe

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Materials Needed

- Cardboard box
- Large tub
- Decorations
- Glue, scissors, etc.
- Small candies or food models
- Large food models

How It's Done

This is an excellent way to teach children what their bodies can and cannot use, and how a block in that pathway can cause unwanted substances to build up and have negative consequences on the body.

Make Herman.

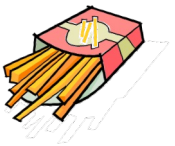
- His torso is made from a box that contains a tube that runs from top to bottom.
- Within the tube is a cross-section of cardboard that has a specific size hole in it. This cardboard restricts the size of food items that can pass all the way through the tube. Food items that are too large will collect within the tube.

Feed Herman

- Have the children feed Herman jelly beans or small candies that will fit through the tube to represent low protein foods. Then have then feed Herman larger, high protein foods that will not fit down his tube.
- Herman's blocked "digestive tube" makes an analogy for a blocked metabolic pathway.

Herman's Maze

Herman is stuck in the middle of a maze.
Help him escape without running into high Phe foods!



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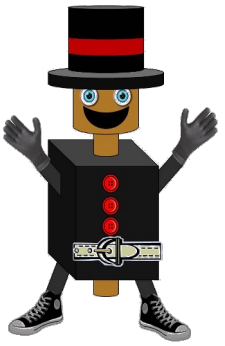

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You Win!

Spaghetti Maze


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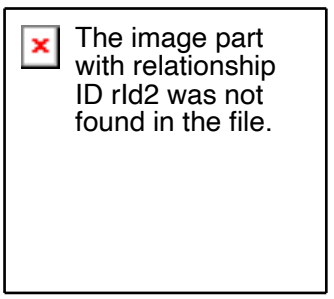
Spaghetti Maze Key

TEACHING AID

The Stuff Between Your Ears

Objective: To introduce brain anatomy and biochemistry.



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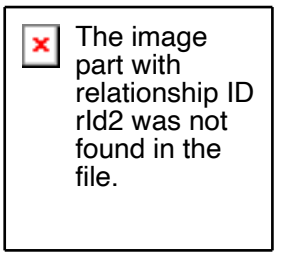
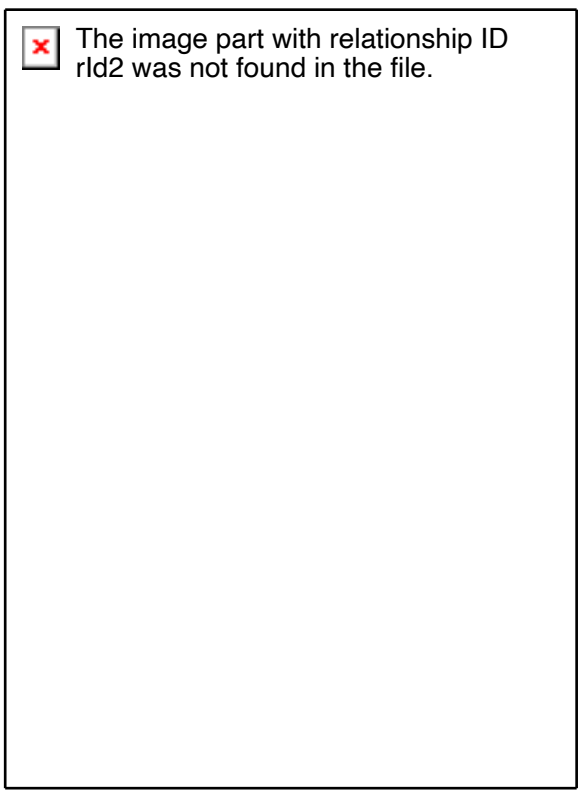




Materials Needed

- Paper or plastic food models

How It's Done

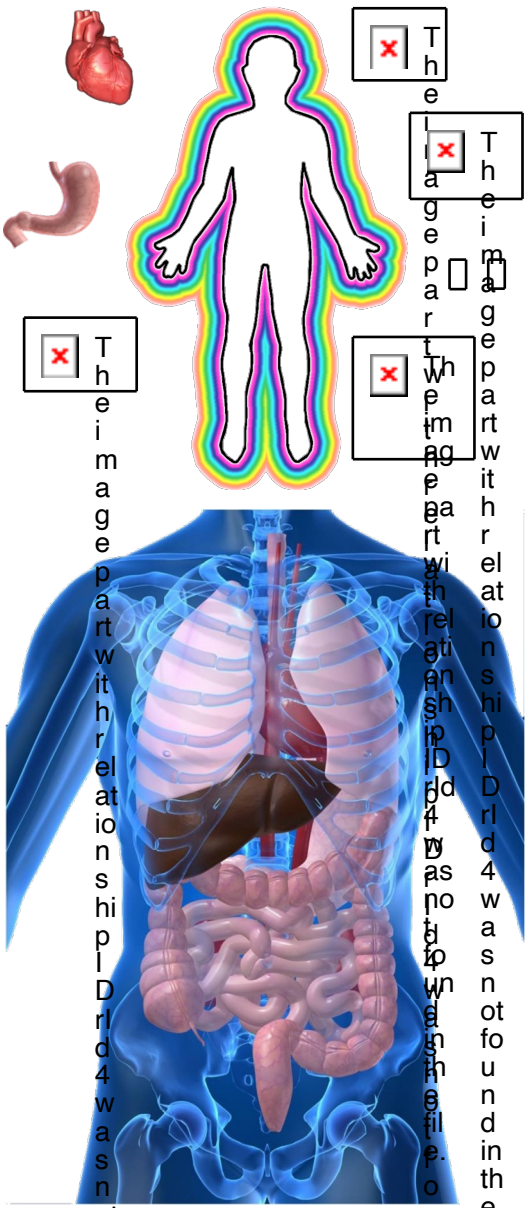
Make a comparison

- A visual aid, such as cauliflower, is a good way to introduce the parts of the brain.
- It is important to discuss:
  - The location and function of both white matter and gray matter.
  - Providing our brains with good nutrition throughout our entire life.
  - What the effects of not following their special diet will be.
- Just as cauliflower requires nutrients and energy to grow, so does our brain. Toxic levels of fertilizer can damage the cauliflower just as excessively high Phe levels can damage the human brain.
- Showing a typical and an atypical cranial MRI may be interesting for older children.

TEACHING AID

Body Parts

Objective: To describe the essential functions of the organs in our body, and to introduce the concept of how they are involved in Hyperphenylalaninemia.



Materials Needed

- Butcher or drawing paper
- Markers
- Glue

How It's Done

This activity can be adapted to a variety of models. There are two main options:

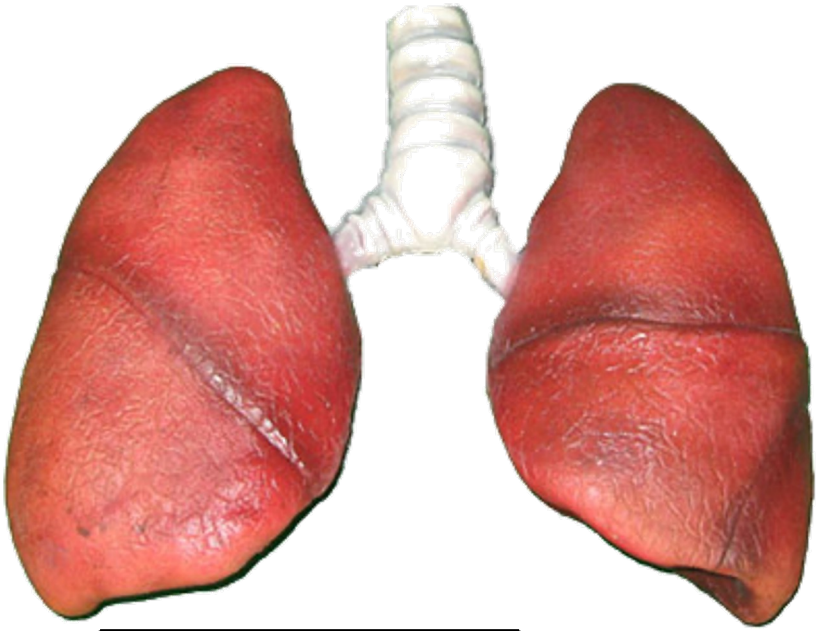
1. Have the participating individuals trace each other's body onto a piece of paper and draw their organs in the appropriate locations.

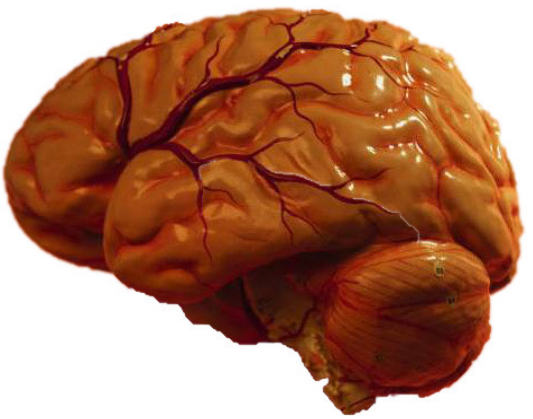
OR

2. Draw a tracing of a body on a board or easel of paper and have the children either draw in the organs or attach pre-made cut-outs of the heart and the liver.

- Use this as an opportunity to discuss the functions of the heart, liver, lungs, stomach, brain etc. and how they are or are not impacted by Hyperphenylalaninemia.
- It may also be useful for the older children to see a fresh liver. These can usually be obtained through a local slaughter house.

BODY PARTS





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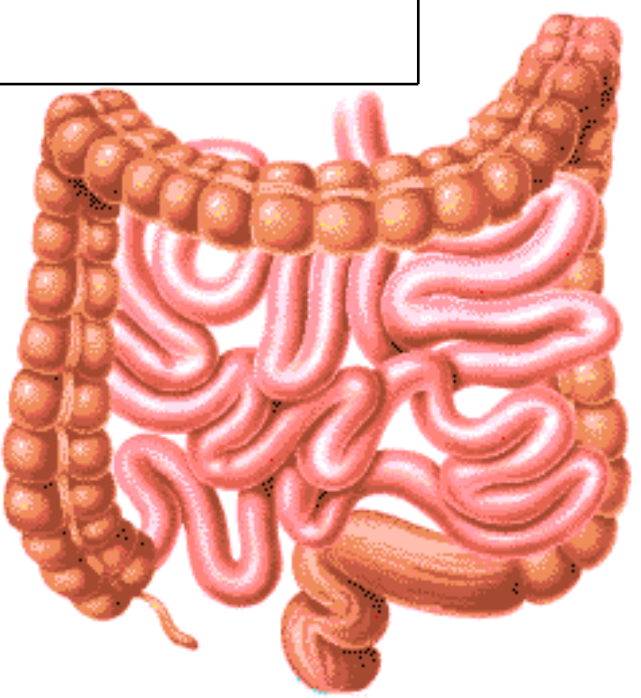

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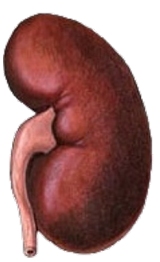

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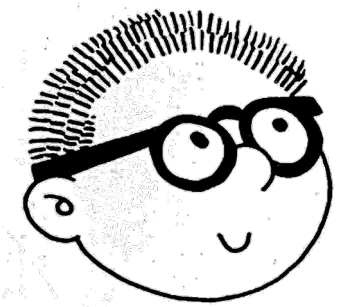
TEACHING AID

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.



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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

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.

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We Are All Alike, Yet Different

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Can You Find The Differences?

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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.

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

How It's Done

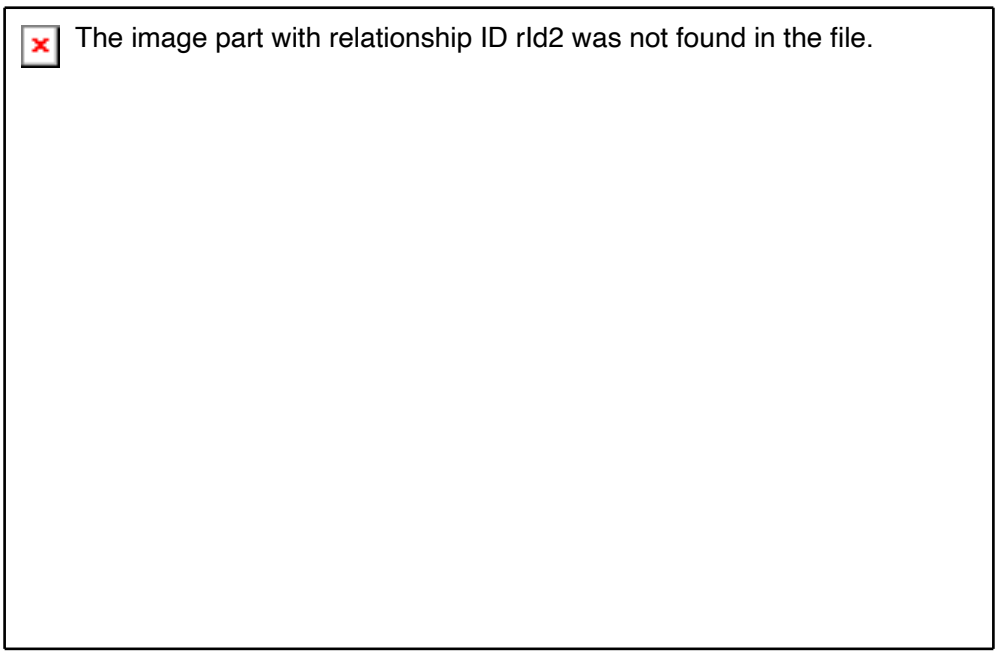
- Discuss the terms "alike" and "different" and use these terms in relation to physical characteristics and other distinguishing traits.
- Compare physical traits of individuals in the room, pointing out ways participants are alike and different.
- Talk about other personal characteristics such as food preferences and or sports skills.
- 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.
- Use the Venn Diagram teaching aid to continue discussing ways participants are alike and different.
- Talk about how some traits are genetic, some environmental, and some are a combination of the two.
- Ask participants to discuss the benefits of variation.

Handouts Needed
Venn Diagram

TEACHING AID

Venn Diagram

Objective: To teach the concept of characteristics and traits being unique or shared with others.



How It's Done

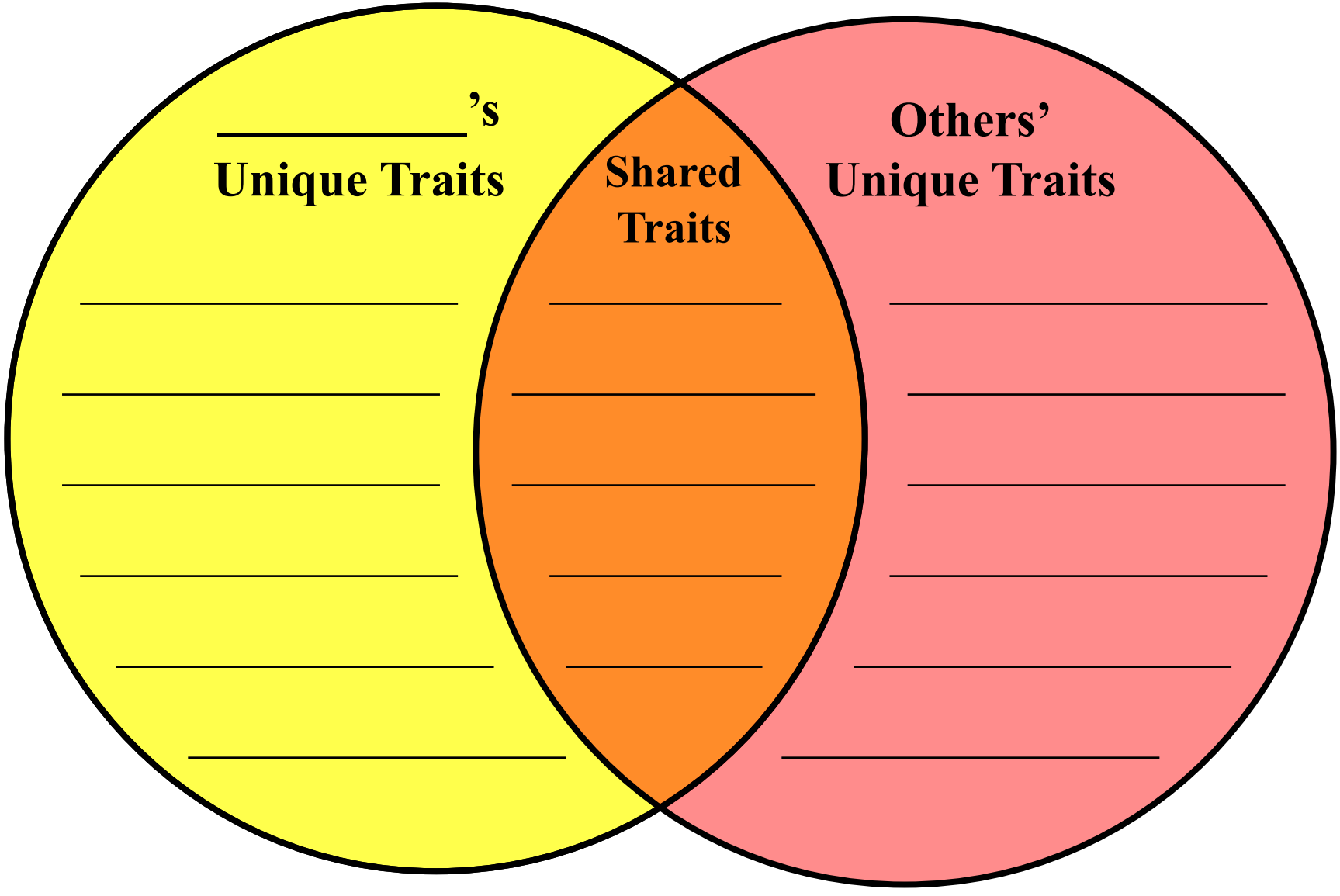
This activity works well with 2 or more children

Using the Venn Diagram handout:

- In the **yellow** circle, have each child write down some of their unique characteristics. Discuss how some of their traits are theirs only, no one else has that trait, that is what makes them unique.
- In the **red** circle, have each child write some of characteristics that are unique to others in the room.
- In the **orange** area in the middle, have each child write the characteristics they share with the others. One shared trait could be their metabolic disorder.
- Discuss the value of having unique traits and characteristics as well as having characteristics that are shared.

Handout Needed
Venn Diagram

Venn Diagram

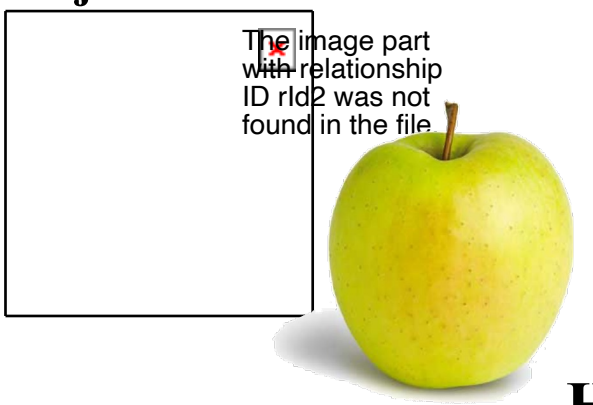


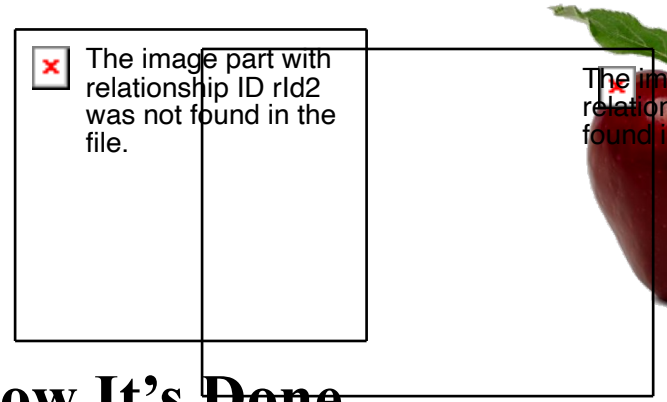
Adapted from "Alike or Different?" Scholastic Inc. 2006

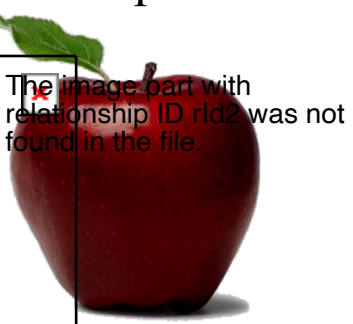
TEACHING AID

Comparing Apples To Apples

Objective: To introduce the concept of genes and genetic uniqueness.







How It's Done

- Bring an assortment of apples to clinic. Start by discussing how the apples are similar. Then note how the apples differ.
- Discuss how there may be other similarities or differences that are not readily visible, such as taste.
- If sufficient time is available, cut up the apples and have the participants taste them. Compare the different tastes.
- Follow with a discussion about members of the group. Talk about similarities and differences that are both visible and non-visible (i.e. PKU)
- Review with the group that our similarities and differences are a result of genes. Tailor the level of discussion to the participant's age and previous exposure to the topic. For example, for younger children one could discuss (or review) that genes function as recipes to make things. A red skinned apple will have a gene to make the color red. A green skinned apple will have a gene to make the color green. People have genes for skin, hair and eye color. They also have genes that impact how the body works. For example some people have genes for PKU and others don't. For older children, you may elect to discuss that we have genes (i.e. recipes) to make enzymes such as PAH. Discuss what happens if an individuals genes to make PAH have been altered such that no PAH is made.
- End by emphasizing that may of our similarities and differences are a result of genes. Stress that the differences we have make us unique and special!

Materials Needed
5 different apples

TEACHING AID

It's In The Code

Objective: To facilitate the introduction and discussion of genetic concepts.

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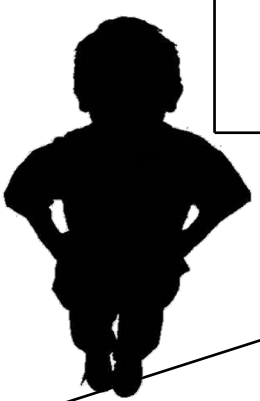

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Handouts Needed

It's In The Code
Jean's Genes
Recipes and Genes

How It's Done

The following is a suggested progression, incorporating the several handouts that can be used to talk about genetics at different levels. Remember to incorporate the children's metabolic disorder and positively reinforce uniqueness as often as possible

It's In The Code

- Compare the similarities and differences between people, animals, and plants: physical features, sources of energy or food, locomotion, and intelligence.
- Discuss these traits as being determined by genetic material called genes. Then ask the children what features might be comparable in people, animals, and plants: fur to hair, stem to legs, sounds to speech, walking to hopping.
- Introduce the concept that the parents of every species contribute the genetic make-up of their offspring, half from the mother and half from the father. Be sure to discuss that parents do not have control over which genes they pass on and which genes they do not. Tracing traits from one generation to the next is a fun activity. Use their own families or make-believe ones. *Be sensitive to variations in family structure.*
- Discuss that genes are contained within chromosomes in nearly every cell of living things. Show a Karyotope and discuss where the gene for PAH is located.

Jean's Genes

This is a teaching aid designed to introduce the concept of genes and inherited traits.

Recipes and Genes

This is a teaching aid designed to introduce the concept that a gene is like a recipe, a set of instructions. A change to those instructions can affect the end product.

It's In The Code

What is similar and what is different Between the objects below?

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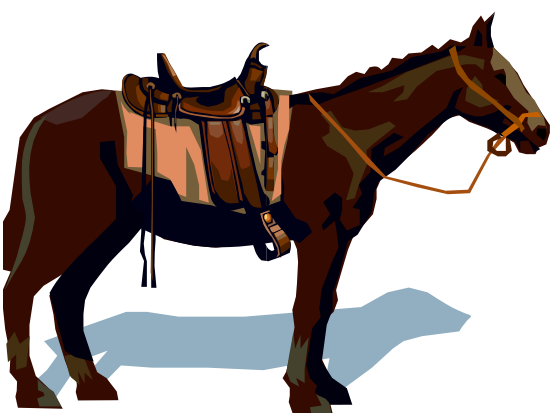
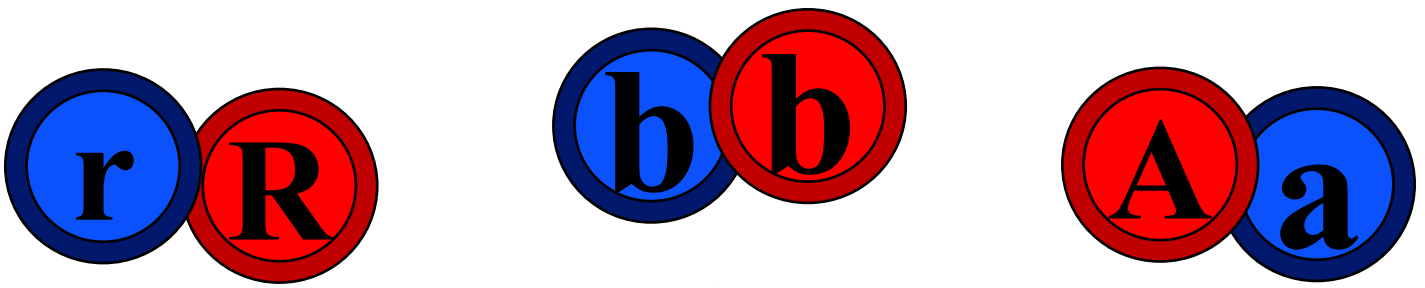


TEACHING AID

Jean's Genes

Objective: To reinforce the concept of genes and uniqueness.**



How It's Done

Preparation:

Label poker chips to represent the genes utilized in this activity (see genotype worksheet handout). Use one color of chips to represent the genes inherited from Dad and another color to represent the genes inherited from Mom. Create random sets (each set to contain one gene for face shape, one gene, for hair color, one gene for eye color, etc.) of maternal and paternal genes, placing them in envelopes labeled according.

Activity:

Discuss the concepts of genes and how they determine individual traits. Provide each participant with an envelope containing a set of paternal genes and an envelope containing a set of maternal gene. Note that these are Jean's genes; the genes she inherited from parents. Instruct the participants to open the envelopes and organize the genes into pairs. Start by finding the gene for face shape inherited from Dad. Line it up with the gene for face shape inherited from Mom. Have the participant record the inherited genotype on the genotype worksheet. Next, have the participant circle the resultant phenotype based on the genotype. After this is done for all the genes, have the participants draw a portrait of Jean based on her inherited traits. Introduce the terms "genotype" and "phenotype" referring back to the appropriate worksheets.

Materials Needed

- Red and blue poker chips
- Markers or colored pencils

Handouts:

Genotype Worksheet

Genotype Worksheet

FACE SHAPE: _____
- ❑ RR = Round
- ❑ Rr = round
- ❑ rr = oblong

HAIR COLOR: _____
- ❑ DD = Black
- ❑ Dd = Brown
- ❑ dd = Blond

EYE COLOR: _____
- ❑ BB = brown eyes
- ❑ Bb = brown eyes
- ❑ bb = blue eyes

NOSE SIZE: _____
- ❑ NN = large nose
- ❑ Nn = medium nose
- ❑ nn = small nose

EAR LOBES: _____
- ❑ EE = hang free, not attached
- ❑ Ee = hang free, not attached
- ❑ ee = attached, do not hang free

LIPS: _____
- ❑ LL = full lips
- ❑ Ll = full lips
- ❑ ll = thin lips

FRECKLES: _____
- ❑ FF = no freckles
- ❑ Ff = no freckles
- ❑ ff = freckles

PKU: _____
- ❑ AA = does not have PKU
- ❑ Aa = does not have PKU
- ❑ aa = does have PKU

HFI: _____
- ❑ HH = does not have PKU
- ❑ Hh = does not have PKU
- ❑ hh = does have PKU

Jean's Portrait

Using the results from the worksheet, draw what Jean will look like.

TEACHING AID

Recipes & Genes

Objective: To introduce the concept that a gene is like a recipe, a set of instructions. A change to those instructions can affect the end product.

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Materials Needed

- Baking ingredients

Handout:

Recipes & Genes

- Holiday Cookies
- Low Protein Waffles

How It's Done

- Discuss that genes carry instruction to make PAH and other substances in our body.
- Relate this to a recipe that instructs on how to make waffles or cookies.
- Have the participant's speculate what might happen to the waffle or cookie if the recipe was changed. For example, speculate on what would happen if the changed recipe called for one teaspoon of salt rather than two teaspoons of salt. What would happen if it called for two cup of salt rather ant two teaspoons of salt? Discussed how some changes can results in a waffle or cookie that may not be "perfect" but is still edible (hence functional). Other changes will result in a waffle or cookie that you cannot eat and would not be functional.
- Relate this to a changes in the gene for PAH and resultant changes in PAH activity or functionality.
- Make products using a recipe provide in the handout. Change the amount of a particular ingredient. Discuss the outcome and how the change in the recipe impacted the end product.

HOLIDAY COOKIES

RECIPE FROM WWW.MYSPECIALDIET.COM

Yields: 20 Servings

Per Cookie: Phe: 5 mg **Protein:** 0.12 g

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Ingredients:

4 oz	Butter, room temperature
2 oz	Sugar
6 ½ oz	Loprofin Low Protein Baking Mix*
½ oz	Butterscotch Instant Pudding (powder)
¾ tsp	Milupa lp Drink

Directions:

1. Place the butter and sugar in a mixing bowl and beat well, until light in color and texture.
2. Stir in the Loprofin and pudding powder. Using one hand, stir and squeeze the mixture until it comes together, adding sufficient Milupa lp Drink to give manageable dough. Transfer to a surface lightly dusted with Loprofin Baking Mix and knead the dough for about until smooth.
3. Roll out the dough to about ¼ inch thickness, on a surface lightly dusted with Loprofin.
4. Cut out shapes. Re-roll the trimmings and continue cutting out the shapes, until all dough is used.
5. Transfer the cookies to lightly greased non-stick baking trays and bake in pre-heated oven for 15 minutes, until a pale golden color.
6. Allow the cookies to cool slightly on the baking trays, carefully remove from the trays and complete cooling on a wire rack.

LOW PROTEIN WAFFLES

RECIPE FROM PKU COOKERY, VIRGINIA SHCUETT

Yield: 5 waffles (4 squares per waffle)

Phe: 14 mg Phe (per ¼ waffle – 1 square)

Ingredients:

330 g (3cups)	Wel-Plan Baking Mix*	¼ tsp	Vanilla
110 g (1 cup)	Wheat Starch	29 g	Egg White (1 Tbls + 1 tsp)
2/3 cup	Vegetable Oil	4 tsp	Baking Powder
1 1/3 cup	Rich's Coffee Rich	½ tsp	Salt
1 ¼ cup	Water	¼ cup	Sugar

Directions:

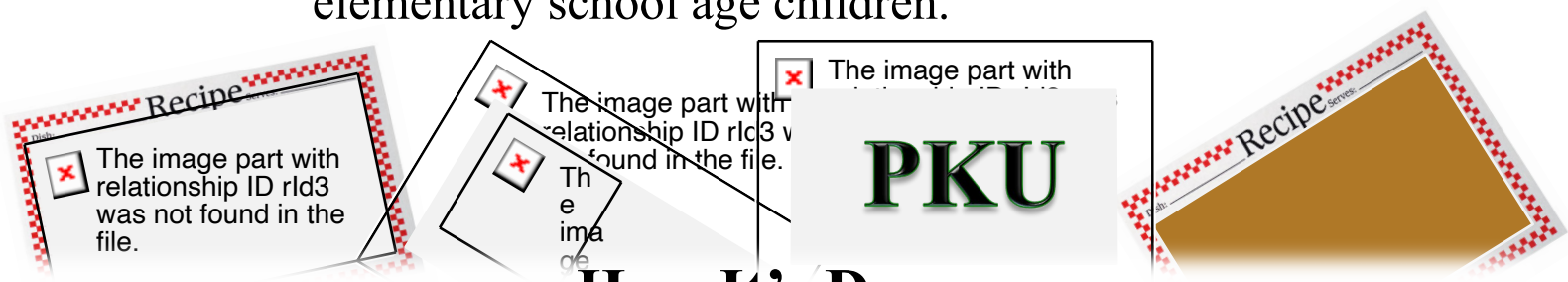
1. In a large mixing bowl, mix Wel-plan baking mix, wheat starch, baking powder, salt, and sugar.
2. Combine oil, Coffee Rich, water and vanilla in a large liquid measuring cup or small bowl.
3. Add to dry ingredients, mixing until smooth.
4. Put egg white in a small bowl and beat with an electric mixer until stiff but not dry. The total volume will be about ¾ cup.
5. Gently mix beaten egg white into waffle batter. Thin batter with a little water if batter is too thick to spread on waffle iron.
6. Brush vegetable oil lightly on top and bottom of a hot waffle iron to prevent any sticking. Use a 1 cup measuring cup to scoop the batter, scraping out quickly onto hot iron (1 cup of batter will make a nice, full 4-square waffle).
7. Bake 3 to 5 minutes. Lid should open easily when done. Open iron and remove waffle carefully.
8. Serve immediately, or cool completely on a wire rack and freeze.

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

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

- 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!!

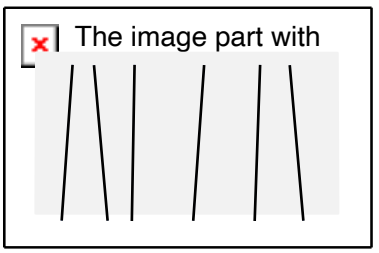
Handout Needed
My Recipe Gene Book

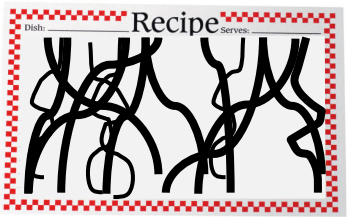
My Recipe Gene Book

Name _____

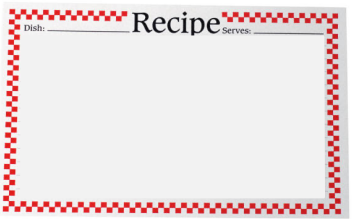
Each box represents a gene or recipe to make important parts of your body. Circle the genes (or recipes) that you have. For example, if you have curly hair, circle the curly hair gene. If you have straight hair. circle the straight hair gene. *Note for instructor*: The hair color genes need to be colored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

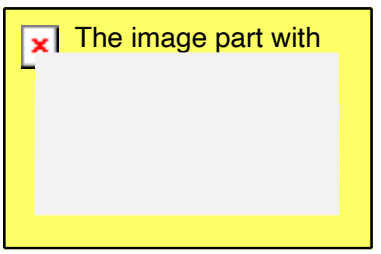
Gene For Hair Texture



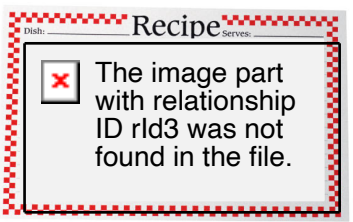


Gene For Hair Color

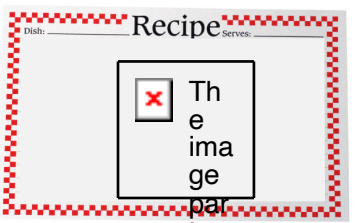




Gene For Eyes



Gene For Nose



Gene For PKU

Gene For HFI

TEACHING AID

I'm Thumb-body Special

Objective: To introduce the concept of genetic differences between people.

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How It's Done

Fingerprints are a fun way to demonstrate the subtle differences between people

- Have the participants first examine their thumbs and fingers closely and notices the fine lines that make up the thumb/finger print.
- Explain that, like snowflakes, no two fingerprints are alike. Discuss that every person in the world has a unique thumb print.
- Help the participants make their own thumb print on the "I'm Thumb0body Special" handout.
- Use as magnifying glass to look closely at the pattern created.
- Compare prints and note the differences. Discuss that these differences are, in general, caused by our genes. Discuss that our genes make each individual special.
- Have the participants to turn their thumb print into a "thumb-body". Ask them to think of the print as a face. Have them use markers to add hair, eyes, nose, etc. Demonstrate this process using your thumb print.
- Encourage them to make additional thumb-buddies. Dependent upon how much time is available, recommend they draw different types of faces (happy, mad, etc.) to reflect how they feel right now, how they feel when they have to have their blood draw, how they feel when they eating lunch with their friends, etc.. Explore, discuss, and validate these feelings.
- End by reviewing that each participant, is unique. Have them list several things that are special about themselves. Ask if they would like to share with other, items that they have listed. Discuss if the participants listed their metabolic disorder (or not), and why.

Materials Needed

- Ink Pad
- Markers

Handout:

I'm Thumb-body Special

I’m Thumb-body Special

Name: _____

Special Things About Me:

_____

_____

_____

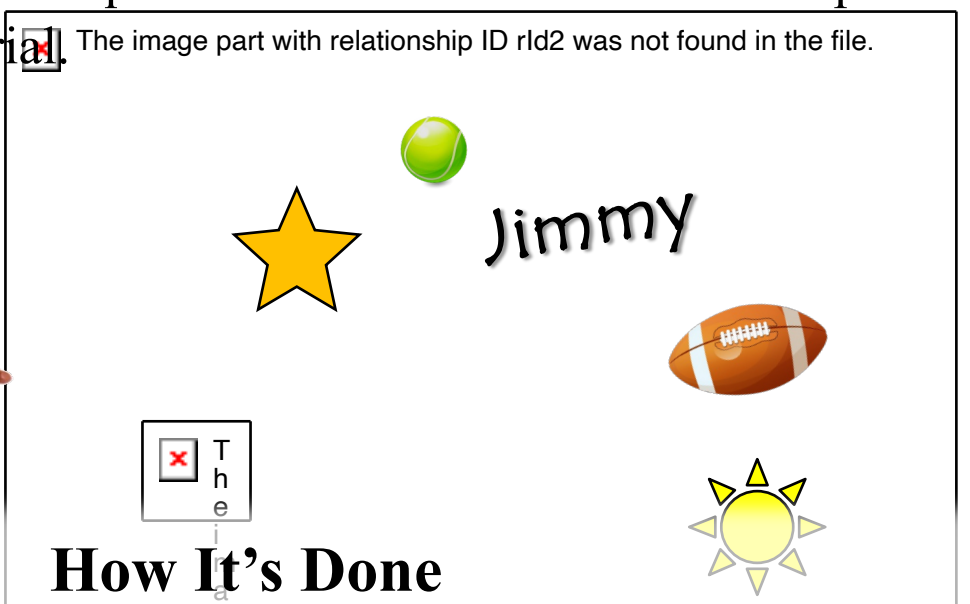
_____

_____

TEACHING AID

Personalized CD

Objective: To emphasize uniqueness and to introduce the concept of genetic material.



How It's Done

- Start by discussing computers and the concept of computer programs. Ask participants make a list of tasks that computers are able to do (i.e. mathematical calculation, check for spelling error, show videos, etc.). Discuss that every computer must have a program, or set instruction, that tells the computer how to do each task. Discuss differences in computer capabilities and speculate on the differences in their computer programs.
- Relate a computer's set of "programs" to an individual's set of "genes".
- Give each individual a blank CD that is to be personalized with programs (i.e. genes) that make them special and unique. Start by having them write their name on the CD. To further personalize it, have them write, draw pictures, or add stickers that represent how they look and or things they like to do. If they have added PKU to their CD, explore why the chose to do so. Do the same if they have not added PKU. Discuss that either way is okay; it is an individual feeling (than may vary from day to day and situation to situation).
- Have each participant share with the group their personalized CD. Discuss similarities and differences between the CDs.

Materials Needed

- Blank CD or Personalized CD Handout
- Markers or color pencils
- Stickers

PERSONALIZED CD

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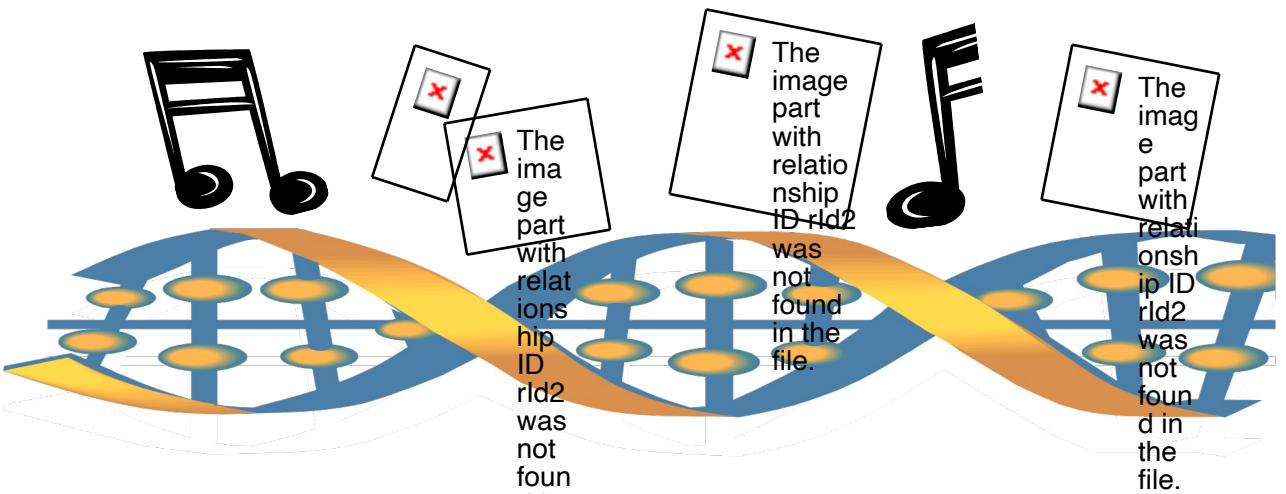
TEACHING AID

The Gene Song

Objective: To use music as a medium for learning about genetic inheritance.



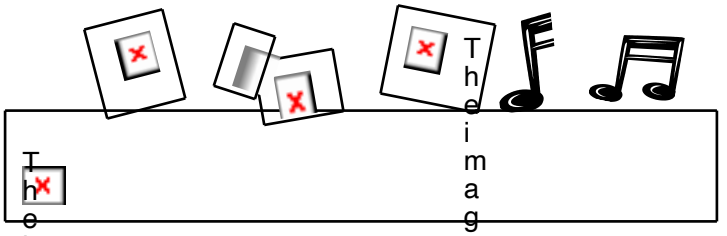
How It's Done

Use the handout "The Gene Song"

- Sing the words provided in the handout to the tune of: "*Head, Shoulders, Knees and Toes*".
- Ask each participant to sing a verse of the song, maybe even add their own lyrics.
- Add hand gestures and dance moves.
- Practice your creation. Stage a production for families or staff, or videotape the performance.
- Have fun! Be crazy!

Handout Needed
The Gene Song

The Gene Song



To the tune of "Head, Shoulders, Knees and Toes"

Genes come from mom and dad (mom and dad!)
Genes come from mom and dad (mom and dad!)
They make us short or make us tall
Genes make us who we are (who we are!)

Genes come from mom and dad (mom and dad!)
Genes come from mom and dad (mom and dad!)
They make our nose big or make our nose small
Genes make us who we are (who we are!)

Genes come from mom and dad (mom and dad!)
Genes come from mom and dad (mom and dad!)
They change things that cannot be seen
Genes make us who we are (who we are!)

Genes come from mom and dad (mom and dad!)
Genes come from mom and dad (mom and dad!)
Mine can't use phenylalanine
Genes give me PKU (PKU!)

TEACHING AID

A Kid's Life

Objective: To dialogue on how various parts of a child's lifestyle make that a child whole.

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How It's Done

- Utilize the handout as a means to help the child identify important aspects of their daily life.
- Create an opportunity for discussion about how each part of their lifestyle contributes to who they are.
- Emphasize the role of diet as a key factor in being able to achieve wholeness.

Handout Needed
A Kid's Life

A Kid's Life

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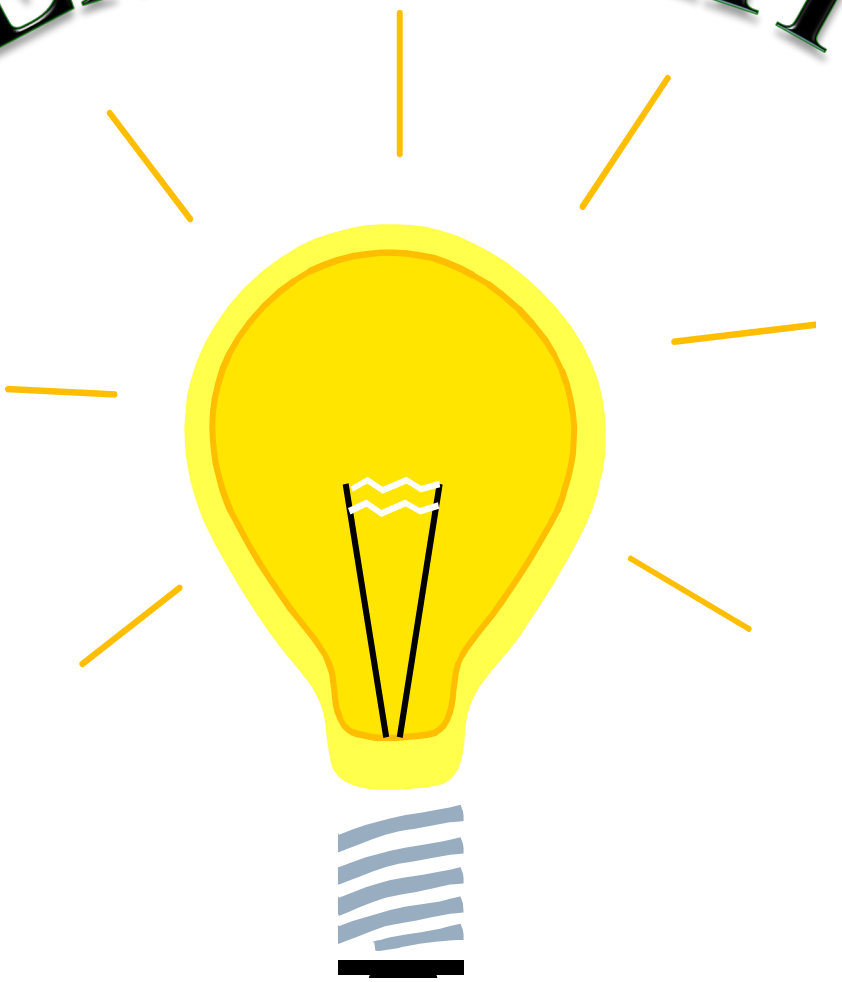
EAT RIGHT



STAY BRIGHT™

Guide For Hyperphenylalaninemia

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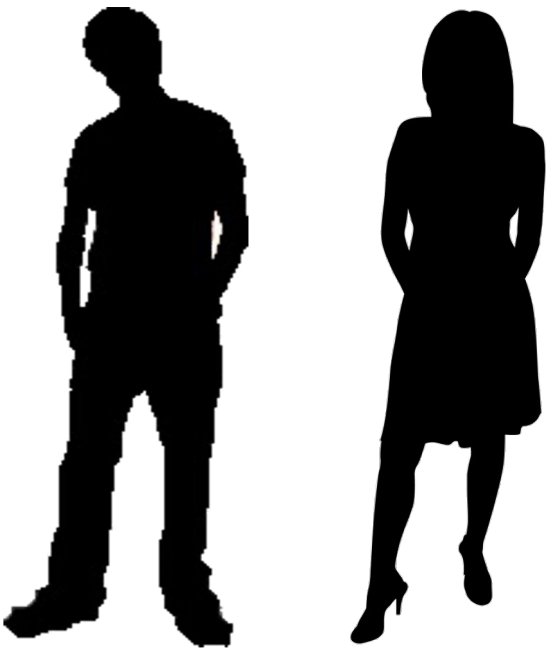
Chapter Three



The Adolescent Years

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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ADOLESCENCE

The adolescent years denote change. Cognitive and emotional changes during adolescence are in their own way as dramatic as physical changes during this period. Throughout the various developmental cycles, we encourage reinforcement of age appropriate knowledge and independence. These practices are critical during adolescence. Adolescence spans from 11-21 years of age. Limits are tested during this period. Important issues vary from throughout adolescence; therefore, we have divided this chapter into three main areas of focus:

- **Early Adolescence (11-14 years of age)**
- **Middle Adolescence (15-17 years of age)**
- **Late Adolescence (18-21 years of age)**

It is important to note that each individual has a unique personality and will transition into the three stages of adolescence at their own pace. Some behaviors are unique to a certain adolescent stage while others will overlap. We suggest that you intertwine the checklists presented in this section, as they seem appropriate on an individual basis.

Early and middle adolescents live in the present. Therefore, their perception of long term consequences resulting from present actions is often minimized. Having a strong knowledge base of their disease while understanding the consequences of their actions, therefore, is a determining factor for successful management of their diet (from superscript #1). In early adolescence, exploration and testing limits is a familiar theme. Beginning in middle adolescence, individuals develop a greater capacity for setting goals.

During late adolescence, individuals focus on achieving autonomy.

Normal development includes fluctuation between unrealistically high expectations and self-esteem, and poor motivation and self-concept. As perceptions fluctuate daily, this creates a seesaw effect. For adolescents with hyperpheylalaninemia, this effect is magnified when phenylalanine levels are consistently high.

In this chapter, we have included:

- **Checklists** that can be used to guide the professional in the care of adolescents at various stages of development.
- **Teaching Aids** that are designed to assist adolescents, at all three stages, in achieving self-assurance, responsibility, and the knowledge and belief that they can succeed.

Above all, we hope that the information and activities in this section will help

CHECKLIST: Early Adolescence & Parents

Planning For The Future[1]

- ❑ Short term goals
- ❑ Long term goals

Genetics

- ❑ Revisit autosomal recessive inheritance[2]
- ❑ Carrier risks and testing for extended family

Biochemistry

- ❑ Diet for life
- ❑ Adverse effects of elevated levels
  - MRI findings
  - Neurological findings
  - Personality changes
  - School and social performance
- ❑ Maternal PKU[3]

Phe Levels, Growth Charts, Interim History

- ❑ Interim Phe levels
- ❑ Interim tyrosine levels
- ❑ Intercurrent illness
- ❑ Puberty/menstrual cycle
- ❑ Heights and weights

Daily Living Routine

- ❑ Weighing, measuring, and preparing formula
- ❑ Cooking/recipes
- ❑ Diet records
- ❑ Blood draws[4] (pg 58 – 59)
  - Time of draw
- ❑ Setting[5]
  - School
  - Home
  - Work[6]

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

CHECKLIST: Early Adolescence & Parents

Psychosocial Issues

- ❑ Increasing autonomy
- ❑ Peers[7]
- ❑ School[8]
- ❑ Sports
- ❑ Family[9]
- ❑ Parental involvement
  - Manipulation/power struggles/conflict resolution[10] (pg 33 – 36)
  - Over-commitment/martyrdom[11]
  - Limit setting[12]
- ❑ Family communication
- ❑ Attitudes[13] (pg 22 – 23)
- ❑ Self-esteem (pg 30)
- ❑ Body image [14]
- ❑ Isolation[15]
- ❑ Finances[16]
- ❑ Impact of diet on family lifestyle

Nutrition Intervention

- ❑ Formula consumption
  - B-12 deficiency
  - Folate deficiency
- ❑ Use of the following items[17]
  - Low protein foods
  - Low protein recipe books (i.e. V. Schuett)
  - Gram scale (pg 56 – 57)
  - Bread machine
  - Low protein food lists
- ❑ Diet records
- ❑ Recipe experimentation
- ❑ Kuvan (pg 21)

CHECKLIST: Middle Adolescence

Planning for the Future[1]

- ❑ Short term goals
- ❑ Long term goals
- ❑ Achieving goals

Self Management Strategies

- ❑ Goal setting
- ❑ Monitoring
- ❑ Self administering consequences
- ❑ Management contracts[18] (pg 62 – 66)

Genetics

- ❑ Recurrence risk
- ❑ Genotype/phenotype correlation/lack of correlation
- ❑ Research advancements/developments
  - Gene therapy
  - Genetic engineering
  - Enzyme replacement therapy
  - PAL (phenylalanine ammonia lyase)

Biochemistry

- ❑ Practical application to diet management
- ❑ Current thoughts regarding pathophysiology of PKU
- ❑ Mean phenylalanine and tyrosine levels
- ❑ Maternal PKU[3]

Daily Living Routine

- ❑ Cooking/recipes
- ❑ Availability of low protein foods
- ❑ Diet records
- ❑ Blood draws
- ❑ Setting[5]
  - School
  - Home
  - Work[6]

CHECKLIST: Middle Adolescence

Psychosocial Issues

- ❑ Peers[7]
- ❑ School[8]
- ❑ Sports
- ❑ Family
- ❑ Parental involvement
- ❑ Manipulation/power struggles/conflict resolution[10] (pg 33 – 36)
- ❑ Over commitment and martyrdom[11]
- ❑ Limit setting
- ❑ Attitudes[13] (pg 22 – 23)
- ❑ Self-esteem (pg 26)
- ❑ Problem solving techniques[19] (pg 33 – 36)
- ❑ Body [14]
- ❑ Isolation [15]
- ❑ Finances[16]
- ❑ Impact of diet on family lifestyle[9]
- ❑ Driving/family rules
- ❑ Dating and the social scene/family rules
- ❑ Risk taking (pg 60 – 61)
- ❑ Routine clinic visits[20]

Nutrition Intervention

- ❑ Formula consumption
  - B-12 deficiency
  - Folate deficiency
- ❑ Use of the following items[17]
  - Low protein foods
  - Low protein recipe books (i.e. V. Schuett)
  - Gram scale (pg 56 – 57)
  - Bread machine
  - Low protein food lists
- ❑ Diet records
- ❑ Recipe experimentation
- ❑ Kuvan (pg 21)

CHECKLIST: Late Adolescence

Planning for the Future

- ❑ Long term goals
- ❑ Medical insurance
- ❑ Formula coverage

Self Management Strategies

- ❑ Goal setting
- ❑ Monitoring
- ❑ Self administering consequences
- ❑ Management contracts[18] (pg 62 – 66)

Genetics

- ❑ Family planning
- ❑ Recurrence risk
- ❑ Spousal carrier testing

Biochemistry

- ❑ Practical application to diet management
- ❑ Current thoughts regarding pathophysiology of PKU
- ❑ Mean phenylalanine and tyrosine levels
- ❑ Maternal PKU[3]

Daily Living Routine

- ❑ Cooking/recipes
- ❑ Availability of low protein foods
- ❑ Diet records
- ❑ Blood draws (pg 58 – 59)
- ❑ Setting[22]
  - School
  - Home
  - Work

CHECKLIST: Late Adolescence

Psychosocial Issues

- ❑ Problem solving skills[19]
- ❑ Family[9]
- ❑ Finances[16]
- ❑ School/vocation
- ❑ Autonomy

Nutrition Intervention

- ❑ Formula consumption
  - B-12 deficiency
  - Folate deficiency
- ❑ Use of the following items[17]
  - Low protein foods
  - Low protein recipe books (i.e. V. Schuett)
  - Gram scale (pg 56 – 57)
  - Bread machine
  - Low protein food lists
- ❑ Diet records
- ❑ Recipe experimentation
- ❑ Kuvan (pg 21)

Experience & Thoughts

1. It has been our experience that having both the adolescent and parents verbalize and discuss goals on an ongoing basis leads to realistic expectations and facilitates future planning.

2. Revisiting autosomal recessive inheritance may be critical in a family where there are unaffected siblings. Understanding the randomness of the disease occurrence may help diffuse the anger and/or guilt of either having the disease, passing on the disease or being unaffected by the disease.

3. We discuss Maternal PKU so often that one would think that it sinks in. That has not always been our experience □. See our next chapter.

4. We encourage and instruct our teenagers to draw their own blood. This encourages independence, autonomy, and a sense of control. (see pg 58 – 59)

5. Adolescents typically have busy schedules. Distraction, temptation, peer pressure, supervision (too much or too little), availability and access to formula and low protein foods all play a role in a child's life and his/her decision making. We have found that parents and staff need to assist adolescents in learning to plan for compliance around a busy school or work schedule.

6. Work can be used as a reason for noncompliance. Inconvenience/inaccessibility to formula/food should be approached as a challenge to develop a schedule that fits within the boundaries of work.

7. We would anticipate at this age that the adolescent has a solid support system and comfort level with the diet. We have found that those adolescents who do not are prone to greater non-compliance at school. Lunchtime can be a critical turning point in the day. The social gathering can either be manageable or be the major cause for compromising diet. The support of peers and teachers can be critical in promoting a feeling of acceptance and safety.

8. Consider whether it is appropriate to inform teachers and coaches about special dietary needs or arrangements. Most middle schools now operate on a team system, which provides more continuity with teachers and more attention to social development. Parent communication with teachers is important. High school students are more independent; if necessary, a school counselor may be informed in lieu of independent teachers.

9. It has been our experience that an alliance within the family, with regard to successful diet management, will help to promote a life long bond and support system.

10. Conflict that is problematic in early adolescence can be magnified in the later teenage years. Questions such as "Are rules clearly stated?" and "Are consequences logical?" should be asked. Addressing issues in the early teenage years will likely pay off down the road. Keep lines of communication open. Conflict cannot be resolved without communication. Commit to communicating constructively.

11. In adolescence, one of our goals is to promote self-esteem and autonomy. Over-involvement can be suffocating and can limit the self-actualization of the adolescent.

12. We encourage parents to be "friendly foes*". Adolescents need parental support, guidance and limit-setting. Parents may feel that their child is ready for more responsibility than they in fact can handle. Inconsistent limits may be worse than no limits at all. (*see Resources/References, REBELS WITH A CAUSE)

Experience & Thoughts

13. Encourage a positive attitude.

14. Adolescents are especially sensitive to unhealthy messages concerning their weight. Adolescents who feel they are “different” are at even greater risk for trying to blend in with peers despite negative consequences.

15. If an adolescent isolates him or herself from peers, clinic staff, and/or family there is reason for concern.

16. In many states, insurance coverage of formula and/or laboratory testing is a concern. It is important to work with the families to acquire coverage.

17. The adolescent should be very familiar with all the components that facilitate compliance.

18. Written contracts may provide adolescents with a sense of accountability and ownership.

19. Adolescents face many challenges that require effective problem solving skills.

20. With patients who are noncompliant we have seen the tendency to cancel clinic appointments. We appeal to the parents as the responsible adults to ensure that clinic appointments are kept. During these visits, we have found that a non-judgmental approach is much more effective. We educate, support, and try and keep communication open between all three parties: parent(s), patient, and clinic staff. A triangle that cannot be complete unless all sides are connected.

21. We have found it necessary that the late adolescent and the health professional develop an adult to adult relationship. This relationship can be influential and supportive in helping to transition the adolescent into adulthood.

22. The transitions to independent living are usually seen during this time.

PRINCIPLES OF DIET PRESCRIPTION

Throughout a Lifecycle

Birth to 6 Months | 6 to 12 Months | 1 to 2 Years | 2 to 7 Years | 8 Years to Adulthood

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

PRINCIPLES OF DIET PRESCRIPTION

Throughout a Lifecycle

Birth to 6 Months | 6 to 12 Months | 1 to 2 Years | 2 to 7 Years | 8 Years to Adulthood | Motherhood

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

PRINCIPLES OF DIET PRESCRIPTION

8 Years Old Though Adulthood

Medical Food

Circle One

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

+

=

Phe, Amino Acids, Vitamins, Minerals & Calories

If an adult only eats 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.

MEDICAL FOOD OPTIONS

8 Years Old Through Early Adolescence

Nutricia

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

Applied Nutrition

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

Abbott Nutrition

- Phenex-2

Cambrooke Foods

- Camino Pro
- Camino Sorbet Stix

Mead Johnson

- Phenyl Free 2
- Phenyl Free 2 HP

Vitaflo

- PKU Coolers
- PKU Express
- PKU Gel

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

DAILY DIET PRESCRIPTION

8 Years Old Through Early Adolescence

Phe 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: 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.

DAILY DIET PRESCRIPTION

Middle Adolescence

Phe 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.

DAILY DIET PRESCRIPTION

Late Adolescence Through Adulthood

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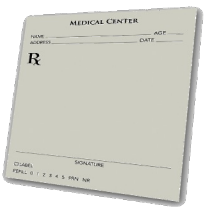


Phe Complete Nutrition

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/Height:**

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

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

Additional Notes:

_____

_____

_____

Large Neutral Amino Acids (LNAA)

An alternative treatment solution for patients 12 years and older who are considering an option to medical food in the nutritional management of PKU.

Why are "Alternative Therapies" like Large Neutral Amino Acids (LNAA) being used?

- Many adolescent and adult patients are looking for alternative therapies to the standard low phenylalanine diet.
- Diet for Life (standard PHE Restriction) is difficult to maintain
- Patients with PKU whom have been treated since NBS are either reaching or have reached adulthood
    - In the USA there are roughly 9000 patients with PKU since NBS initiated (est. 210 live births per year X 43.5 years [1965])\
    - Approx. 5500 more patients with PKU who predated NBS are in dependent living homes/institutions (avg live span 75 years-31.5 years pre-1965 X est. 185 live births per year).

Lanaflex™ is the only...

- ☑ Nutritionally complete alternative treatment option that contains vitamins and minerals.
- ☑ Low volume treatment that reconstitutes to a 2 fl oz dose taken 3 times day, with protein containing meals.
- ☑ Normal alternative treatment for patients struggling with social acceptance.

Other LNAA Products

Solace Nutrition:

- PreKUnil
- NeoPhe

Applied Nutrition:

- PheBloc

Where LNAAs Should Be Used With Caution/Close Monitoring

- Patients on psychotropic medications (SSRIs)
- Patients who may be using amino acid supplements for reasons other than treatment (e.g. weight lifters)
- Women of childbearing age

Where LNAAs Is Not Indicated For Use

- Young children who are compliant with diet
- Any patient who is in good control and compliant with classic diet
- Pregnant women (no data)
- Lactating women (no data)

Adapted from *"Lanaflex - Guidelines for Use"* Rockville, MD: Nutricia North America.

Lanaflex™

Large Neutral Amino Acids (LNAA)

The report of the Medical Research Council Working Party on PKU recommended the continuation of dietary management beyond childhood, preferably for life. However, compliance with PKU medical foods is not optimal and Prince et al have reported that less medical food is taken than the amount actually prescribed.

There is a tendency for individuals with PKU to relax dietary control as they increase in age, forming a so-called 'off-diet' population in which Phe concentrations are high and nutritional status may be compromised. Individuals often self restrict high biological protein, when they come off diet, and this can lead to a diet low in essential amino acids.

Nutricia North America supports "diet for life" for all patients with PKU. Research has shown that adherence to a well-managed diet plan has the best possible clinical outcomes for patients with PKU.

Lanaflex has been developed to address the population of individuals with PKU who are no longer following a strict Phe-restricted diet. Lanaflex offers the potential to help control brain and plasma Phe concentrations and support normal nutrient status.

Indications for Use

Lanaflex is indicated for individuals over 12 years of age with proven PKU, who no longer adhere to a strict Phe-restricted diet. These individuals are often referred to by clinicians as being "off diet". Lanaflex is not recommended as an alternative to the Phe-restricted diet but as an option if individuals with PKU choose to come off diet.

Amino Acid Profile

All of the essential amino acids (except Phe) are present in Lanaflex. The amino acid profile used in Lanaflex is a "balanced profile" of amino acids, i.e. there are no excessively high or low concentrations of the essential amino acids or lysine.

Micronutrient Profile

Vitamins, trace elements and calcium, magnesium and phosphorus have been included in the product to help ensure normal nutrient status. The level of the micronutrients in Lanaflex is set at a level that meets at least 80% of DRI-for-age micronutrient requirements.

Dosage

Lanaflex is available in 15.8 g stick packs, which contains 5.2 g protein equivalents [PE].

Adapted from *"Lanaflex - Guidelines for Use"* Rockville, MD: Nutricia North America.

More information available at www.Nutricia-NA.com

GLYCOMACROPEPTIDE (GMP)

After years of research, development, and collaboration with the University of Wisconsin Madison research team, Glycomacropeptide (GMP) is the first ever intact protein to be used to treat PKU!

For the first time, PKU patients may benefit from a slower, more gradual, and sustained elevation in plasma amino acid concentrations that only an intact protein offers.

GMP can help prevent the extreme swings in plasma amino acid concentrations typical in amino acid based diets and reduce the possibility of catabolism. In addition, the GMP is naturally high in the large neutral amino acids threonine, isoleucine, and valine. LNAAs compete with phenylalanine at the blood-brain barrier, resulting in lower brain phe levels. (J Clin Invest. 1999; 103(8): 1169-1178).

What is Glycomacropeptide (GMP)

Glycomacropeptide (GMP) is a naturally occurring dairy protein that contains minimal phenylalanine (Phe) and high levels of large neutral amino acids (important for keeping Phe from crossing the g=blood-brain barrier), making it perfect for use as a PKU formula replacement or as a food ingredient for people with PKU. Its high concentration of sialic acid, a carbohydrate structure in GMP, my aid in normal brain development and have anti-bacterial properties as well.

GMP is one of several proteins that make up whey, accounting roughly for 15 – 20% of whey's protein. Whey protein makes up 20% of milk protein and is a byproduct of cheese making. The Health benefits and functional properties of GMP have increased it use as an ingredient for nutritional formulations and health foods over the last 15 – 20 years. Because why proteins are part of milk and have a history of use as food ingredients such as in infant formulas, they are generally recognized as safe.

BENEFITS:

- GMP is an intact protein that is better utilized by the body
- Helps to better maintain blood Phe levels between meals
- Low volume - mix with 4 fluid ounces of water for 75 grams of protein equivalent
- 32 mg of DHA Omega-3 Fatty Acids per serving (DHA Omega-3 for a healthy brain and heart)
- Probiotic cultures to support digestive health
- Fortified with vitamins & minerals
- Great, neutral, non-medicinal taste!
- Combines well with you favorite foods

More information available at www.cambrookefoods.com

Adapted from *" CamBrooke Foods. Introducing Camino Pro® BetterMilk™ with Glycomacropeptide (GMP). Ayer, MA: CamBrooke Foods,*

What is the Function of BH4 in the Body?

Normally, BH4 helps the enzyme known as PAH (short for phenylalanine hydroxylase) work. The PAH enzyme breaks down the amino acid Phe, found in many foods, into other needed chemicals in the body. In PKU, the PAH enzyme isn't working, which allows too much Phe to build up in the blood. This can eventually affect the brain if left untreated.

What is KUVAN?

KUVAN (Sapropterin Dihydrochloride Tablets) is a medication for PKU that functions like BH4, a substance that naturally occurs in the body.

How Does KUVAN Work?

In many patients, KUVAN increases the activity of the PAH enzyme that isn't working properly. In other words, the PAH enzyme that isn't working "wakes up" and starts to process the Phe in some patients with PKU. This helps to lower the amount of Phe in the blood in these patients.

Should I Consider KUVAN?

The Phe-restricted diet is challenging, and so is consistently keeping your Phe level low. A low-Phe diet still allows a certain amount of Phe to pass from the food you eat into your bloodstream. KUVAN, together with a Phe-restricted diet, helps you lower your blood Phe level and keep it low day after day. KUVAN addresses the problem from a different angle than diet does. KUVAN works by stimulating the PAH enzyme to break down the Phe in your body, thereby lowering your blood Phe levels.

Does KUVAN Work for Everyone?

KUVAN does not work for everyone. However, studies have found that some people with mild, moderate, and severe PKU have responded to treatment with KUVAN. It is not possible to know whether KUVAN will work until you start taking it. Your doctor will determine whether or not KUVAN is working by checking to see if your blood Phe drops while the Phe-restricted diet is held constant.

If you do not initially respond to KUVAN, your doctor may consider another trial of KUVAN if there is reason to believe that diet, change in use of medical food, or other factors affecting blood Phe level (such as fever or illness) may have affected your results. Unfortunately, KUVAN may not work for everyone.

Will KUVAN Work Long Term?

Yes. In clinical trials, the benefit of KUVAN continued throughout the length of the study. KUVAN has been studied for periods from 1 to 30 weeks.

What are the Possible Side Effects of KUVAN?

In studies, side effects in patients taking KUVAN generally occurred at a similar rate as they did in patients who received placebo (a pill without any medicine in it).
The most common side effects reported when taking KUVAN included:

- Headache
- Diarrhea
- Abdominal pain
- Upper respiratory tract infection (like a cold)
- Throat pain
- Vomiting
- Nausea

The side effects listed are not all the side effects seen with KUVAN. You should talk to your doctor or pharmacist if you have concerns about these or other side effects.

Ingredients of KUVAN

Active Ingredient: sapropterin dihydrochloride.

Inactive Ingredients: ascorbic acid, crospovidone, dibasic calcium phosphate, D-mannitol, riboflavin, and sodium stearyl fumarate.

177

Kuvan Tablets are mottled, off-white to light yellow, and debossed with "177".

KUVAN is a product of BIOMARIN
More information available at Kuvan.com

Adapted from *Frequently Asked Questions About KUVAN*. BioMarin Pharmaceuticals Inc., 2010. Print.

TEACHING AID

The Altitude Of Your Attitude

Objective: To measure gains on a personal improvement scale, not by comparison with others; to determine how attitude affects reaching individual goals of diet compliance.

Handout Needed
The Altitude of Your Attitude

How It's Done

Use the "The Altitude of Your Attitude" handout

- Open conversation about emotional ups and downs.

Ask each participant to identify where they are on the mountain.

- How does that compare with where they were at the last visit?
- How does one measure personal improvement?
- How do you feel when you've reached the top of the mountain?
- What are some of the struggles that can keep you from reaching the top?

HANDOUT: The Altitude Of Your Attitude

THE SKY'S THE LIMIT!

How High Will You Climb?

TEACHING AID

How Do You Feel?

Objective: To increase the individual's understanding of their feelings around special occasions and being on diet.

How It's Done

Use the handout "How Do You Feel?"

- Present the participants with a scenario in which they are with a group of people who are not on a diet.
  Here is an example of what your could say:
  - *After school, your friends decide to go to the local restaurant and order burgers and french fries. How does this make you feel?* Participant Response.
  - *If your friends respond* use different possibilities, *how does it make you feel?*_____
  - *How does it make you feel not to eat the same food your friends are eating?*_____
- Ask each participant to identify his/her own feelings from the different facial expressions on the handout.
- Discuss why people want to be alike and how being alike makes them feel. Stress that while there are similarities in people, being the same is an illusion.
- Help each participant to identify how one emotion or feeling can lead to another (i.e. embarrassment leads to anger). Discuss the benefits of being able to identify their emotions in difficult situations, and how that can improve their ability to understand themselves and stay on their diet.
- Around holidays, this activity is especially effective to elicit discussion about being on a restricted diet during special occasions.

Handout Needed
How Do You Feel Today?

How Do You Feel?


Aggressive


Agonized


Anxious


Apologetic


Arrogant


Bashful


Blissful


Bored


Cautious


Cor tin g


Confident


Curious


Determined


Disappointed


Disapproving


Disbelieving


Disgusted


Distasteful


Ecstatic


Enraged


Envious


Exasperated


Exhausted


Frightened


Frustrated


Grieving


Guilty


Happy


Horrified


Hurt


Hysterical


Indifferent


Idiotic


Innocent


Interested


Jealous


Joyful


Lonely


Mischievous


Miserable

Negative

Obstinate

Optimistic

Pained

Paranoid

Perplexed

Prudish

Puzzled

Regretful

Relieved

Sad

Satisfied

Shocked

Sheepish

Skeptical

Smug

Surly

Surprized

Suspicious

Sympathetic

Thoughtful

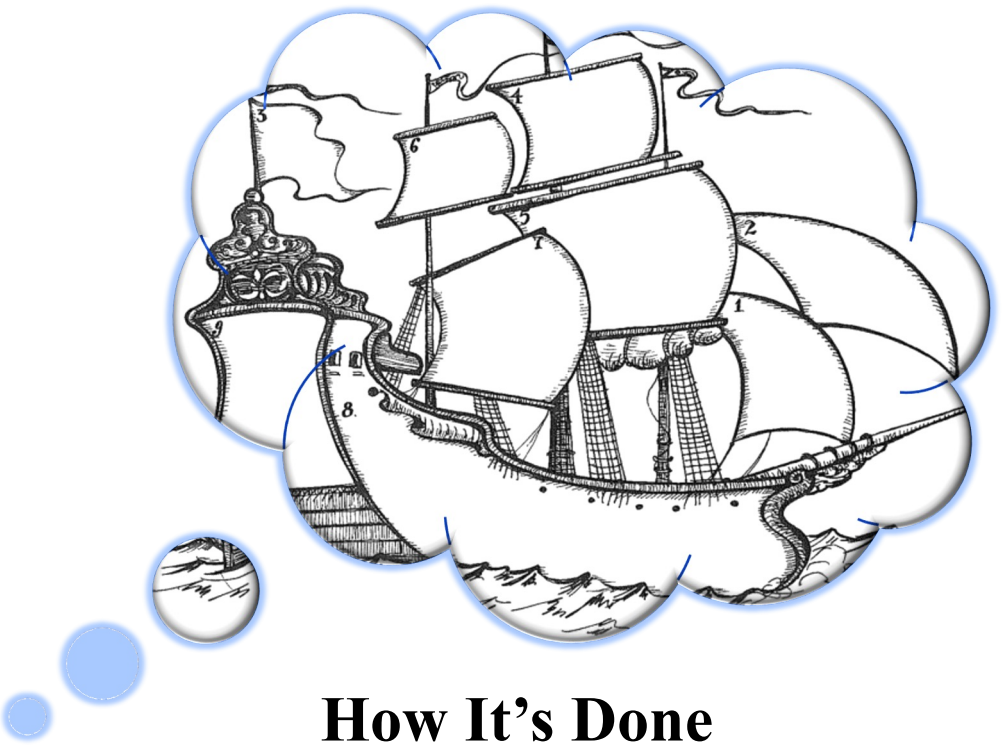
Undecided

Withdrawn

TEACHING AID

Self Talk

Objective: To improve self-esteem by learning to use positive internal messages regarding differences and being on the diet.



How It's Done

*Use the handouts from **Making Choices**:*

- Distribute a copy of "The Personal Ship" to each participant and ask them to complete the activity as described on the sheet.
- Encourage participants to be creative and to share ideas. Another activity for this teaching aid is the handout "How Well Do You Like Yourself?" and "Scale For Self Esteem".
- Ask individuals to think critically and honestly about their answers.

Additional Activity:

- The handout "Do You Hear Yourself? Are You Listening?" to discuss the impact of self talk, both positive and negative.
- Discuss ways to consciously change thinking towards positive internal messages.

Handouts Needed

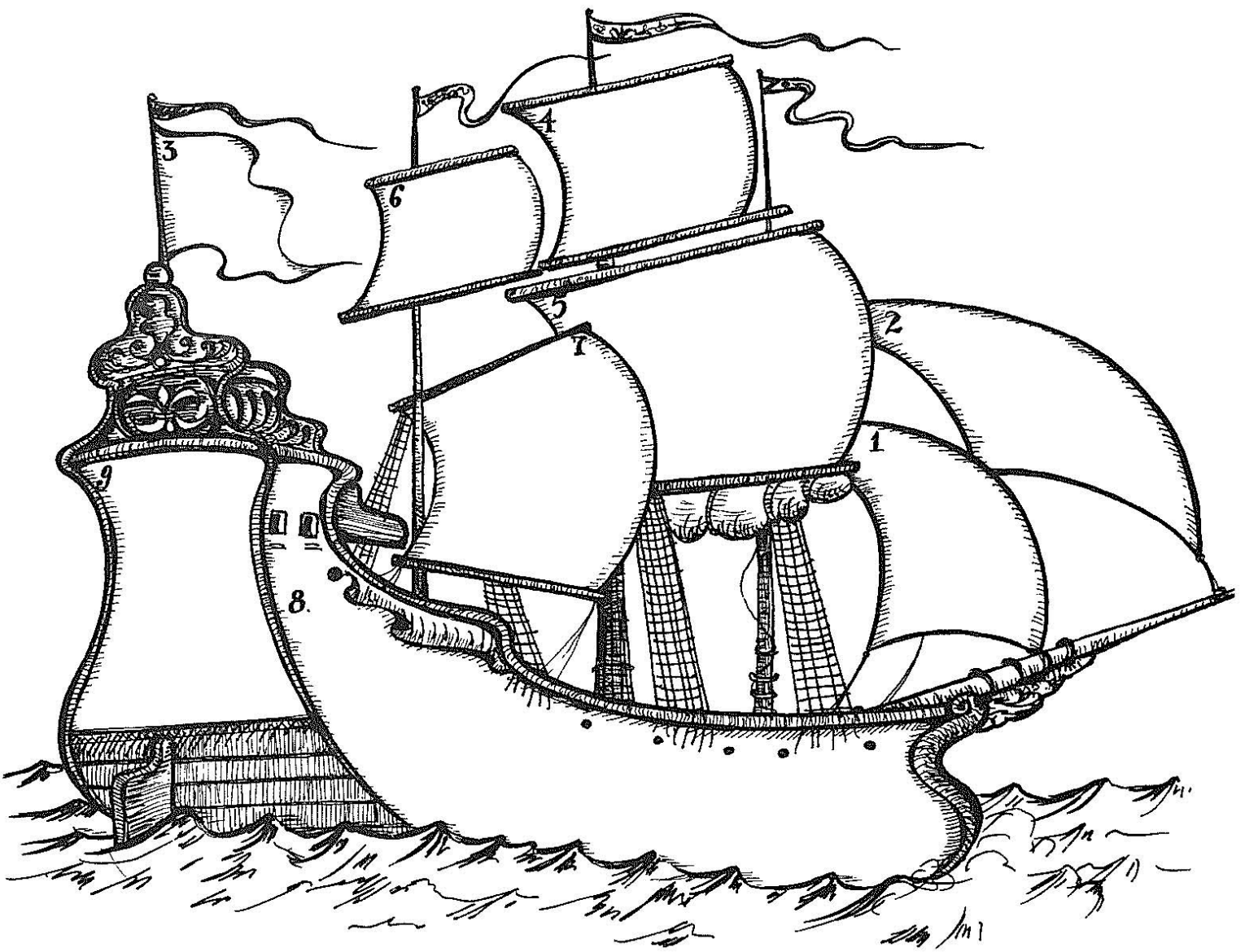
- The Personal Ship
- How Well Do You Like Yourself?
- Scale For Self Esteem

Adapted from M. Halter and B. Fierro Lang Making Choices

Personal Ship

Complete your Personal Ship by writing or drawing something in the numbered space which corresponds with the numbers below and would indicate the answers to the following questions:

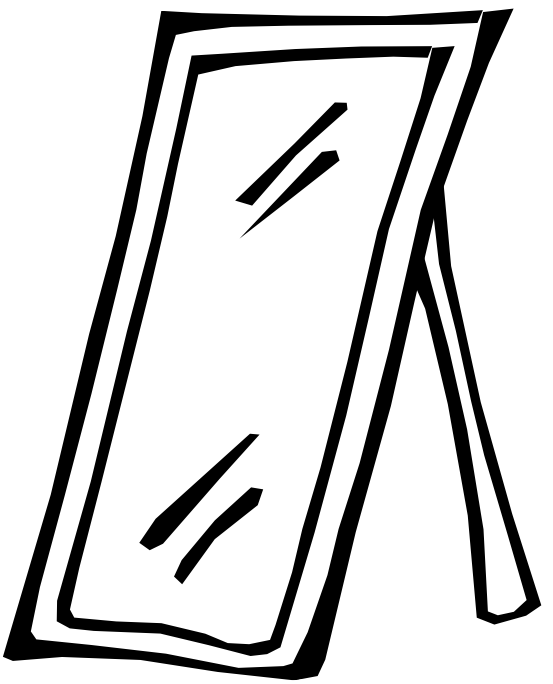
1. My favorite possession
2. What I do best
3. Greatest success in the past 12 months
4. Unrestrained by money and commitments, what I would do in the next 12 months
5. Three words that best describe me
6. What I am really trying to get better at
7. Three successful experiences I've had in my life (fun and success)
8. Biggest mistake that I learned frorn
9. Three words I would like people to use to describe me



How Well Do You Like Yourself?

Complete the survey below using the following scores:

15 = True almost all of the time
10 = True most of the time
5 = Sometimes true
0 = If it's seldom true



___ 1. I am kind and loving.

___ 2. I am an optimistic person.

___ 3. I make friends easily.

___ 4. I am happy with my life the way it is.

___ 5. I look forward to each new day.

___ 6. I would rather be me than anyone else.

___ 7. If I were someone else, I would choose me for my friend.

___ 8. When I feel strongly about something, it feels good to express my feelings.

___ 9. Regardless of my grades, I feel good about the work I do.

___ 10. Other people treat me nicely most of the time.

___ 11. My life is very interesting.

___ 12. Most of the time I am in a good mood.

___ 13. When I make a mistake I can laugh at myself.

___ 14. My energy level is usually high.

___ 15. I get plenty of exercise and eat well.

___ 16. I am happy with the way I look.

___ 17. If I were a person of the opposite sex, I would want to ask me for a date.

___ 18. My friends and family value my ideas and listen to what I say.

Scale For Self Esteem

Total Score	What Your Score Means
200-225	Very high self esteem, positive self-worth. You like yourself.
120-195	Self-esteem and self worth are good. You accept yourself and your life. Self image could be improved with conscious effort.
45 -115	Self esteem is so-so. Acceptance of yourself and your self worth is less than desirable. You need to work on improving your self image. You may want to practice being your own best friend and follow some of the advice given below.
Below 45	Very low self esteem and self worth. Health and personal growth could suffer. You need to develop relationships which give you encouragement and support. You also need to work on changing your own negative attitudes and behaviors. It might help to talk about this with someone you trust. Try to practice the advice given below.

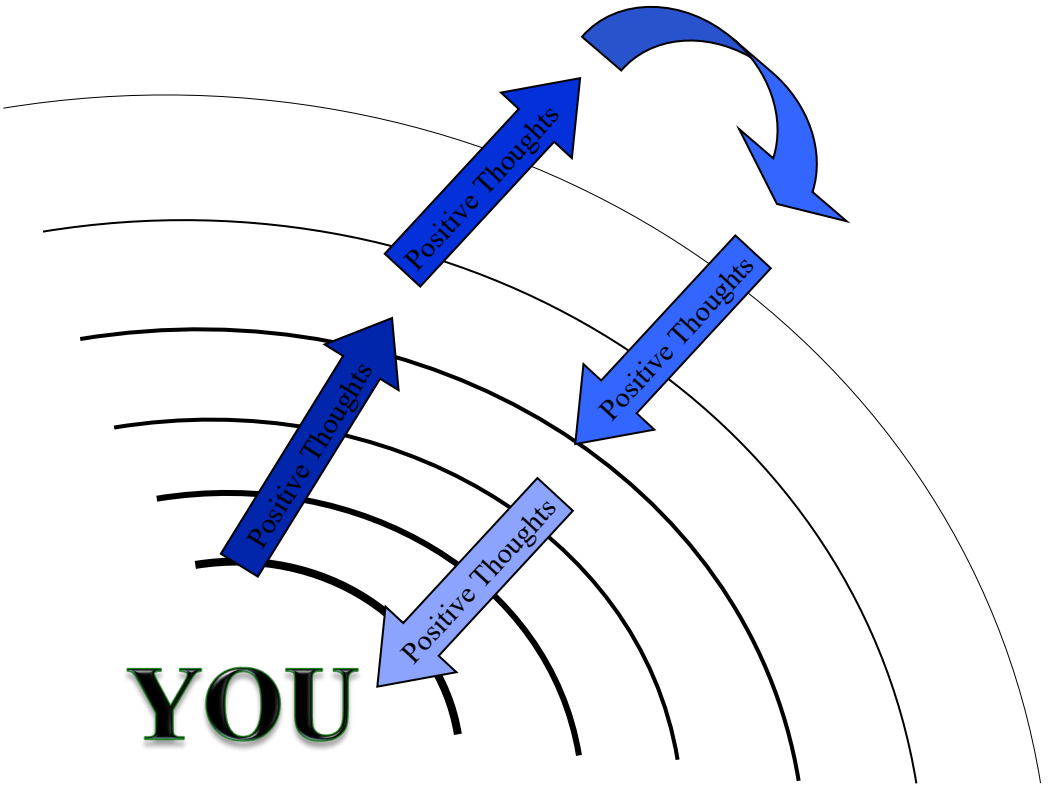
Tips To Help Improve Your Self Esteem

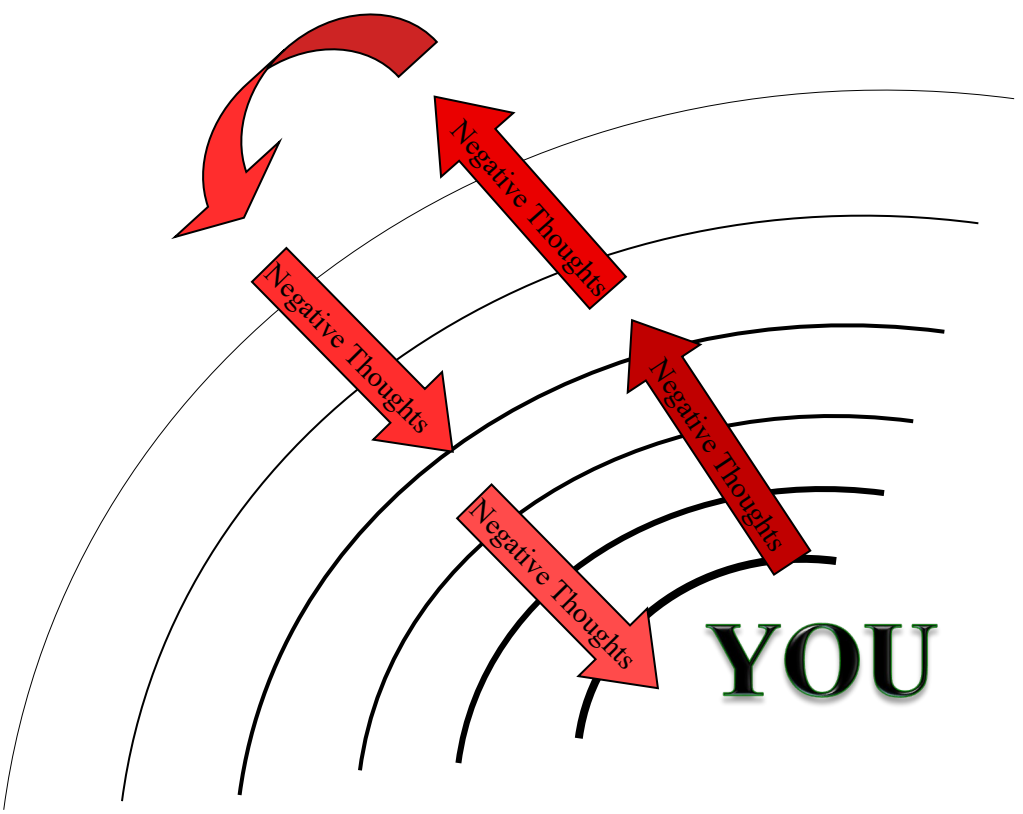
- One of the best ways to improve our self esteem is to be nice to ourselves. Begin saying nice things to yourself. Start each day with a positive comment to yourself about yourself and reinforce that comment throughout the day.
- Surround yourself with positive people and people who are positive about you. Don't allow others to put you down.
- Accept yourself as you are right now and begin to work to change those things that you don‘t like or want to improve about yourself.
- Set goals for yourself - small, medium and large. Congratulate yourself as you accomplish even the smallest of goals.
- Substitute "I can" and "I'll do my best" for "I can't" in your vocabulary. Begin to see yourself as successful and capable of reaching your goals.

Start Now & Never Give Up!

Adapted from Page 61, Making Choices

Do You Hear Yourself?
Are You Listening?

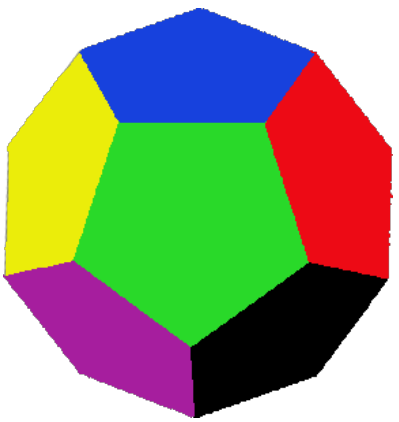




TEACHING AID

Who Are You?

Objective: To promote self reflection about one's own traits and to identify differences and commonalities between participants, including genetic disorders.



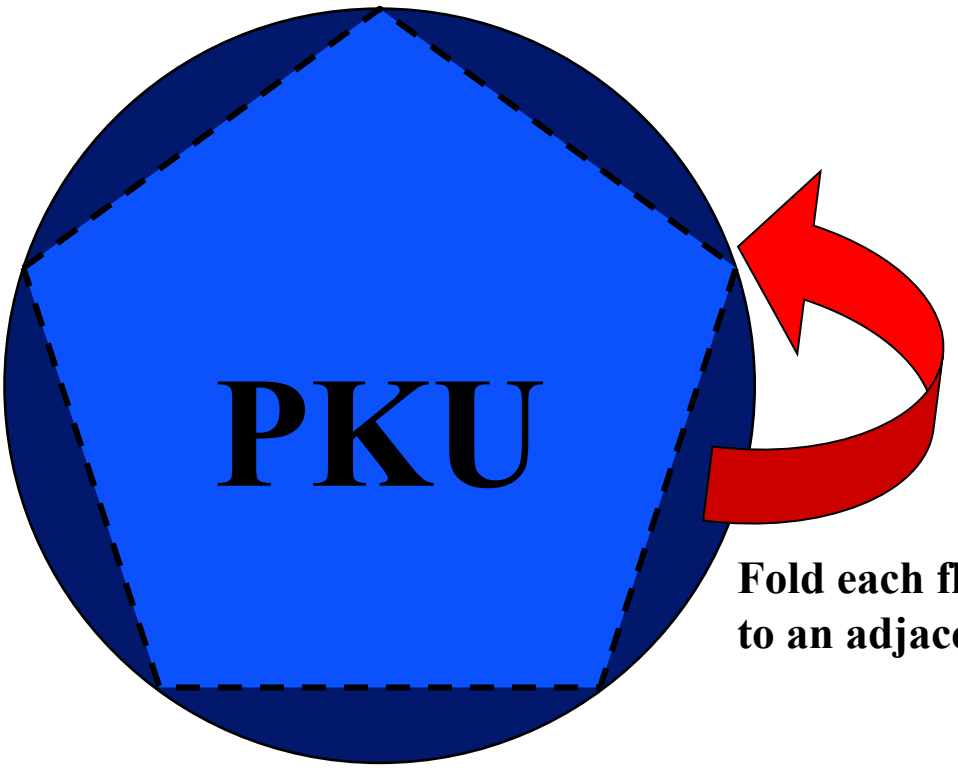
How It's Done

Use the handout "Who Are You?"

- Provide each individual with 12 colored paper circles (approximately 2-3 inches in diameter).
- Have individuals to write to one aspect of their life on each colored circle.
  - One circle MUST say PKU.
  - Other circles may include: friends, family, sports, music, etc.
- Fold each circle near the edges (about ¼ inch) so that it has five small flaps (see handout).
- Glue each flap on the main circle to a flap on another circle. Each circle will touch 5 other circles. Continue this process until all 12 circles are used to create a sphere.
- When completed, the sphere represents the individual's life, made of components of what makes them unique. Discuss that if any of the pieces were missing, the person would be different and the sphere would not be complete. Comparison between different balls can be utilized to discuss similarities and differences between peers. PKU is the item that everyone in that group has in common.

Handout Needed
Who Are You?

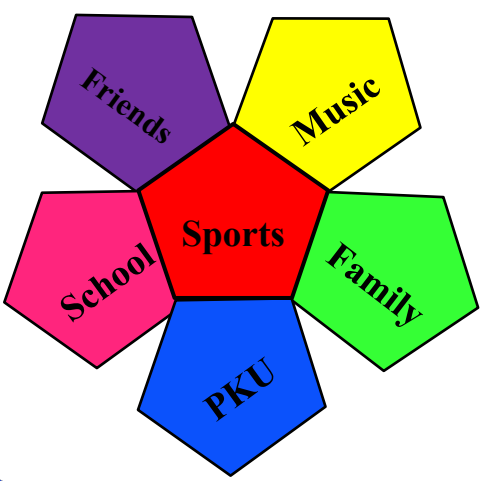
Who Are You?

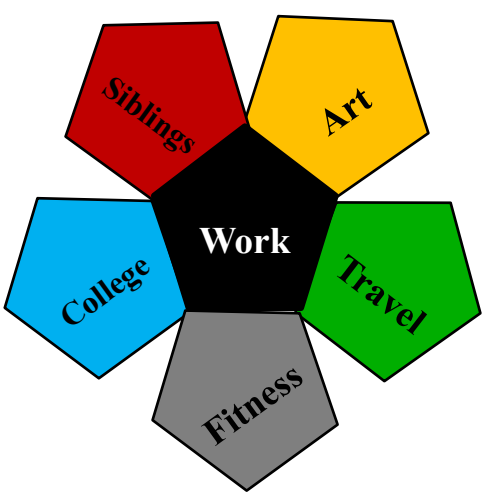


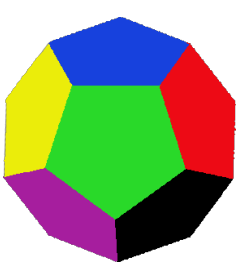
Fold each flap and glue to an adjacent circle

This is an example of an individual circle.

Each flap will be glued to a flap on an adjacent circle. Circles will be surrounded on all 5 sides to create a spherical dodecahedron (12 sides), similar to the appearance of a soccer ball. Flaps should be folded away from the words.





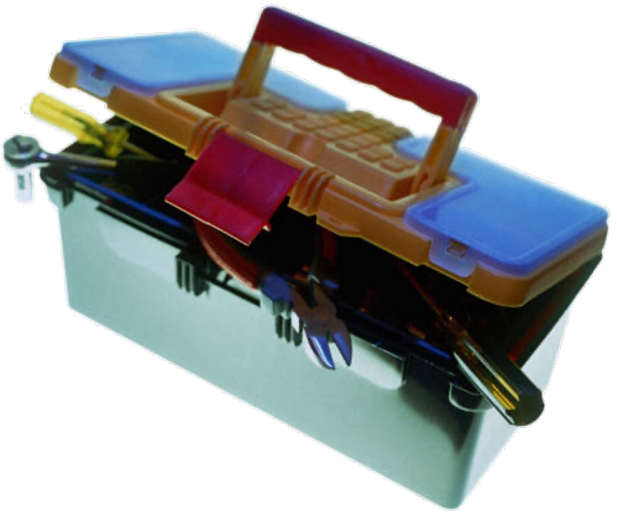


You will need a total of 12 brightly colored circles.
Label each one with something that represents you.

TEACHING AID

Problem Solving Tool Kit

Objective: To provide individuals with tools to manage and successfully resolve problems relating to PKU management.



How It's Done

Use the handout "Problem Solving Tools"

- Discuss key elements in problem solving beginning with defining the problem. Discuss negotiation tools and resolution tools including listing the solutions.
- Ask individuals to think of a recent conflict that they have had. Individuals should write down their conflict using "I Messages." These should include how they feel about the problem and what they want to have happen. Individuals should answer the questions:

  "How did I resolve this conflict?"

  "Did I use effective problem solving tools?"
- If problem solving tools were not used, discuss how the scenario could be changed to incorporate these tools. Would the outcome change?
- Discuss with group specific examples of how they might use these new tools in future situations.

Handouts Needed

- Problem Solving Strategies
- Problem Solving Worksheet
- I Messages

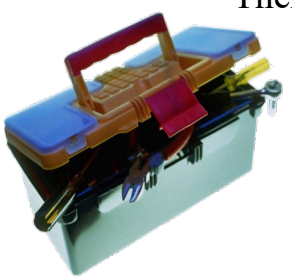
Adapted from M. Rapoff, Adherence to Pediatric Medical Regimens

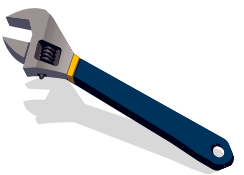
Problem Solving Strategies

Everyday we face situations which require us to use problem solving tools. These strategies become even more important when we are confronted with peer pressure and busy schedules. When the outcome of your decisions directly affects your health, it is especially important that you take time to think about solutions you choose for your dilemmas.

Effective problem solving generally involves five steps:

1. **Identify**. Recognize and be able to define exactly what the problem is.
2. **Brainstorm**. Develop a list of possible solutions.
3. **Plan**. Choose what you feel is the best solution, make a plan and follow it!
4. **Improve**. Decide if your solution was effective or not and make changes.
5. **Never Give Up!** If your plan didn't work, decide what you need to change. Then, go back and try something else!



Effective problem solving takes practice and consistency. Some of the strategies listed below may help to have a complete tool kit.

- Keep a journal to identify situations when you are tempted to compromise your diet. When you see patterns developing, try to think through and anticipate solutions.
- Use the idea of "self-talk" to reinforce positive thoughts about yourself, your diet and your management plan. The more you cycle negative messages, the less likely you will be to follow your diet plan.
- Talk through scenarios with other young adults who have chronic disorders, friends, clinic staff, siblings or parents. Discuss hypothetical situations and brainstorm possible solutions.
- Mentally role-play situations before they occur. Picture yourself making constructive choices despite peer pressure.
- Set up your own consequences and stick to them. Allow yourself limits and rewards.
- Create a contract with clinic staff and your parents which will encourage you to maintain your diet. Include both positive and negative consequences depending on the choices you make.

Adapted from Michael A. Rapoff <u>Adherence to Pediatric Medical Regimens</u>

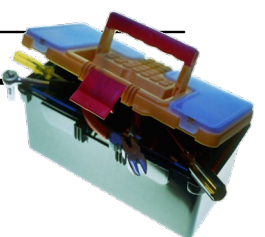
Problem Solving Worksheet

1. **Identify the problems with compliance:**

2. **Brainstorm a list of possible solutions for being more compliant:**

3. **Draw a star by what you feel is the *best* solution. Below, list the steps necessary to make your plan work:**

4. **After you put your plan into action, decide if your solution was effective or not.**
   What would you suggest now to change or improve your solution:



Example

5. **Don't give up! Put your changes and improvements to action!**

1. Identify: "I don't always drink my formula at school."

2. Brainstorm: "Store formula in refrigerator in main office; to me, it tastes better cold."
and/or "Use a new water bottle with a straw; it won't look or smell as different"
and/or "Keep it in my locker and drink during passing periods."

3. Plan:
1) Go to clinic store and get new water bottle.
2) Weigh and measure my formula in the morning so it's ready to go when I leave for school.
3) Remember to take my water bottle with me.
4) Bring my water bottle to lunch with my friends.
5) Drink all of the formula in the bottle before 5th period class.
6) Bring water bottle home every night to clean and refill.

4. Improvements: ***Problem*:** Formula still gets warm. ***Solution*:** Keep a Blue Ice pack in my locker.

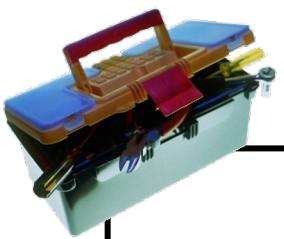
Adapted from Michael A. Rapoff <u>Adherence to Pediatric Medical Regimens</u>

I Messages

"I messages" are great tools. They provide a way to express your feelings and effectively communicate with someone without putting them on the defense. "You messages," by contrast, put someone in an argumentative position of having to defend themselves. Try using this strategy to rephrase your thinking and communication.

"I Messages" Involve 4 Steps:

1. State the feeling or problem
2. Describe the behavior
3. Explain why the behavior is a concern
4. Define what you want to see happen/change



Sample Situation

You Want To Say A "You Message"

"You are so irresponsible! You never drink your formula without being reminded and you're risking your health by cheating on your diet!"

Instead You Say An "I Message"

"I feel frustrated and concerned when I see your diet being compromised Because I know it can affect your health and attitude. I want to see you take more responsibility for following your diet.

Try This Situation:

You want to say a "**You Message**"

"You are control freaks! You never trust me to take care of myself and you're always butting into my business!"

Rephrase it, and instead you say an "**I Message**"

I feel _____

when _____

because _____

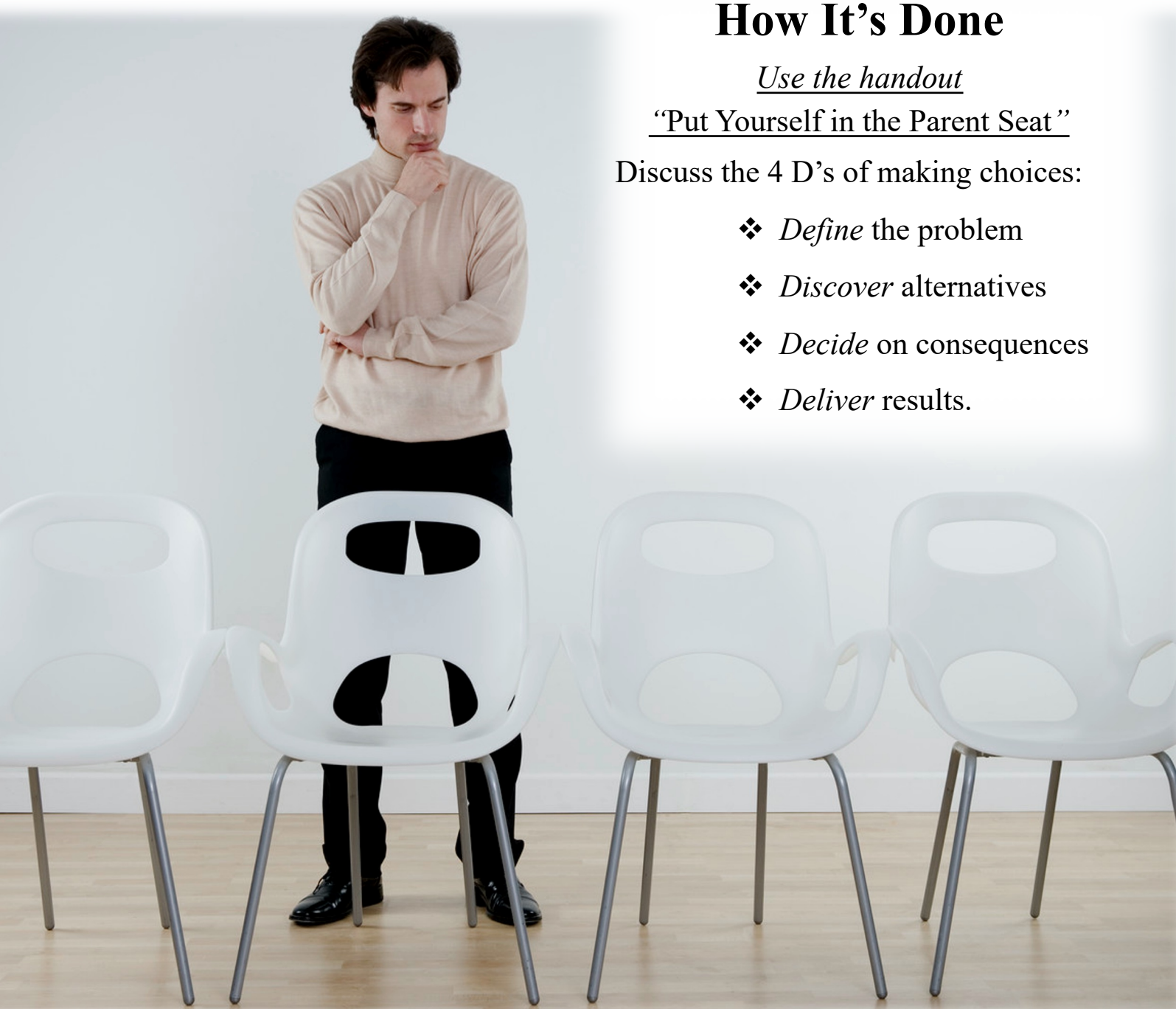
I want _____

Adapted from Michael A. Rapoff Adherence to Pediatric Medical Regimens

TEACHING AID

Put Yourself in the Parent Seat

Objective: To recognize that parents and adolescents may interpret the same scenario very differently; to open communication and discuss various points of view.



How It's Done

Use the handout

"Put Yourself in the Parent Seat"

Discuss the 4 D's of making choices:

- *Define* the problem
- *Discover* alternatives
- *Decide* on consequences
- *Deliver* results.

Handout Needed
Put Yourself in the Parent Seat

Adapted from M Halter and B. Fierro Lang Making Choices

Put Yourself In The Parent Seat

Scenario: Donna, a 16 year old with PKU, was asked by her high school friends to join them at McDonald's on Friday after school. Because Donna doesn't like to drink her PKU formula at school, she is supposed to go straight home after school to drink it. She decides to go with her friends instead.

When Donna returns home, her mother and father are concerned because they know she did not drink her PKU formula. You are the parent; respond to the following:

Define the problem**:**

I

feel_____

when

you_____

and I

wish_____

Discover and share alternatives to the unacceptable behavior. What could Donna have done differently?

_____

_____

_____

___

Decide on consequences. Be sure they are fair, firm, logical, and can be clearly Measured and understood by Donna. Remember: the goal is to teach, not punish!

_____

_____

_____

___

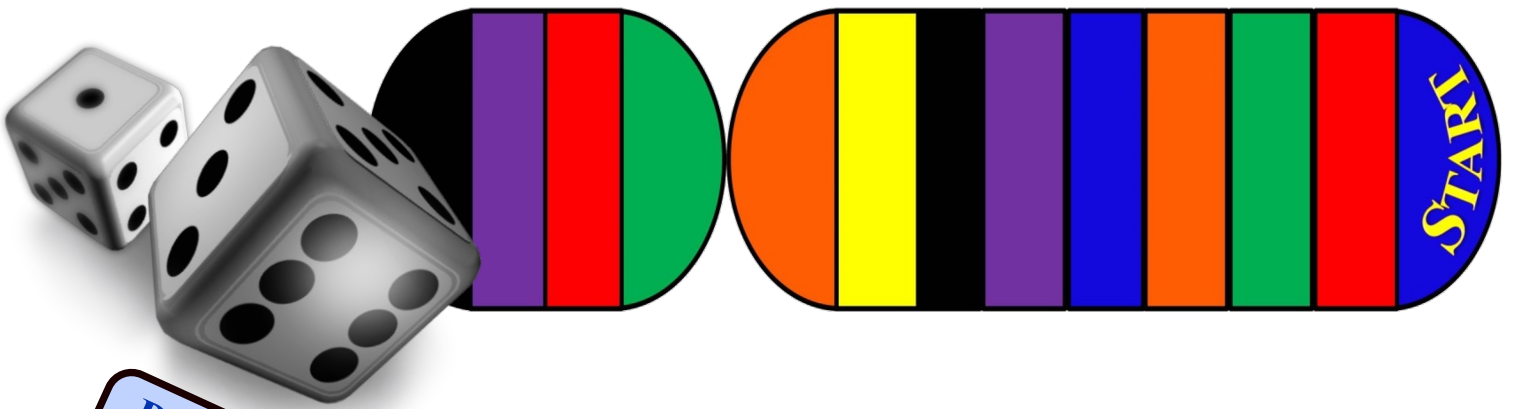
Deliver results: Will the consequences above accomplish the desired change in behavior? Consider the following questions:

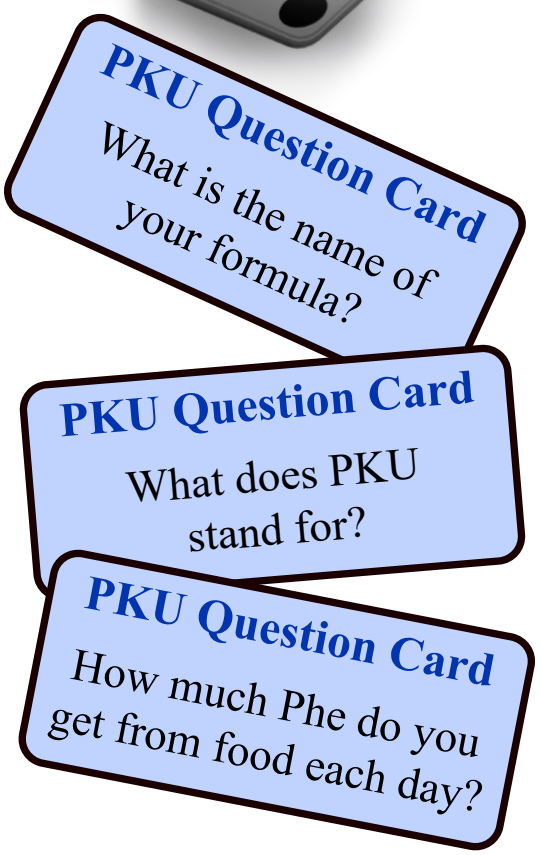
- What are the medical consequences for Donna if she chooses to be noncompliant?
- What was the specific behavior that was unacceptable?
- What could Donna's parents do differently in the future?
- Do Donna and her parents understand what is expected in the future?
- Were expectations clearly stated that Donna will know exactly what consequences to expect if the behavior continues?

TEACHING AID

PKU Adventure Game

Objective: To assess the Hyperphenylalaninemia knowledge base while providing a fun learning environment.





Materials Needed

- PKU Adventure Game
- PKU Question Cards
- Dice
- Playing pieces

How It's Done

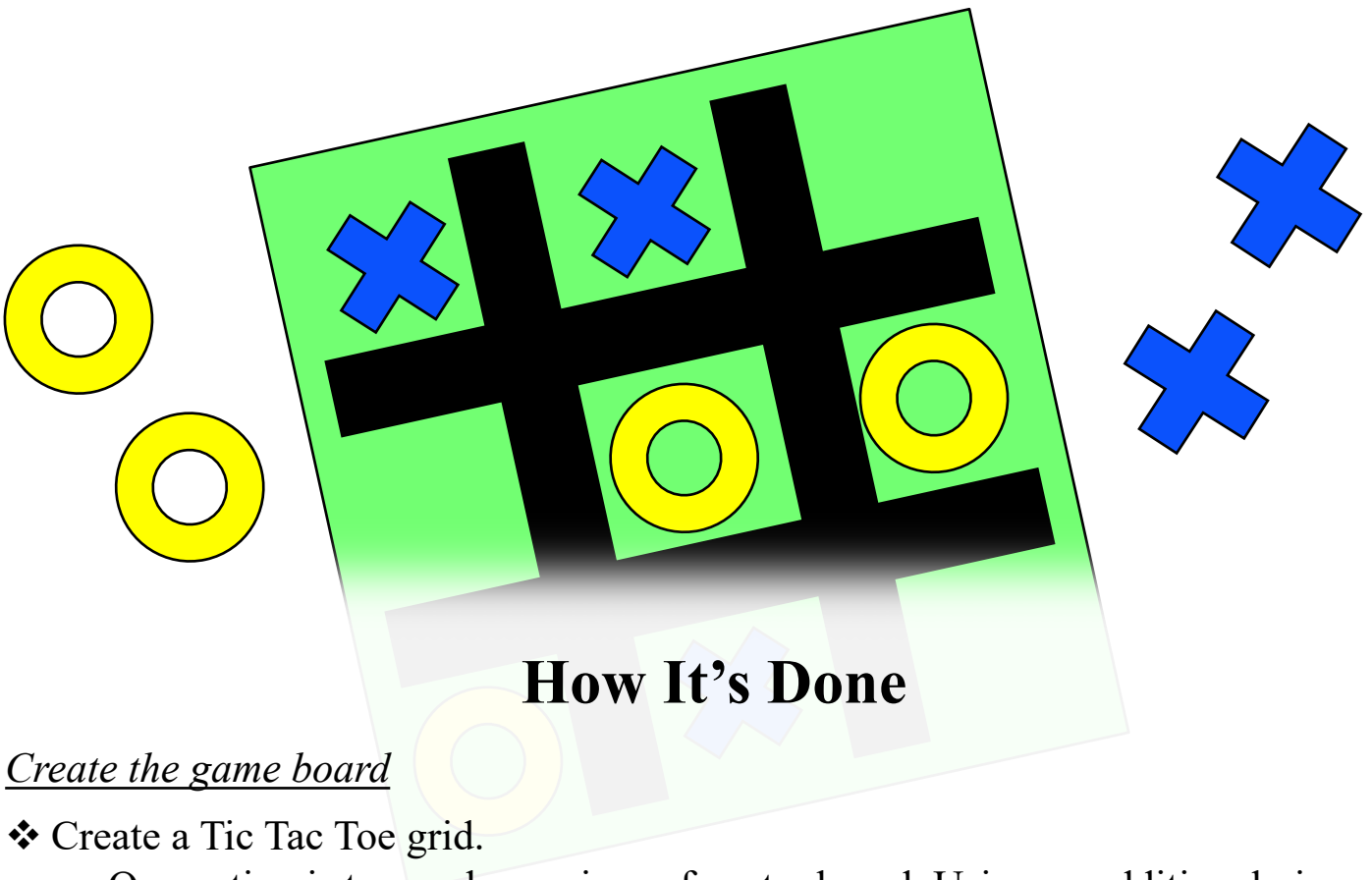
Set up a board game

- Use an existing game board, create one, or purchase a ready to use **PKU Adventure Game.**
- Create question cards that are appropriate to the participant's age and level of understanding.
- Provide a game piece for each participant.
- Each participant will role the dice at the beginning of their turn.
- A question card is drawn and the questions is asked.
  - A correct answer allows the player to move their game piece ahead the number of spaces determined by the number they rolled on the dice.
  - An incorrect answer results in no movement of the game piece.

TEACHING AID

Tic Tac KNOW

Objective: To review knowledge and encourage discussion of various aspects of hyperphenylalaninemia.



How It's Done

Create the game board

- Create a Tic Tac Toe grid.

  One option is to us a large piece of poster board. Using an additional piece of poster board, create10 smaller squares and label 5 with "X" and 5 with "O." Velcro strips placed in the boxes and on the back of the squares help to keep the game board in place or allow it to be propped up.

Play the game

- Individuals earn a chance to place one of their playing pieces by correctly answering a question about PKU, their diet, health, etc.
- This game can be played by two individuals or in teams (adolescents versus parents is a fun way to test knowledge of diet).
- By changing the types and difficulty of questions asked, this activity can be modified for any age level.

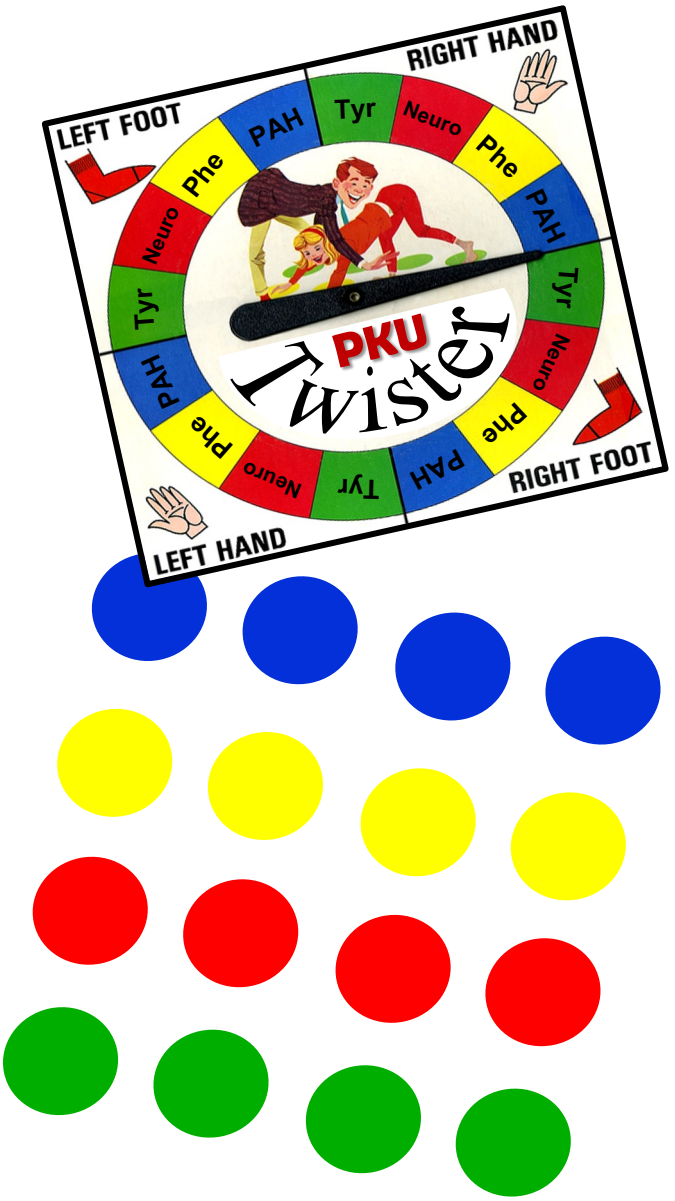
Materials Needed

- Tic Tac Toe Game
- Trivia Questions

TEACHING AID

PKU Twister

Objective: To reinforce the topics commonly discussed in group: phenylalanine, PAH, tyrosine, and neurotransmitters.



Materials Needed

❖ Twister Game

Handout:

Twister Questions

How It's Done

Using a Twister Game

❖ Mark each color on the twister spinner board with a different topic; Phenylalanine, PAH, Tyrosine, Neurotransmitters (or make up other topics you want to discuss).

❖ When the spinner lands on a color/topic a question from that category will be asked (see Twister Questions Handout). The participants will place the appropriate hand or foot on the colored dot.

Example:

Question: What is the amino acid that there are high levels of in PKU?

Answer: Phenylalanine (move appropriate hand or foot to yellow dot).

❖ Questions should be kept relatively simple as this is just an exercise to help reinforce major topics. By changing the types and difficulty of questions asked, this activity can be modified for any age level.

❖ This game can be played by up to four

PKU Twister Questions

Phenylalanine

- What is the name of the amino acid that is high in untreated PKU
- In PKU the treatment range of what should be a level of 2-6mg/dl or 120-360umol/l?
- In untreated PKU the amino acid that can cause irreversible brain damage in infants is what?
- What is the amino acid that is broken down into tyrosine?
- In PKU we count either protein or milligrams of _____?
- Many adults state that if (blank) levels are high they have headaches and feel tired.
- The Phe level that we always need to maintain within treatment range is (blank)?

Tyrosine

- What is the amino acid that phenylalanine gets broken down into?
- What is the name of one amino acid that is important for making the chemicals that help our brain cells communicate?
- What is the amino acid that we get in our medical food (formula) that we cannot make in our bodies?

PAH

- What is the most important enzyme in the PKU pathway?
- What is responsible for breaking down Phe in Tyr?
- BH4 is a cofactor for this enzyme.
- If we have very little to none of this enzyme we have classic PKU.

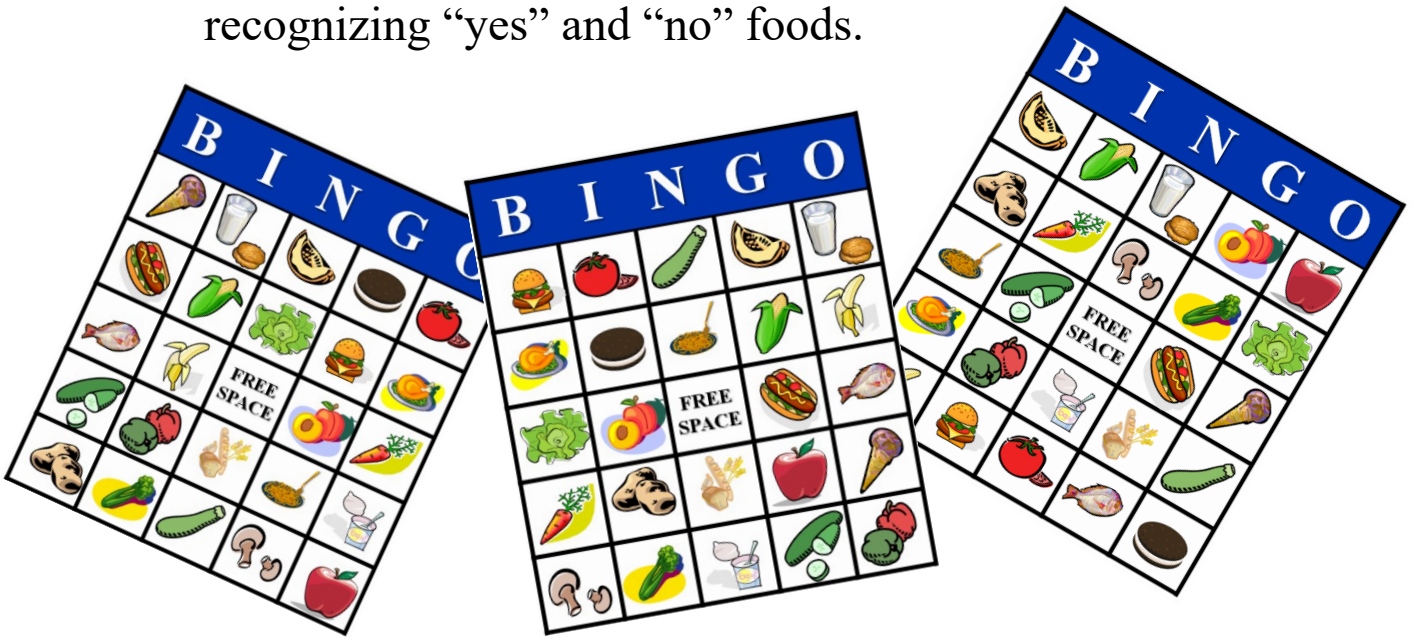
Neurotransmitters

- These are the chemicals that help our brain cells communicate.
- Low tyrosine levels might keep these from working well.
- High phenylalanine levels in blood may keep these message centers from working well.
- If you have low tyrosine and decreased messages to the (blank) you may have a greater challenge being organized.
- Dopamine and serotonin are (blank).

TEACHING AID

"Yes" "No" BINGO

Objective: To increase participants understanding of their diet by recognizing "yes" and "no" foods.



How It's Done

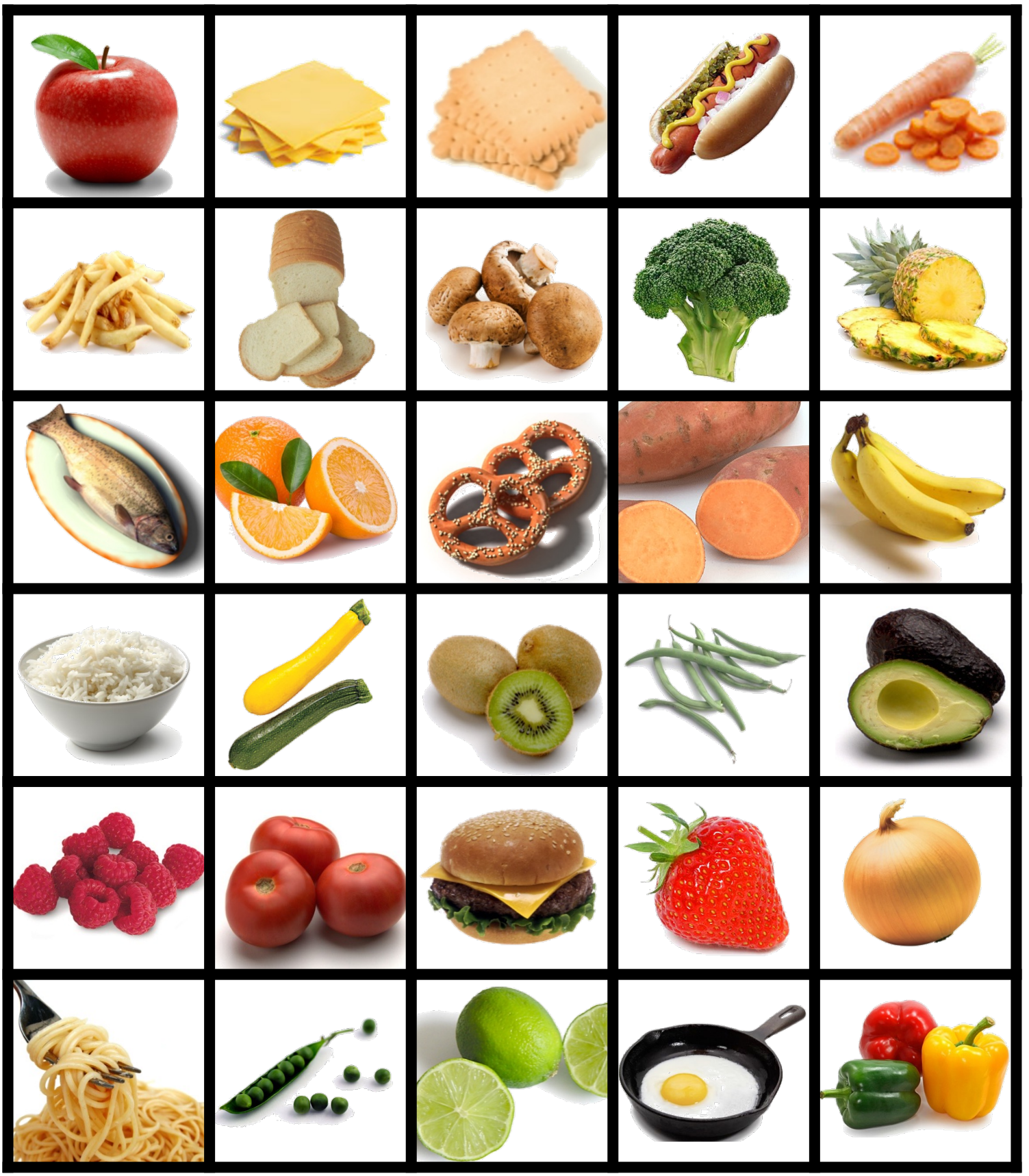
This game can be as simple or complex as you decide to make it. For younger participants, limit discussion to "yes", "no", and "sometimes" foods. If participants are older, lead discussion to include topics such as serving size, cooking method, etc.

- Using the cut out game pieces mixed in an envelope, draw one square and call the name of the food out load. Have participants mark the appropriate square with a piece of candy, and have the participants say if it is a yes, no, or sometimes food (for older participant, begin to discuss what a typical serving size is and how much Phe is in one serving).
- Once a participant has five in a row (in any direction) they should shout "BINGO". To prove they have a BINGO they will need to read off the foods that make up their BINGO, and say if the food is a yes, no, or sometimes food.
- Game boards can be shuffled or traded between participants and all game pieces put back into the envelope , this game can be played until all the foods have been discussed.

Materials Needed
- Bingo Game Pieces
- Bingo Game Boards
- Candy (as markers)

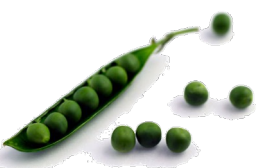
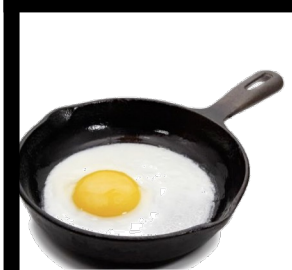
BINGO GAME PIECES

Cut out these game pieces. As you draw them, have the participants place candy on the appropriate square and discuss each food until someone get's 5 in a row. Discuss that some of these foods are only "yes" foods if they are a low protein option (i.e. rice, bread, pasta).



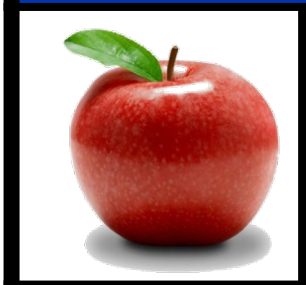
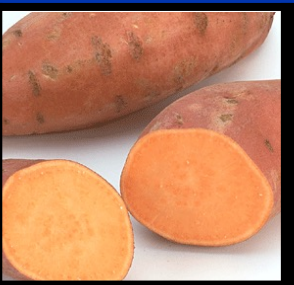
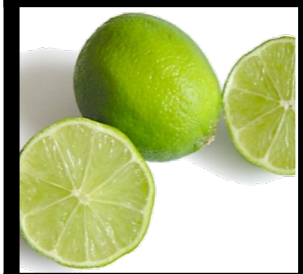
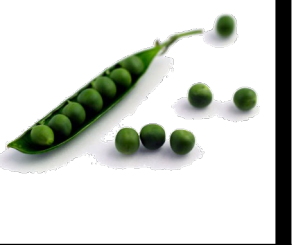
BINGO GAME BOARD

Place candy on the appropriate square and discuss each food until you get five in a row.

B	I	N	G	O
				
				
		**FREE SPACE**		
				
				

BINGO GAME BOARD

Place candy on the appropriate square and discuss each food until you get five in a row.

B	I	N	G	O
				
				
		FREE SPACE		
				
				

BINGO GAME BOARD

Place candy on the appropriate square and discuss each food until you get five in a row.

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				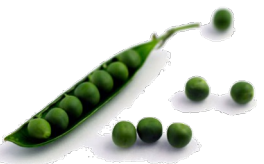
				
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				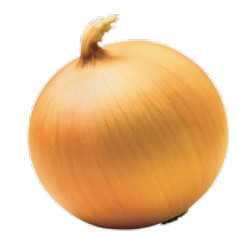
		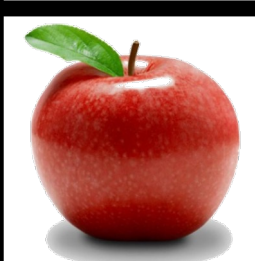		

BINGO GAME BOARD

Place candy on the appropriate square and discuss each food until you get five in a row.

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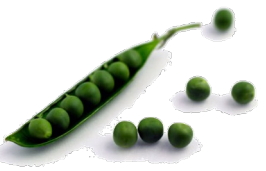
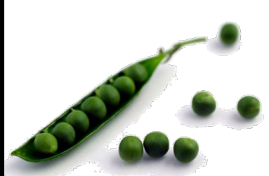
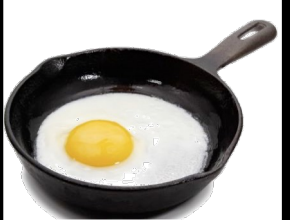
				
				
		**FREE SPACE**		
				
				

BINGO GAME BOARD

Place candy on the appropriate square and discuss each food until you get five in a row.

B	I	N	G	O
				
				
		FREE SPACE		
				
				

TEACHING AID

PKU Jeopardy

Objective: To review prior knowledge and encourage discussion and updates of important concepts related to PKU.

GENETICS	HEALTH	DIET	KUVAN	FOOD	PKU
$200	$200	$200	$200	$200	$200
$400	$400	$400	$400	$400	$400
$600	$600	$600	$600	$600	$600

How It's Done

This activity is based on the game show Jeopardy.

- Create 5 categories that may include: Management, Genetics, Foods, Health, Diet, or other categories applicable to prior teaching.
- For each category label five index cards with $200, $400, $600, $800, and $1000 respectively and on the back, write a question (and its answer) pertaining to that category. Remember that more difficult questions are worth more money. Continue until you have 5 questions per category.
- Tape index cards in columns onto a poster-board in the appropriate columns with $ signs showing.
- Individuals or teams earn points for correct answers as in Jeopardy.
- Encourage discussion and instruction of incorrect responses and praise understanding of important concepts.

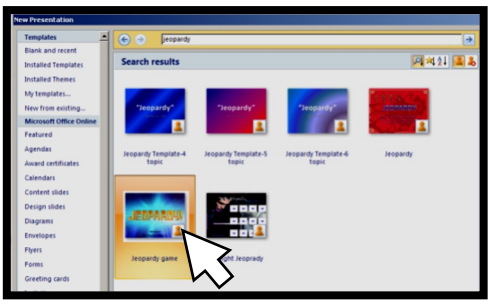
Materials Needed

- Poster-board
- Index cards
- Jeopardy Answers & Questions
- Prizes (optional)
- Laptop with PowerPoint (optional)

Option: Use the **Jeopardy Power Point Game**

JEOPARDY POWER POINT GAME

Using the Jeopardy Game Template from Power Point is a simple, fun way to customize this activity without having to recreate question cards for each age group.



Step #1: **Open A New Presentation**.

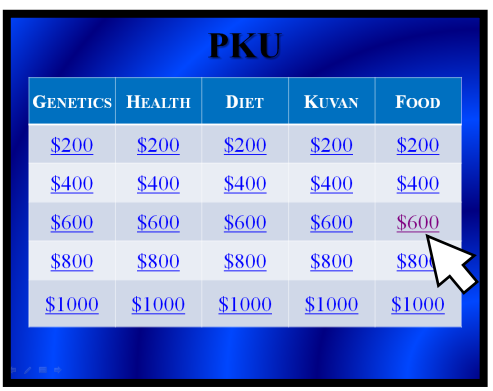
Search templates for "Jeopardy" and select the template you prefer.

Step #2: **Title the Game Topic**, such as PKU

If you have participants with metabolic disorders other than PKU, you could make the topic "Metabolic Disorders"

Step #3: **Develop Categories**.

There can be as many as 5 categories.



Step #4: **Develop Questions.**

Each category will have up to 5 questions with increasing values: $200, $400, $600, $800, and $1000. Be sure the question fits the topic, and as the value increases, the question difficulty must increase as well.

There is also the option for a Bonus Question (not shown in example).

<u>Remember:</u> In the game of Jeopardy, the questions are phrased like answers and the answers are phrased like questions. For example, instead of

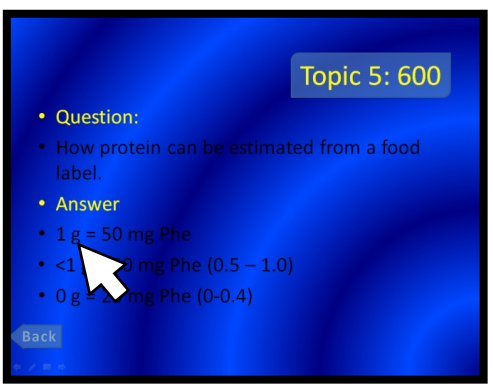
Q: What is the enzyme that is broken in PKU?

A: Phenylalanine Hydroxylase (PAH)

The question would be phrased like:

Q: The enzyme that is broken in PKU?

A: *What is* Phenylalanine Hydroxylase (PAH)



A sample Jeopardy Power Point Game is included as a separate file. Be sure to add questions about information covered in your clinic.

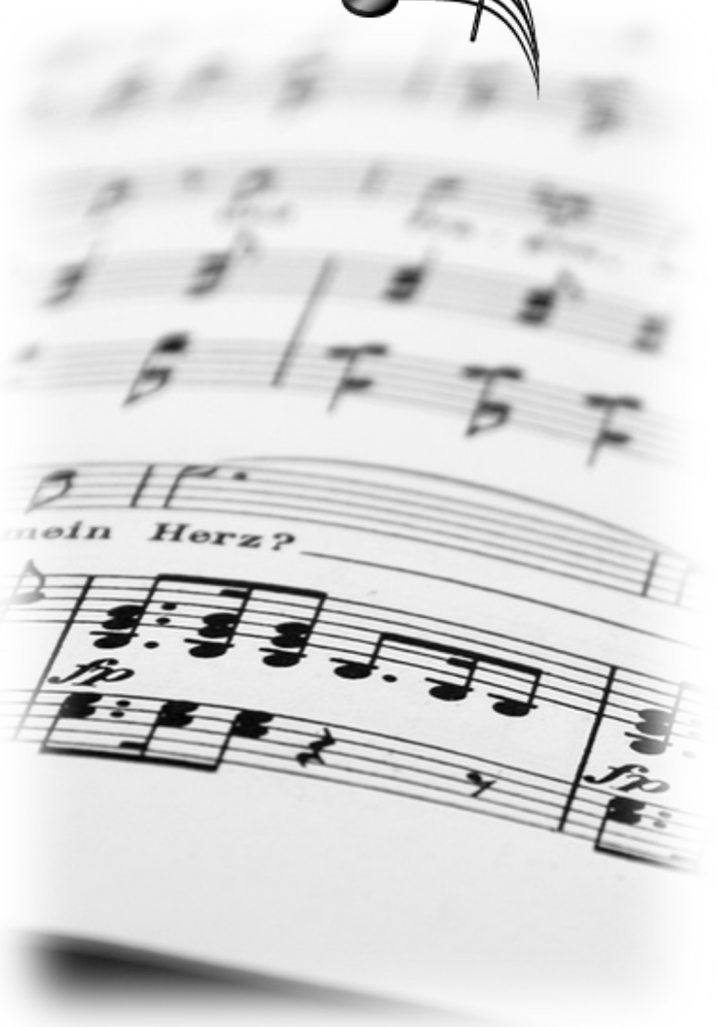
Step #5: **Play The Game!**

Play with teams or individuals. Start the slide show by hitting the F5 key. Use the curser to select the category and question value the first team selects. Always use the "Back" link after each question is completed.

TEACHING AID

The Golden Key

Objective: To use music as a medium for enhancing self image and to provide an opportunity for self-expression with respect to hyperphenylalaninemia.



Handout Needed
The Golden Key Song Sheet

How It's Done

Use the handout "The Golden Key."

- Rap to the words provided in the handout section. Have fun! Be crazy! Ask each participant to add his/her own words to the song. If needed, assist by suggesting lead-in lines such as:
  - My (friends, family, etc.) all think I'm (very cool, so unique, etc.)…

    OR
  - When I'm (alone, at school, etc.) and feeling (sad, fine, etc.)…
- Practice your creation.
- Stage a production for families or staff, or videotape the performance.

The Golden Key

"The Golden Key"

A Rap Song

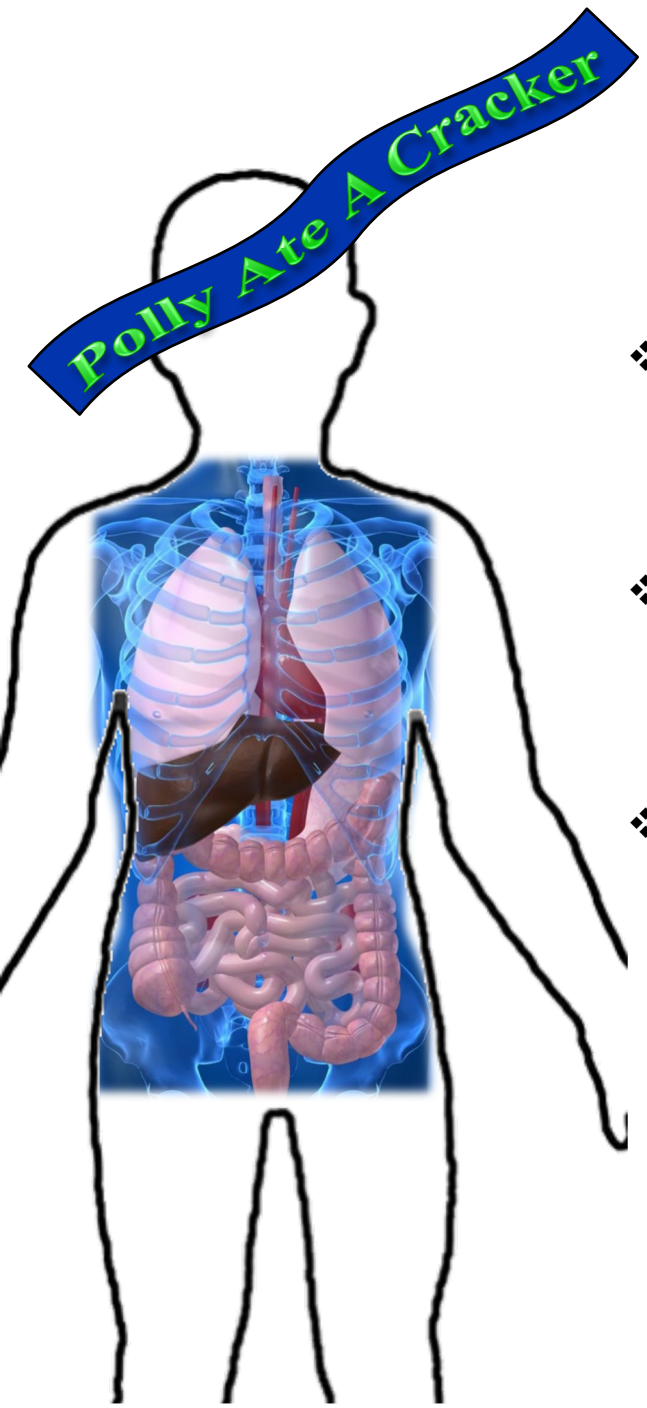
Too much **Phe** is bad for **me,**
it hurts my **brain** I must re-**frain**,
from eating **meat** and Nutra**Sweet**.
To drink my special milk's **a must,**
I'm told this by my mom I **trust.**
I weigh my for-mu-la and **food**
which keeps me in a happy **mood**.
It's **cool** you see to be like **me**,
staying on diet is the golden **key**…
the golden key, the golden key, the golden key…

I **come** to clinic and see the **group**,
it's ***"bad"*** to be part of the metabolic **loop**.
We **learn** a lot about our **genes,**
we **learn** what all those long words **mean.**
Our **diets** are different but we're **okay**,
we take care of ourselves the IMD Clinic **way**.
We're **bad**, we're **cool**,
when it comes to our diets, **We're no fools!**
Staying on diet is the golden key…
the golden key, the golden key, the golden key…

TEACHING AID

Polly Ate A Cracker

Objective: To illustrate how protein is digested, broken down into amino acids, and transported to various parts of the body.



Materials Needed

- Butcher Paper
- Markers

How It's Done

Begin with a discussion of Autosomal Recessive Inheritance.

- Using a large piece of butcher paper, trace the body of a volunteer.
- Ask the group to brainstorm body organs and to draw them into the outline with different colored markers. As organs are added, discuss their general significance or purpose.
- Ask participants, "When you eat a cracker, what happens to the cracker?" Discuss that crackers contain several types of ingredients. The protein in crackers is made of amino acids.
- Print the sentence, "Polly ate a cracker" on a large strip of paper. Place the sentence in the mouth of the traced body and guide it down to the stomach and intestine where the sentence is broken down into words or proteins. The words are then taken to the liver where they are broken down into letters representing amino acids. The amino acids are then rearranged to form different proteins. The excess letters or amino acids that could not be processed by the liver, then travel into the blood along with the new proteins. Excess amino acids travel in the blood until they reach the brain. This helps to explain how food intake eventually affects the brain.

Suggestion: Follow this activity with "The Naked Egg."

TEACHING AID

The Naked Egg

Objective: To illustrate how elevated Phe levels may demyelinate white matter and affect neuron function.

Materials Needed

- Jars
- Eggs
- Vinegar
- Wire
- Beads

How It's Done

Precede this activity with "I Ate A Cracker" to demonstrate how Phe from food arrives at the brain.

- Approximately 2-3 days prior to the activity, prepare an uncooked, whole egg in a clear jar and cover it with clear vinegar. Allow the egg to sit while the acetic acid of the vinegar slowly dissolves the eggshell.
- On the day of the activity, provide an uncooked, whole egg and clear jar to each participant. As they cover their eggs with vinegar, participants will see the bubbles immediately forming on the surface of the eggshell. This provides a basis for your discussion of white matter degeneration due to high Phe levels in the blood, which travels to the brain.
- Begin a discussion of white versus gray matter in the brain. Gray matter may be described as the control center and white matter equates to insulation for rapid messaging.
- Pieces of wire may provide a helpful illustration of neurons without insulation. Wires with beads on them represent insulated neurons. Explain that messages can jump between beads, rather than slowly traveling along each point of the wire, stressing the point that insulation is necessary for quick messaging.
- Allow participants to take the jars home and observe the eggshell degeneration over the next several days.

TEACHING AID

Weighing &Measuring

Objective: To use a standard system of measurement to quantify metabolic formula; to evaluate accuracy and technique for measuring and mixing formula; to assess participants knowledge of their diet prescription.

How It's Done

This activity will show participants that volume and weight aren't always equal.

- ❖ You will need a gram scale, measuring cups, measuring spoons, a blender and reference charts (see handout).
- ❖ Using measuring tools have each participant weigh their formula and compare with their prescription . **The gram scale is the gold standard!**
- ❖ Discuss differences in various methods of mixing metabolic formulas, the varying gram amounts, the addition of fluid and different concentrations. All of these factors will affect the accuracy of the prescription.

Materials Needed

- ❖ Gram scale
- ❖ Measuring cups
- ❖ Measuring spoons
- ❖ Blender or shaker cup

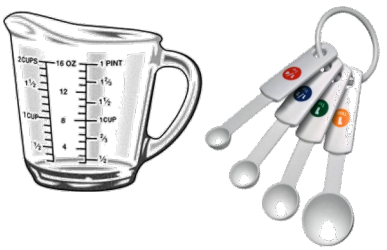
Handout:

Weighing & Measuring References

Weighing & Measuring References

1 Gram of Protein = 50 mg of Phe

Household Measures



Some household items are great measuring tools!

Measurement	Similar In Size
1 Tbsp	2 Dice
½ c	Light Bulb
1 c	Baseball

Remember the Rule of Thumb

Measurement	Similar In Size
1 Tablespoon	2 Thumbs
1 teaspoon	Tip of Thumb*
1 Cup	Fist
½ Cup	Cupped Palm

* The tip of your thumb begins at the base of your fingernail.

Food weight and size will vary. Always use a gram scale when available.

Key					
c	Cup	L	Liter	Pt	Pint
fl oz	Fluid ounce	lb	Pound	Tbsp	Tablespoon
gal	Gallon	mg	Milligram	tsp	Teaspoon
g	Gram	ml	Milliliter	Qt	Quart
Kg	Kilogram	oz	Ounce		

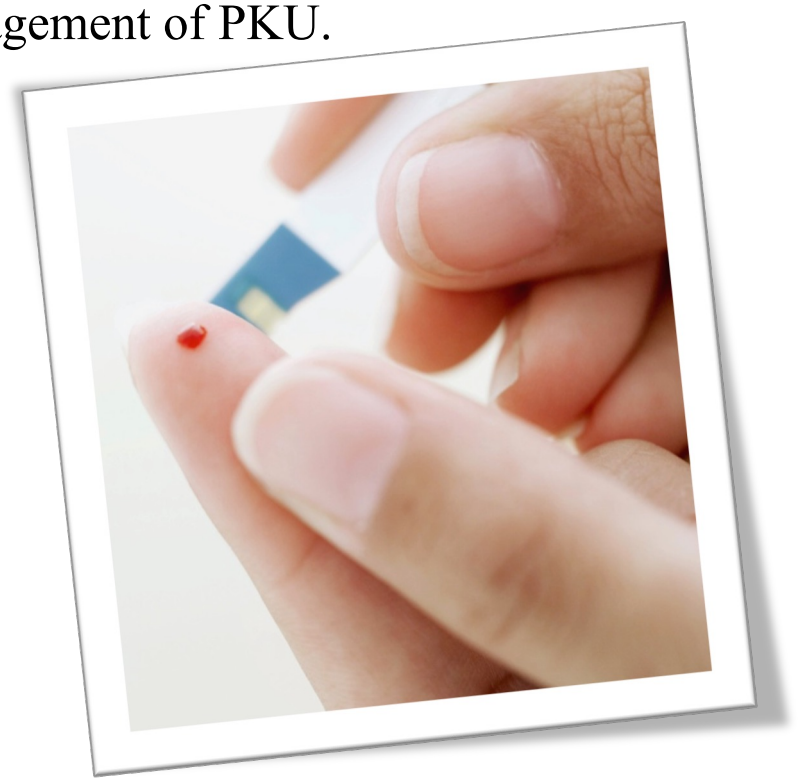
Household Measurements	
1 c	16 Tbsp
¾ c	12 Tbsp
2/3 c	10 Tbsp + 2 tsp
½ c	8 Tbsp
3/8 c	6 Tbsp
1/3 c	5 Tbsp + 1 tsp
¼ c	4 Tbsp
1/6 c	2 Tbsp + 2 tsp
1/8 c	2 Tbsp
1/16 c	1 Tbsp
1 Pt	2 c
1 Qt	2 Pt
1 Tbsp	3 tsp
1 c	48 tsp

Metric vs. US Measurements	
1 ml	1/5 tsp
5 ml	1 tsp
15 ml	1 Tbsp
30 ml	1 fl oz
50 ml	1/5 c
100 ml	3.4 fl oz
240 ml	1 c
470 ml	1 Pt (2 c)
950 ml	1 Qt (4 c)
1 L	34 fl oz 4.2 c 2.1 Pt 1.06 Qt .26 gal
3.8 L	1 gal (4 Qt)
1 g	.035 oz
28 g	1 oz
100 g	3.5 oz
454 g	1 lb
500 g	1.1 lb
1 Kg	2.205 lb 35 oz

TEACHING AID

Blood Drawing

Objective: To provide instruction on self blood drawing for adolescents and to demonstrate the importance of regimen tasks in the management of PKU.



How It's Done

Use the handout "Blood Drawing"

- Paint your face white. (Talcum powder will do.) Create some blood (lipstick) dripping from the corners of your mouth. Don black pants and a white shirt. Put on your black cape and put in your fangs—you are now Dracula and ready to instruct on self-blood drawing!
- *Actually, our preferred method is to solicit the help of our favorite phlebotomist and ask him/her to do this.*
- Provide your patient with fangs. (Stock up supply during Halloween period.)
- Teach and practice self blood draws.
- Have participants collect their own blood sample for clinic visit labs.
- This is great to do around Halloween.

Materials Needed

- Lancets
- Collection tubes
- Tube labels
- Order sheets
- Vampire teeth
- Dracula cape
- White face paint
- Fake blood

Blood Drawing

1. Selecting the puncture site

- Finger is the most commonly used puncture site
- Avoid previous puncture sites
- Avoid very tip of finger

2. Warming the puncture site

- Warming finger prior to blood collection can increase blood flow and make the draw easier
- Easiest way is to place the hand in warm water or wrap a hot moist towel around the hand

3. Cleansing the puncture site

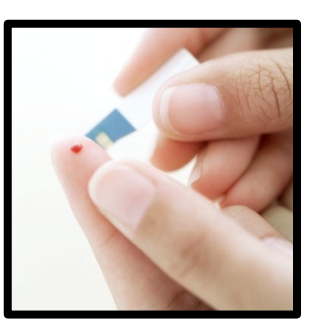
- Preferred antiseptic is alcohol or betadine
- Antiseptic must remain in contact with the skin for at least one minute
- Let the skin air dry

4. Puncture technique

- Puncture the skin in one continuous deliberate motion using the appropriate lancet
- Direction should be perpendicular to “finger prints”
- The first drop of blood should be wiped away with a sterile swab

5. Collecting the sample

- Apply moderate pressure to the puncture site
- Avoid squeezing with force in that this will cause the red cells to break open
- Collect blood in a BD microtainer serum separator tube or on a filter card
- Label the sample with your name and date of collection



TEACHING AID

Risky Business

Objective: To evaluate and encourage discussion of various risk taking behaviors as they pertain to health and PKU.

How It's Done

Risk means different things to different people

- Ask the group to define the term "risk."
- Does everyone agree on what is or is not considered "risky" behavior?
- Would their parents, teachers and peers agree?
- Distribute the survey, "Risky Business." Allow each participant to independently complete their own survey, ranking certain behaviors as high risk, low risk, or no risk.
- Facilitate group discussion of individuals' responses and address areas of concern.

Handout Needed
Risky Business

Adapted from M. Halter and B. Fierro Lang Making Choices

Risky Business

Causes of Disease	List at least one example:
Heredity	
Environment	
Life Style	
Germs	

With regards to your inherited metabolic disease or in general, mark the box with the appropriate risk level for the following behaviors.	High Risk	Low Risk	No Risk
Smoking a pack of cigarettes a day			
Drinking formula once a day			
Eating foods high in Phe once a year			
Drinking from the same cup as someone who has a cold			
Having unprotected sex with anyone			
Eating rotten food			
Accepting an offer from a friend to share their ice cream once a week			
Living in the same house as someone with PKU			
Living in the same house as someone with HIV			
Wearing someone's pierced earrings			
Not being on a phenylalanine restricted diet			
Taking mega doses of vitamins			
Taking another person's prescription medicine			
Drinking three glasses of orange juice a day			
Not wearing your seat belt while driving			

Adapted from M. Halter and B. Fierro Lang Making Choices (see resources)

TEACHING AID

Management Contracts

Objective: To encourage ownership of and consequences for personal decisions; to assist in individual and family management of compliance with treatment.

How It's Done

Each contract will have to be written specifically for the patient

- ❖ See examples of management contracts in the handout section. Examples are intended as guidelines and should be modified to meet individual needs.

Handouts Needed
Guidelines for Negotiation
Sample Contracts

Adapted from Michael A. Rapoff Adherence to Pediatric Medical Regimens

Guidelines for Negotiation

As adolescents learn to manage chronic disease, they must also learn accountability for their choices. Constructive negotiation involving the adolescent, his/her parent(s) and the clinic staff is an essential part of this process. Management contracts provide a way to record and revisit what has been worked out in negotiations. They also establish both positive and negative known consequences, which removes the decision of punishment from the parents and places the choice and control on the adolescent.

Tips for Negotiation:

- Choose a convenient, uninterrupted time for negotiation. If necessary, establish a weekly appointment for family meetings.
- Avoid negotiations following a big "blow-up." Postpone the negotiation until all parties are more willing to listen.
- Select a facilitator for the meeting. This person is responsible for ensuring that everyone has had a chance to speak and listen and that the tone remains positive.
- Phrase constructive criticism as "I Messages." For instance, rather than placing blame by saying, "You have been lazy and irresponsible about your diet, " try, "I am concerned that your phe levels have increased. I would like to see you be consistent in following your diet prescription."
- Communicate constructively. This is not a power struggle or an attack. If an individual continues to speak in a negative and critical way, he/she may be asked to leave the meeting to "cool off." Individuals will learn that decisions that affect them may be made without their input if they continue to be disruptive.
- If you identify a problem, also offer a solution. These meetings are meant to be constructive, not destructive.
- When all parties have had an opportunity to raise and discuss solutions, they will vote on a plan. Make sure the plan includes a specific way to monitor efficacy and establishes a specific time limit in which to evaluate the plan.
  - Develop a written contract in which everyone has input and is allowed to sign.
  - Contracts should focus on specific, measurable goals.
  - Specify positive and negative consequences which will occur as a result of the individuals choices. Parties must establish ahead of time which privileges may be added, suspended or removed and for what length of time.
  - Clearly state how the behavior will be monitored and who will be responsible for assessing this.
  - At the end of the contract time limit, parties will meet to review and modify the contract if necessary.
- See sample contracts: Diet, Kuvan , High Phe Levels, and Termination of Diet

Sample Contracts

Diet Agreement

I, _____, hereby make a commitment to follow my diet prescription. I understand the factual information regarding PKU and long-term consequences of being off diet, which have been presented to me. I am aware that following diet includes the following:

- ❑ Knowing my diet prescription
- ❑ Weighing my own formula
- ❑ Mixing my own formula
- ❑ Drinking the formula according to my diet prescription, until completed
- ❑ Counting (and recording) mgs of Phe
- ❑ Keeping my Phe levels within treatment range (2-6mg/dl)
- ❑ Having blood drawn monthly for Phe/tyrosine levels
- ❑ Coming to clinic visits twice per year
- ❑ Knowing the risks of maternal PKU
- ❑ Maintaining accurate diet records

I understand that if I choose <u>not</u> to follow these guidelines, I will be at risk for neurological disease. I am aware of the consequences and have listed them below:

_____

_____

_____

Signatures: Patient_____ Date_____

Parent(s)_____

Clinic Representative_____

Sample Contracts

Kuvan Agreement

I, _____, hereby make a commitment to follow my diet prescription while on this Kuvan trial. I understand the Phe/tyrosine levels for the first four weeks will provided after the first month of the trial. I am aware that participating in the Kuvan trial requires:

- ❑ Not making any changes to food intake.
- ❑ Taking Kuvan daily at the same time as food.
- ❑ Having blood drawn weekly for Phe/tyrosine levels
- ❑ Maintaining accurate diet records

I understand that if I choose not to follow these guidelines, I will withdrawn from the trial.

Signatures: Patient_____ Date_____

Parent(s)_____

Clinic Representative_____

Sample Contracts

High Phe Levels Agreement

I, _____, agree that I need to bring my Phe levels down to an acceptable range (2-6 mg/dl). In order to accomplish this, I understand that I must regulate my diet and drink my formula as prescribed. I also understand that if I choose not to follow my diet, there will be the following complications: (ex: I will be admitted to the hospital. This means that I will be temporarily removed from school and will have to leave my job and sports.)

Signatures: Patient_____ Date_____

Parent(s)_____

Clinic Representative_____

Termination of Diet Agreement

I, _____ recognize that the decision to discontinue diet is against medical advice. I understand that this decision places me at risk for neurological disease, B_{12} deficiency and folate deficiency. I have been encouraged to maintain my relationship with the clinic in spite of this decision. The clinic will continue to maintain an open door policy and encourage ongoing communication.

Signatures: Patient_____ Date_____

Parent(s)_____

Clinic Representative_____

As a female patient, in addition to the aforementioned risks, I am aware of the risks and issues of maternal PKU.

Signatures: Patient_____ Date_____

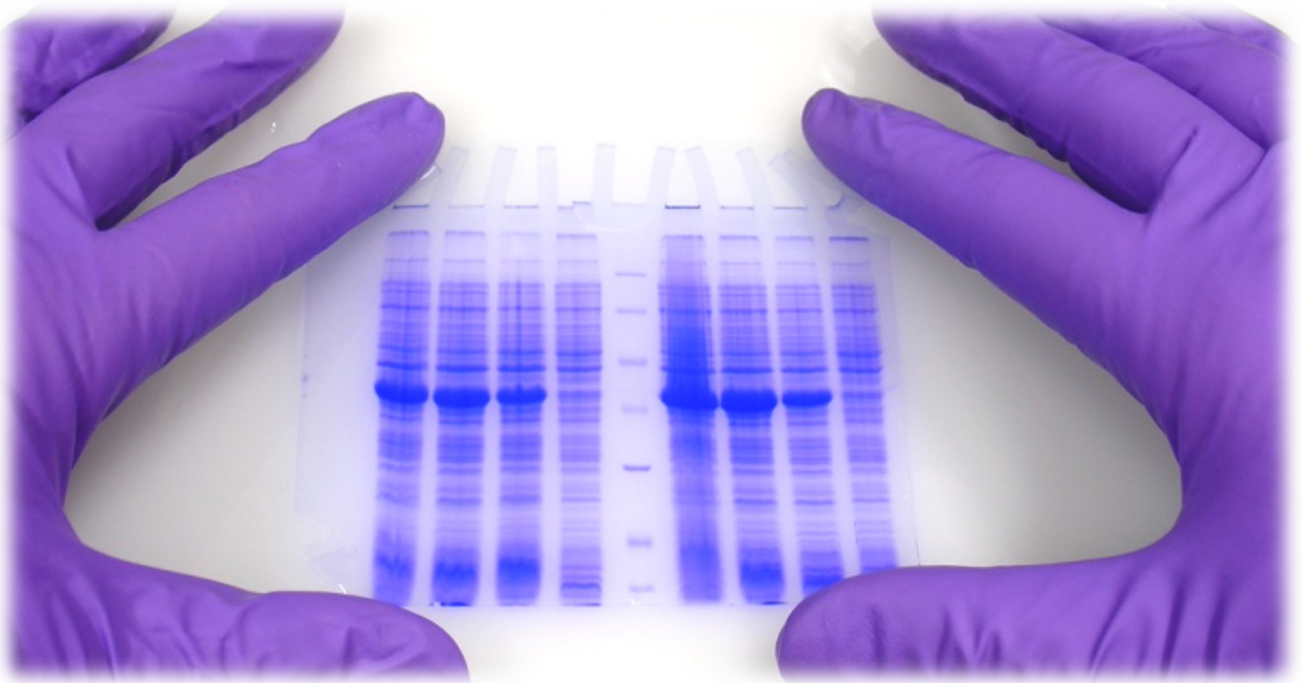
Parent(s)_____

Clinic Representative_____

TEACHING AID

DNA Extraction

Objective: To educate the participants about DNA and its function.



How It's Done

- See DNA Extraction Handout for specific instructions of how to do a DNA extraction.
- Review with the participants the concept of DNA, genes, and chromosomes. Talk about their functions in the body. The Unraveling Your Genes Handout may be used to help lead discussion.
- Use the Exotic Tea Recipe Handout to illustrate the concept of codons and various types of mutations (point mutations, deletions, insertions).

Materials Needed

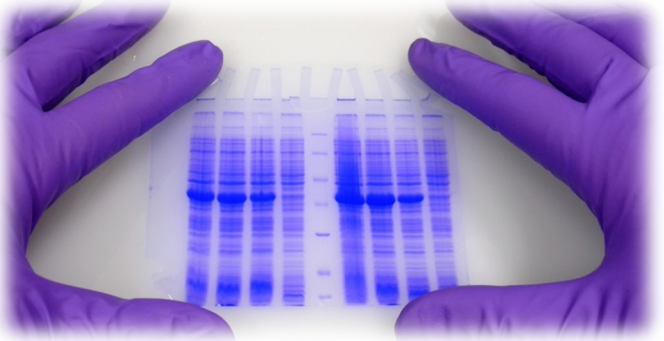
- Graduated cylinder
- Water
- Plastic cups
- Plastic spoons
- Metal sieve
- Stirring rod
- Liquid soap
- Wheat germ
- Meat tenderizer
- Baking soda
- Rubbing alcohol

Handouts:

DNA Extraction
Unraveling Your Genes
Exotic Tea Recipe

Adapted from genome.gov National Human Genome Human Research Institute http://www.genome.gov/11511420

DNA EXTRACTION



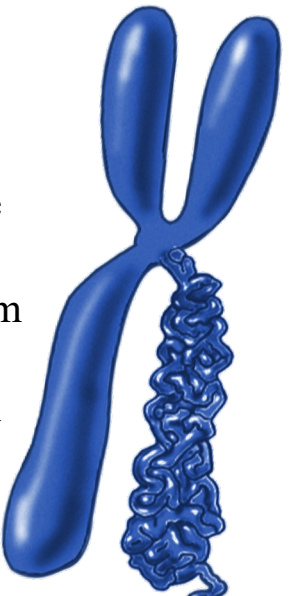
How It's Done

1. Using the graduated cylinder, measure out 100 ml of water and pour it into the plastic cup.
2. Add 1 large spoonful of wheat germ to the water and mix using a plastic spoon.
3. Add one pump of liquid soap, stir for 1 minute.
4. Add 1 small spoonful of meat tenderizer and 2 small spoonfuls of baking soda. Stir to mix for 1 minute.
5. Strain the wheat germ solution by putting a metal sieve over a plastic cup and pouring the wheat germ solution into the sieve.
6. Once the wheat germ has settled, remove the sieve and transfer three or four droppers full of the wheat germ liquid in the cup to a tube.
7. Dribble alcohol down the side of the tube, adding an amount of alcohol equal to the wheat germ liquid. Try not to mix the two layers. Let the tube sit for approximately. You will see large and small bubbles appear at the interface between the two layers. You will also see the formation of white, stringy material. This is the DNA, it does not dissolve in alcohol.
8. Carefully swirl a rod at the interface of the two layers using small circles to spool or wrap the DNA around the rod. If you keep swirling and are careful not to mix the two layers, you might be able to pull out a big wad of DNA. The wad of DNA that you have collected on the rod is composed of millions of DNA strands. When many of these strands are swirled together they make a large sticky, slimy glob. Because this DNA sample has been purified very quickly, there are proteins and long chain sugars mixed in with the DNA. Scientists use a similar procedure to produce highly purified DNA. You can touch your DNA sample to see how it feels.

Unraveling Your Genes

CHROMOSOMES

They are important. They carry the genetic information we inherit from our parents. Every cell in our body has 46 chromosomes: 23 inherited from Mom and 23 inherited from Dad. Each chromosome is made up of coiled, string like material. If you grab the end of the string like material and unravel the chromosome, you will find that the string like material is actually…

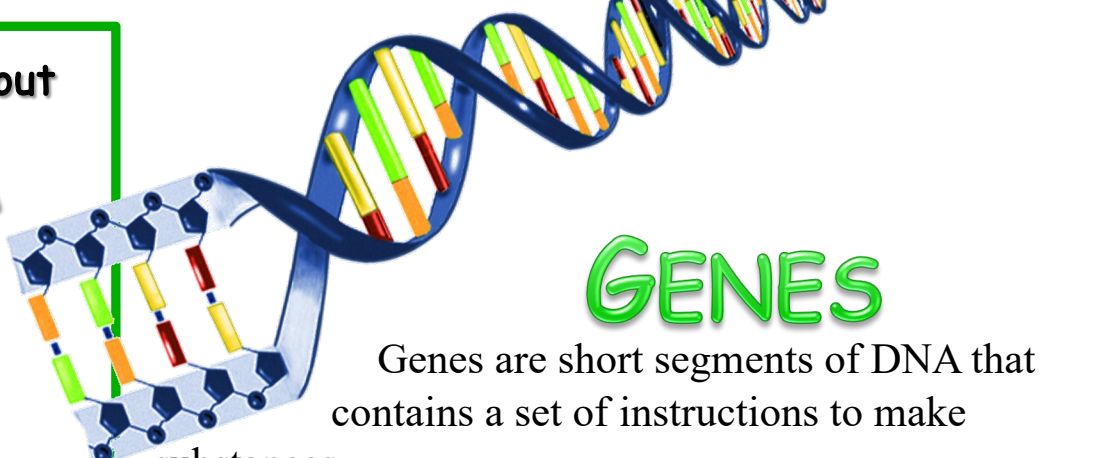


"Chromosome Inside Nucleus" *Image Gallery*. National Institute of General Medical Sciences.

DNA

Deoxyribo**N**ucleic **A**cid

DNA is a chemical molecule made from 4 different "bases" or chemical letters. Like the alphabet, these chemical letters can spell out instructions to make things.



Fun Facts About DNA

- 5 million strands of DNA can fit into the eye of a needle.
- 1 cell = 6 feet of DNA
- 5 trillion cells in the body = 5.7 billion miles of DNA
- Your DNA could stretch to the sun and back 30 times, or circle the earth 228,000×

GENES

Genes are short segments of DNA that contains a set of instructions to make substances our body needs to function. In general, one gene contains one set of instructions to make one protein our substance our body needs. There are hundreds of genes on each chromosome. Together, they genes combine to make our own special blueprint or recipe to make our body.

Exotic Tea Recipe

Genes use chemical letters to write out its instructions or recipes. It does this using three letter words. Let's look at a gene that is a recipe for a very exotic, topical drink that you might want to serve at your graduation from high school. The recipe is so complicated, in print it would cover a whole page, so we are just going to look at the very last sentence.

Exotic Blackberry Honey Tea

…add one cup tea and add ice and mix end

Possible changes in the recipe are noted below in **red**:

…add one kup tea and add ice and mix end

…add one cup sea and add ice and mix end

…add one cup tea end

…ado nec upt eaa nda ddi cea ndm ixe nd

…add one ciu pte aan dad dic ean dmi xen d

Draw a square around the changes that are due to a substitution of a letter.
Draw a circle around the changes that are due to a deletion of letters or words.
Draw a triangle around the changes that are due to a insertion of a letter.

Discuss how these changes effect:

- Your ability to make the tea?
- The flavor of the tea?
- Whether or not you would serve the tea at your party?

TEACHING AID

Mutations

Objective: To introduce the concept of genetic mutations and how they can impact the gene and gene product.

How It's Done

Introduce the activity by reviewing the concept of a gene and how a genes functions as a set of instructions

- Write the sentence "Wash the dishes after dinner." on a sheet of paper or on a whiteboard. Discuss with participants that the sentence provides instructions; hence in some ways is analogous to a gene.
- Erase or mark out the words "after dinner" in the sentence. Discuss how the change might impact the sentence's instructions. (For example, the dishes will still be washed; however, they may not be washed in a timely manner. This may result in an accumulation of dirty dishes over time.) Relate the change in the sentence to a change in a gene. Introduce the concept of a deletion being a specific type of mutation or change in a gene. Discuss how deletions in a gene can result in a gene product that works but at a reduced rate. Explore how varying size and placement of deletions might have varying effects. Use the sentence "Wash the dishes after dinner." to illustrate this. (For example, discuss how deletions of single letters - such as the "es" in "dishes" - would impact the sentence and work done).
- Put the words "after dinner" back in the sentence. Change the letter "w" in the word "wash" to the letter "b". The sentence now reads "Bash the dishes after dinner. " Discuss how this impacts the sentence and the work done. Relate this back to genes and gene products. Introduce the concept of point mutations in genes. Discuss the impact of other possible point mutations. (For example, replace the "e" in the word "dinner" with an "a". In this case, it is likely that the person reading or hearing the sentence would still understand the meaning of the sentence. This being the case, the sentence retains good functionality.)
- Finally, talk about what happens if the sentence is unchanged but water is not available. Determine if the work will get done. Compare this to a loss of cofactor .

Materials Needed
- White board
- Markers

What does this message mean to you?

_____

_____

_____

Deletions:

Wash the dishes.

What does this message mean to you? How does it differ from the original statement ?

_____

_____

_____

Wash the dish after dinner.

What does this message mean to you? How does it differ from the original statement ?

_____

_____

_____

Point Mutations:

Bash the dishes after dinner.

What does this message mean to you? How does it differ from the original statement ?

_____

_____

_____

Wash the dishes after dinnar.

What does this message mean to you? How does it differ from the original statement ?

TEACHING AID

GO! Speed Racers

Objective: To reinforce the concept that a gene is a set of instructions; altered instructions lead to altered gene products with variable functionality.

Lime Racer

Nitro Muscle

Blue Bullet

Materials Needed

- 3 Lego© racing vehicles
- 3 sets of instructions

How It's Done

Have participants work in groups if there are more than three.

- Start with 3 disassembled Lego© racing vehicles.
- Provide each groups with parts and a set of instructions for assembling their vehicles. Note that participants must follow instructions step by step.
- Supply Group One with an unaltered set of instructions (these are provided by Lego©).

  This car, once assembled, represents a functional enzyme.
- Supply Group Two with a set of instructions that have been altered. Delete the steps that instruct how to attach the lights, muffler, seats, or other non-essential parts of the vehicle. You will have to create these flawed instructions.

  This car, once assembled, represents an enzyme that has decreased functionality, but still has some residual activity (i.e. non-PKU HPA)
- Supply Group Three with a second set of instructions that have been altered. Delete the steps that instruct how to attach the wheels or motor

  This car, once assembled, represents an enzyme that is non- functional; has no residual activity (i.e. PKU)
- Encourage discussion of how altered instructions (altered genes) lead to less or non functioning end products, such as the race car without wheels or an engine.
- For added effect, have the groups race their vehicles.

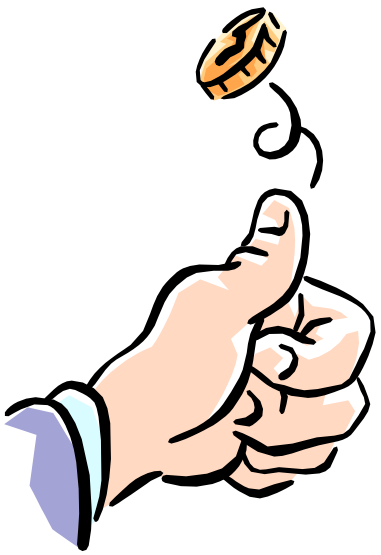
Kuvan Discussion Option:

- Talk about what would happen if the instructions to add the hood latch were flawed or missing: Discuss how the car shape might change with the hood up, blocking the windshield.
- Discuss how a mechanic could fix the hood latch, and return the hood to its proper position. Relate the mechanic to Kuvan as it's role as a Chaperone.

TEACHING AID

Recessive Traits

Objective: To explore the concepts of autosomal recessive traits and random chance.



Materials Needed

- Coins (2 per team)

Handout:

Recessive Traits Portrait

How It's Done

Begin with a discussion of Autosomal Recessive Inheritance.

Participants must understand that each individual has two copies of autosomal genes, one from each parent. They must also understand that recessive traits require that a recessive form of the gene be passed from both parents. Dominant traits, by contrast, require that only one dominant form of the gene be present.

- Provided each participant with two coins and a copy of the handout, "Recessive Traits."
- Explain that parents can not choose which genes they pass down to their children. Discuss how this occurs by random chance, much like the flip of a coin.
- For each trait listed on the handout, participants will flip their two coins and note their results (HEAD-HEAD, HEAD-tail, or tail-tail). Using the Recessive Traits Handout they will determine the phenotype for the trait dependent upon the genotype (HEAD-HEAD, HEAD-tail, or tail-tail) obtained. Have the participants circle the phenotype on the handout.
- Once all the traits have been determined, have the participants create a portrait based on their results. Have participants share their portraits and point out similarities and differences between the different portraits. Point out that a child who is affected with PKU or other metabolic disorder would not look different, however, their body would function different.

Recessive Traits Portrait

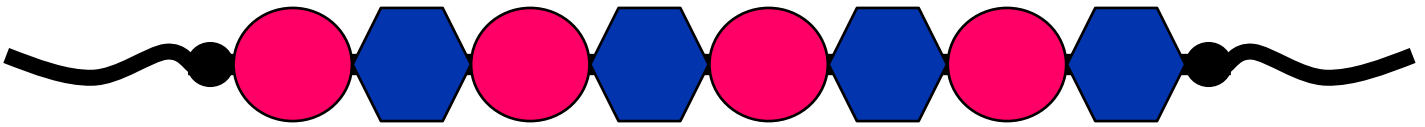
Trait	Gene		H & H	H & t	t & t
Eye color	H = brown	t = blue	Brown	Brown	Blue
Hair Color	H = brown	t = blonde	Brown	Brown	Blonde
Hair Texture	H = curly	t = straight	Curly	Curly	Straight
Eyebrows	H = bushy	t = thin	Bushy	Bushy	Thin
Eyelashes	H = short	t = long	Short	Short	Long
Dimples	H = no dimples	t = dimples	No dimples	No dimples	Dimples
Freckles	H = freckles	t = no freckles	Freckles	Freckles	No freckles
Chin	H = round	t = pointed	Round	Round	Pointed
Mouth	H = big	t = small	Big	Big	Small
Nose	H = big	t = small	Big	Big	Small
Ears	H = big	t = small	Big	Big	Small
PKU	H = Working PAH	t = Altered PAH	No PKU	No PKU	PKU

PKU is not something you can see, but if the child has it, write PKU somewhere within the frame.

TEACHING AID

PKU Bracelet

Objective: To illustrate the role of PAH's function in metabolizing Phe.



How It's Done

- Begin with a short lesson or review of genes and the PAH enzyme. See Handout: PKU Bracelet, for some suggestions.
- **Left Hand**: Show the participants the cartoon of a functional PAH enzyme as illustrated on the handout.
- Have each participant make two bracelets representing the functional enzyme. Using yarn, have them string together the blue and pink beads, alternating the colors. Tie a knot at both ends so the beads don't slide off.
- Tie the bracelets on the participants left wrist.
- Have each participant (or instructor) put a glove on their left hand. Demonstrate that, with the glove on, their hand can still open and close like a mouth. As a result the PAH enzyme is able to work; it can break down the Phe.
- **Right Hand**: Show the participants the cartoons of the non-functional PAH enzyme as illustrated on the handout .
- Have each participant make two bracelets representing non-working PAH enzymes. Using yarn, have them string 10 pink beads together. Tie a knot at both ends. Using yarn, have them string three red then three blue beads together. Tie a knot at both ends.
- Tie both bracelet on the participants right wrist.
- Have each participant (or instructor) put a (tight) sock on their right hand. Demonstrate that, with the sock on, their hand cannot open and close like a mouth. As a result the PAH protein cannot break down the Phe.
- **Wrapping up**: Discuss the consequences of altered enzyme activity

Materials Needed

- 1 glove and 1 sock of the same color
- Scissors
- Yarn

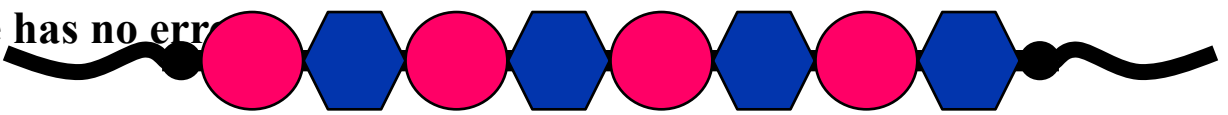
Per Participant:

- 11 blue beads
- 21 pink beads

PKU Bracelet

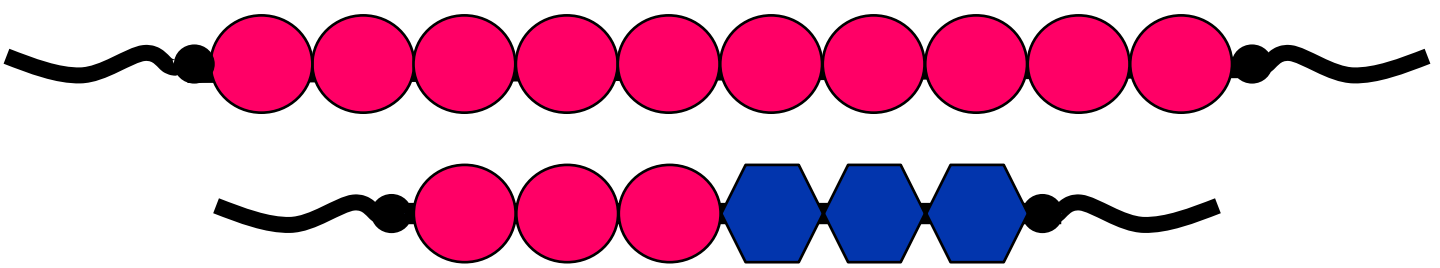
The PAH enzyme breaks down Phe in the food we eat. That is it's job. The enzyme is made from instructions carried on our genes. Each person has two copies of the PAH gene (one from Mom and one from Dad). The gene needs to be correct, without spelling errors or other mistakes, in order to make a PAH enzyme that works. If the enzyme does not work well, it can not break down Phe.

This is an example of what a PAH enzyme would look like if the PAH gene has no err



This enzyme can breakdown Phe.
Make two bracelets that looks like this protein.

This is an example of what their PAH proteins might look like if the PAH gene has errors or mistakes:



These enzymes cannot breakdown Phe because they are not correctly put together. They do not work.
Make a bracelet that looks like each of these proteins.

TEACHING AID

Random Chance

Objective: To demonstrate random chance as it applies to inheritance of autosomal recessive traits.

How It's Done

This activity is based on the famous shell game seen at carnivals, though used with four cups rather than the typical three, and 4 balls rather than one.

- Start by reviewing autosomal recessive inheritance. Use punnet squares with varying parental geneotypes to illustrate inheritance of PKU. Determine risks for a child to have PKU if both parents are homozygous normal, if one parent is homozygous normal the other a carrier, if both are carriers, if one parent has PKU and the other is homozgyous normal, and lastly if one parents has PKU and the other is a carrier. Emphasize that these are risk and that parents can not choose which genes they wish to pass down. Illustrate this by doing the "shell game".
- Take four cups, turn them upside down. Label two as "Mom" and two as "Dad" . These represent maternal and paternal chromosomes that carry the PAH gene. Label ping-pong balls: 4 balls with "PAH" to represent a PAH gene that is unaltered and four as "pah" to represent a PAH gene that is altered. Show the cups and the balls to the participant and explain what they represent.
- Play the game using two carrier parents: Place one "PAH" ping-pong ball and one "pah" ping-pong ball under the "Mom" cup . Do the same the "Dad" cup. Have participants watch you do this and write the parental genotype on paper or the whiteboard. Slides all four cups around, swapping them back and forth faster than the eye can follow. (If you can not do this fast enough, ask the participants to close their eyes.) Have participates select one gene from Mom and one from Dad. Write the genotypes selected (i.e. genotype of the offspring) on paper or a white board under the parental genotype. Discuss whether or not a person with this genotype would be affected with PKU.

  Place the genes back under the cups and repeat the activity. After about six repetitions, review all the offspring genotypes obtained and relate this back to the punnet square and calculated frequency of the various genotypes.
- Repeat the activity with a different parental geneotype, f… r parent, and/or one parent with PKU. End by reviewing that parents have no choice in which genes they donate. The process is random and out of their control.
- Repeat the activity with a different parental geneotype. End by reemphasizing that parents have no choice in which genes they donate; the process is random chance.

Materials Needed

- Four cups
- Eight ping pong balls
- Permanent marker

Handout: PKU Punnet Square

PKU Punnet Squares

	PAH	PAH
PAH		
PAH		

Risk of PKU _____

	PAH	pah
PAH		
pah		

Risk of PKU _____

	PAH	pah
PAH		
PAH		

Risk of PKU _____

	pah	pah
PAH		
pah		

Risk of PKU _____

	pah	pah
pah		
pah		

Risk of PKU _____

PAH = Functioning PAH

pah = Non-functioning PAH

PKU Punnet Squares

	PAH	PAH
PAH	**PAH/PAH**	**PAH/PAH**
PAH	**PAH/PAH**	**PAH/PAH**

Risk of PKU 0%

	PAH	pah
PAH	**PAH/PAH**	**PAH/pah**
pah	**PAH/pah**	**pah/pah**

Risk of PKU 25%

	PAH	pah
PAH	**PAH/PAH**	**PAH/pah**
PAH	**PAH/PAH**	**PAH/pah**

Risk of PKU 0%

	pah	pah
PAH	**PAH/pah**	**PAH/pah**
pah	**pah/pah**	**pah/pah**

Risk of PKU 50%

	pah	pah
pah	**pah/pah**	**pah/pah**
pah	**pah/pah**	**pah/pah**

Risk of PKU 100%

PAH = Functioning PAH

pah = Non-functioning PAH

TEACHING AID

Enzyme Bill

Objective: To illustrate the concept of an enzyme.

Lead in by asking participants what enzyme is not working properly in individuals with PKU. Have participants write the full and abbreviated name of the enzyme on the white board or on paper. Discuss how PAH functions; how it acts as a catalyst promoting the conversation Phe to Tyr. Have the participants write the reaction on the white board or on paper. Discuss that this change or reaction would not occur in the absence of PAH. To illustrate this do the following activity.

How It's Done

- Provide participants with 5 paper clips. Ask them to create a chemical reaction or change the results the paperclips being hooked together. Note that when doing this they must not use their hand. Have a contest to see who can hook the most together. Set the stage with a "Ready, set, go!"
- Ask individuals to share how many paperclips they were able to hook together. Ask why the number is zero. Discuss that the change or reaction could not occur on its own.
- Enter Enzyme Bill to the rescue. With fanfare, introduce Enzyme Bill, providing one Enzyme Bill to each participant. Have participants clip the paper clips to the dollar bill as shown in the corresponding handout. Pull the ends of the dollar bill and see the resulting joined paper clips. Discuss how Enzyme Bill worked as a catalyst for the reaction. Note he had to have a specific shape in order for this to happen. He had to be folded into the Z shape in order to work properly.
- Have the participants tear Enzyme Bill, either at the top of the bill or the side. Try the reaction again. Talk about the results. (Some tears may not impact function, others will).
- Have participants tear Enzyme Bill in half. Try the reaction again. Relate these changes back to PAH activity and PKU.
- *Optional Kuvan Discussion*: Discuss what would happen if you could restore the original shape of the dollar bill (i.e. fix the tear with tape). Relate this to Kuvan and its function as a chaperone.

Materials Needed

Per Participant:

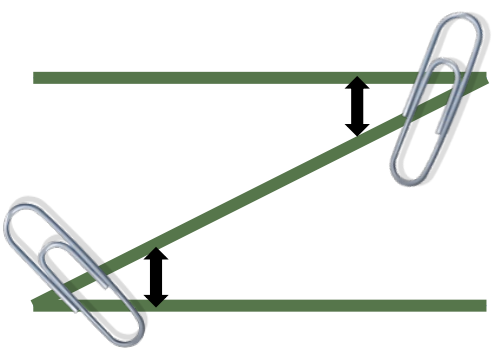
- Dollar Bill Handout
- Two Paperclips

Enzyme Bill

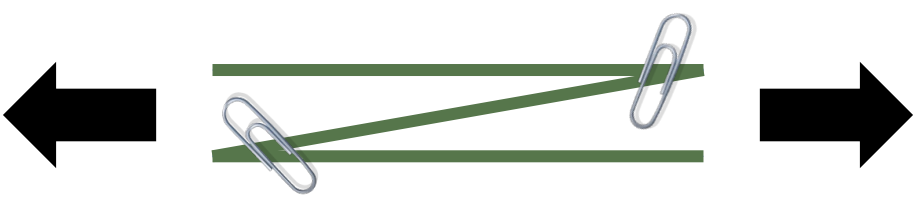
1. Begin with a dollar bill.

2. Fold the dollar bill into thirds, so it forms a "Z" shape. This is the necessary conformation of the enzyme.

1. Attach paper clips to the outer end and opposing inside fold. Do <u>not</u> clip all three folds together.



2. Grab the ends of each side of the dollar bill. In one quick motion, pull ends in opposite directions. The paper clips should join together.



Enzyme Bill


FEDERAL RESERVE NOTE
THE UNITED STATES OF AMERICA
THIS NOTE IS LEGAL TENDER
FOR ALL DEBTS, PUBLIC AND PRIVATE
L 11180916 G
WASHINGTON, D.C.
H 3 12
12
L 11180916 G
12
Anna Escobedo Cabral
Treasurer of the United States.
SERIES 2003 A
John W Snow
12
Secretary of the Treasury.
WASHINGTON
ONE DOLLAR

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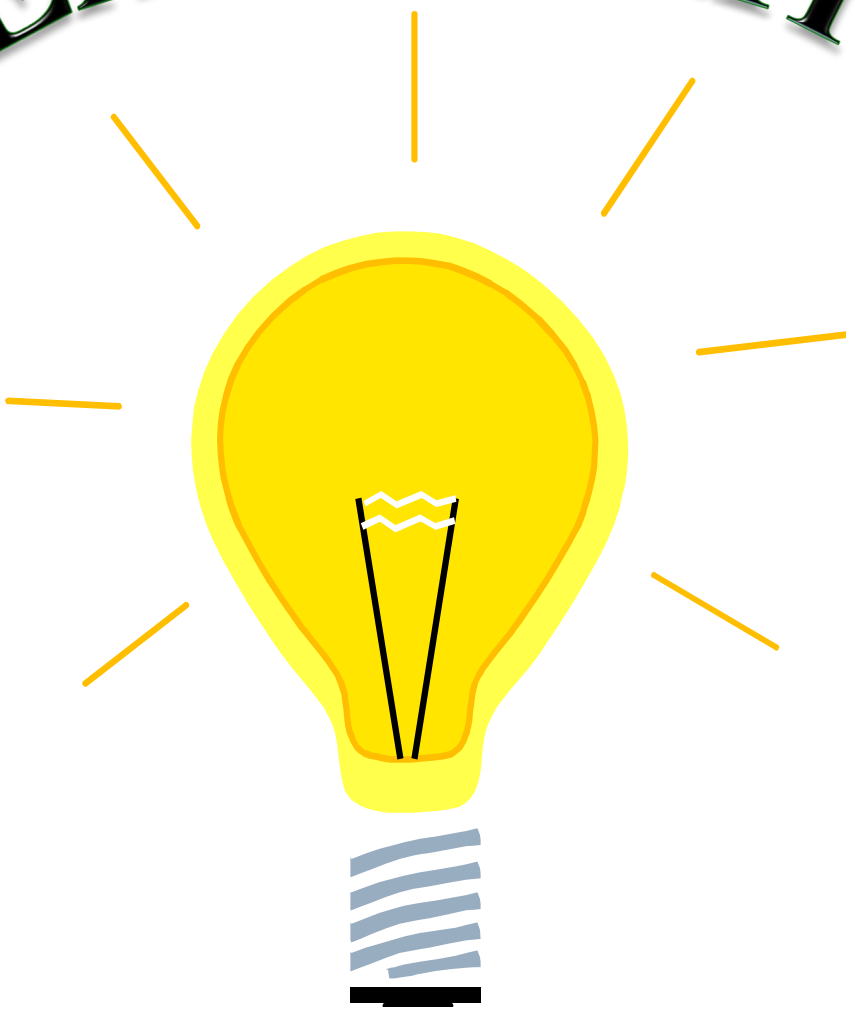
Why Can't I Eat That? Helping Kids Obey Medical Diets. J. Taylor and S. Latta, R&E Publishers, P.O. Box 2008, Saratoga, CA 95070. Available from Ross.

EAT RIGHT



STAY BRIGHT™

Guide For Hyperphenylalaninemia

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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.

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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[1]
  - Joy
  - Ambivalence
  - Fear
  - Denial
- ❑ Reproductive options[2]

Review of General Hyperphenylalaninemia Information[3]

- ❑ 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[4]
- ❑ Work
- ❑ Finances
- ❑ Impact of diet on family life

Prenatal and Newborn Care[5]

- ❑ 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

Birth to 6 Months | 6 to 12 Months | 1 to 2 Years | 2 to 7 Years | 8 Years to Adulthood | Motherhood

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

PRINCIPLES OF DIET PRESCRIPTION

PKU and Pregnancy

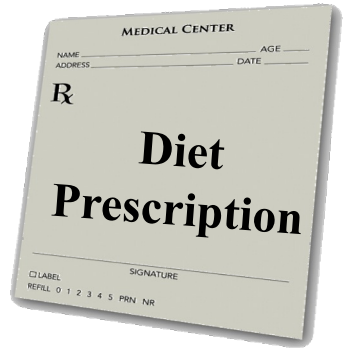
Medical Food

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

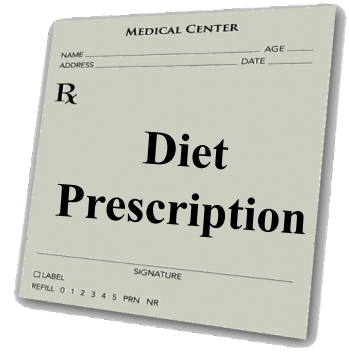
Medical Food

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.

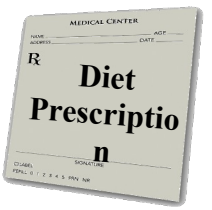
See Reference: "PKU & Pregnancy, For Women With PKU." March of Dimes.

DAILY DIET PRESCRIPTION

PKU and Pregnancy

+

=



Phe

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)
			Totals		

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 reinstitution 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
- Lophlex
- Lophlex LQ
- Periflex Advance
- XPhe Maxamum

Applied Nutrition:
www.medicalfood.com
- PhenylAde 40
- PhenylAde Drink Mix

Vitaflo:
www.vitaflousa.com
- PKU Coolers
- PKU Express

Cambrook Foods:
www.cambrookefoods.com
- CaminoPro PKU 15

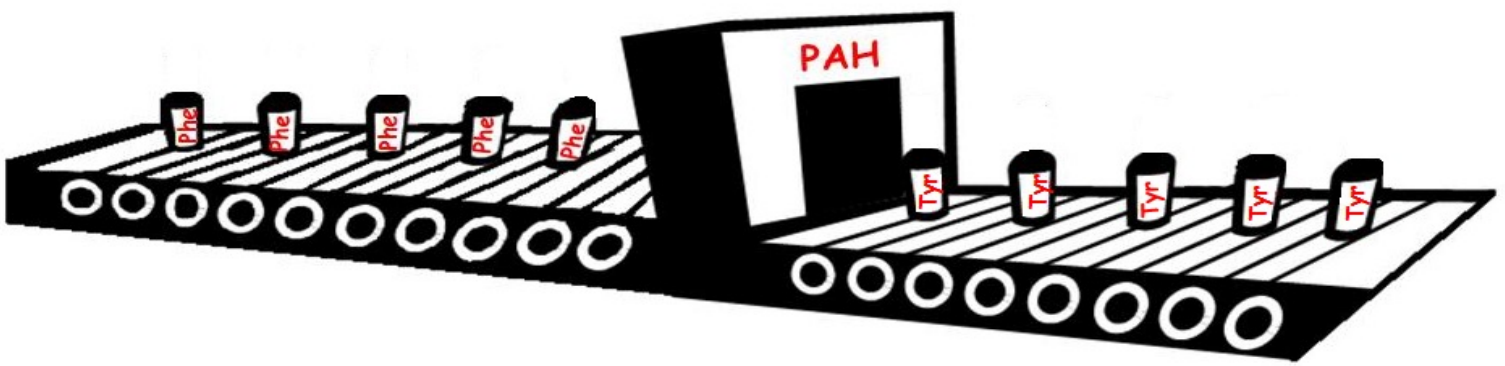
Abbott Nutrition:
www.abbottnutrition.com
- Phenex-2
- Phenyl-Free 2

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.**

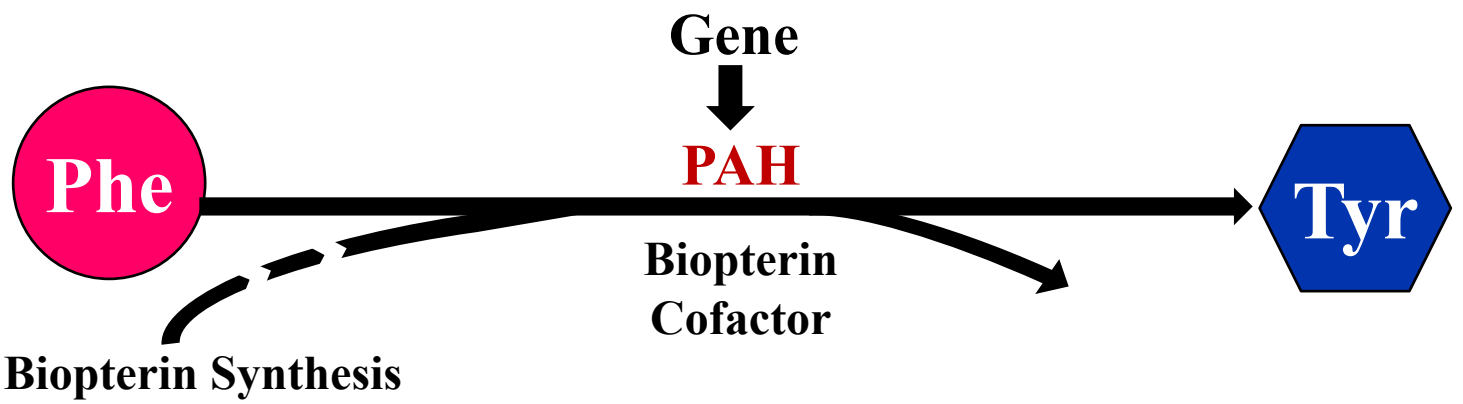
The Result: Too much **Phe** and not enough **Tyr**.

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

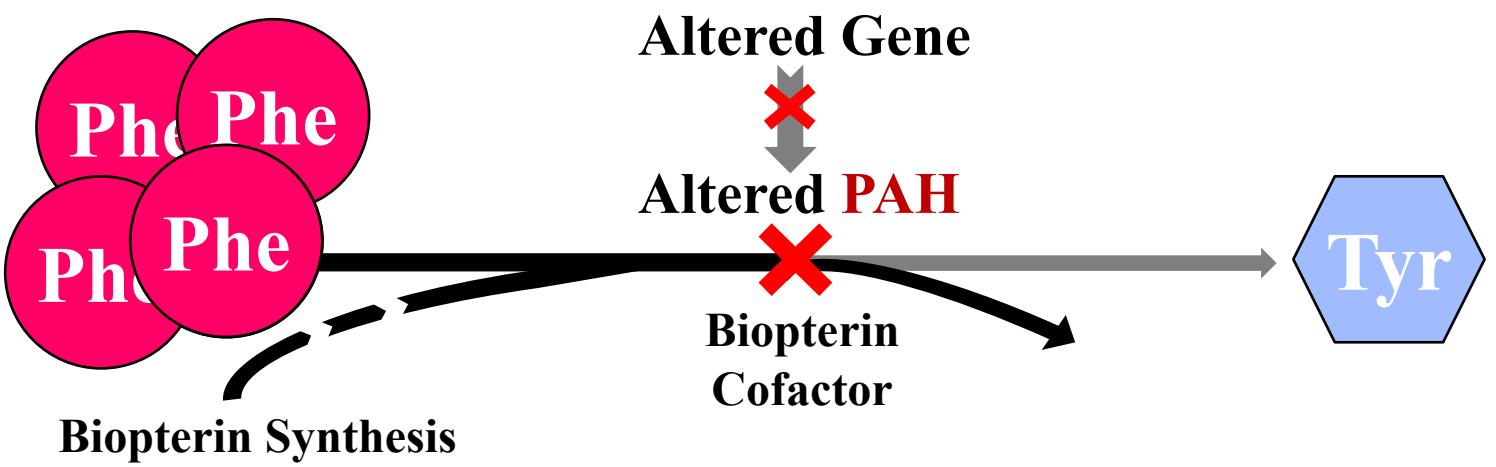
Pathways

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

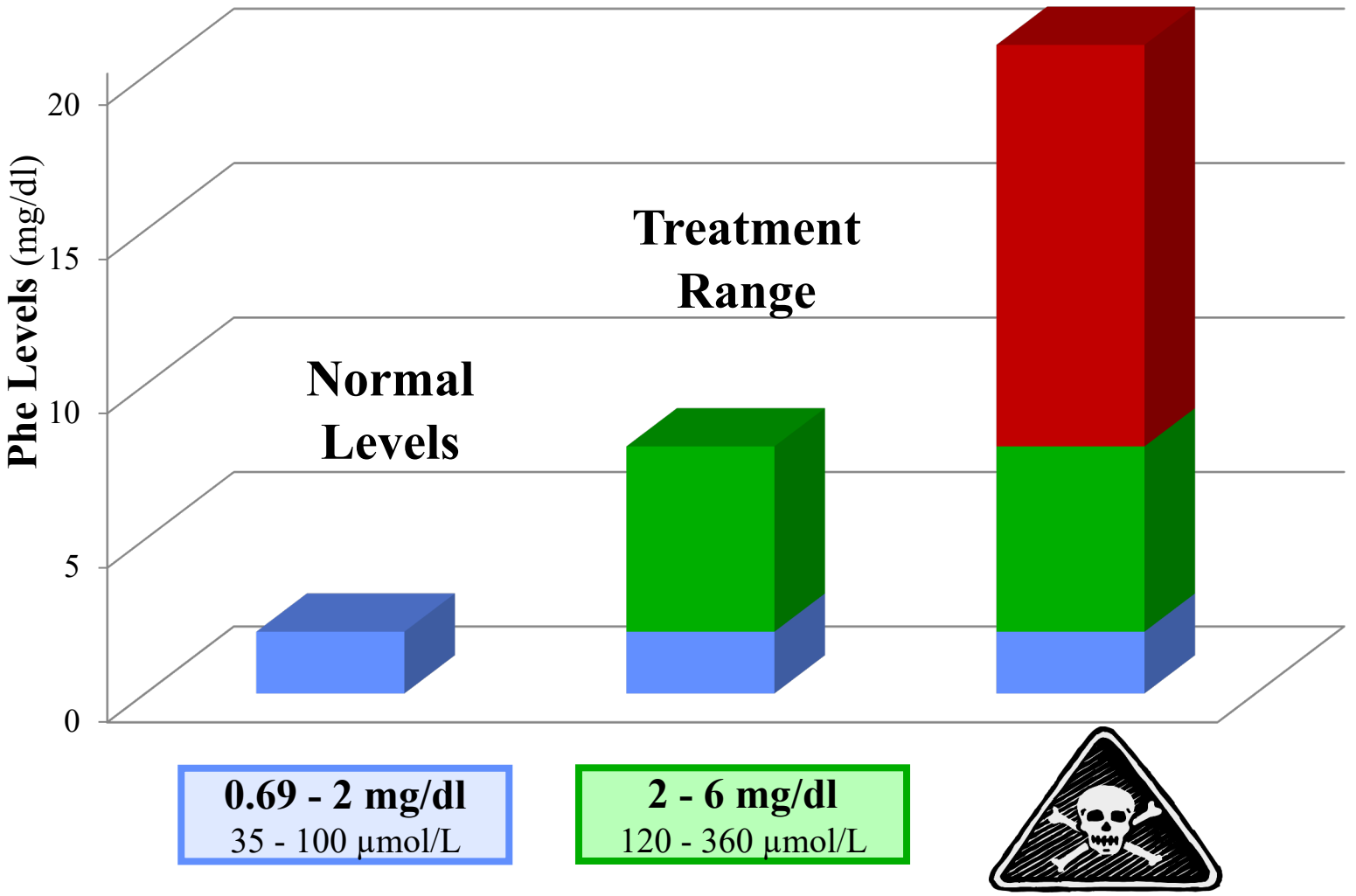
Normal Phenylalanine Pathway



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 µmol/L (2 – 6 mg/dl)

Low	Normal	Non- PKU HPA	Mild PKU	Classic PKU
< 35 µmol/L (< 0.5 mg/dl)	35 - 120 µmol/L (0.5 - 2 mg/dl)	120 - 600µmol/L (2 - 10 mg/dl)	600 - 1200 µmol/L (10 -20 mg/dl)	> 1200 µmol/L (> 20 mg/dl)

Hyperphenylalaninemia (HPA)

[------hyper-----][-----phenylalanin-----][------emia-----]

high levels of → *phenylalanine* → *in the blood*

Phenylketonuria (PKU)

[------phenylketon-----][------uria-----]

phenylketone → *in the urine*

Information taken from Nutrition Management of Patients with Inherited Metabolic Disorders by Phiyllis Acosta

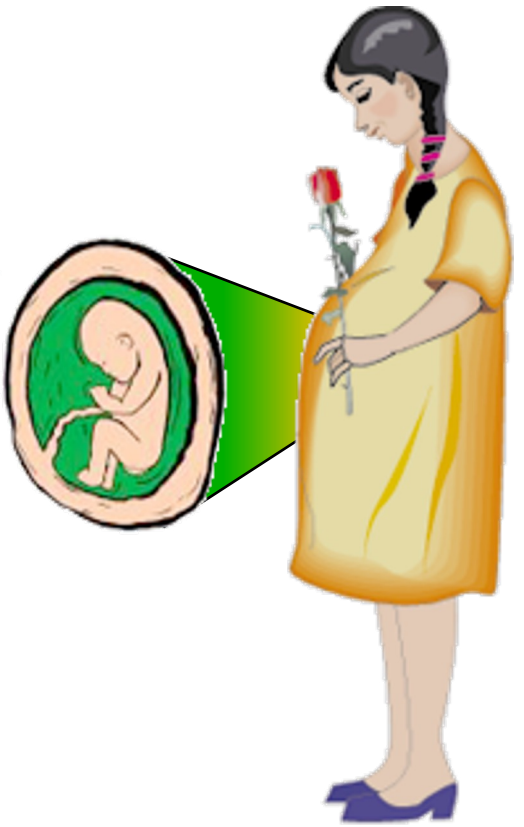
PREGNANCY TREATMENT RANGE

2.0-6.0 mg/dl

or

120 - 360 µmol/L

Phenylalanine levels are within treatment range



DANGER!!!

Phenylalanine levels greater than

6.0 mg/dl

or

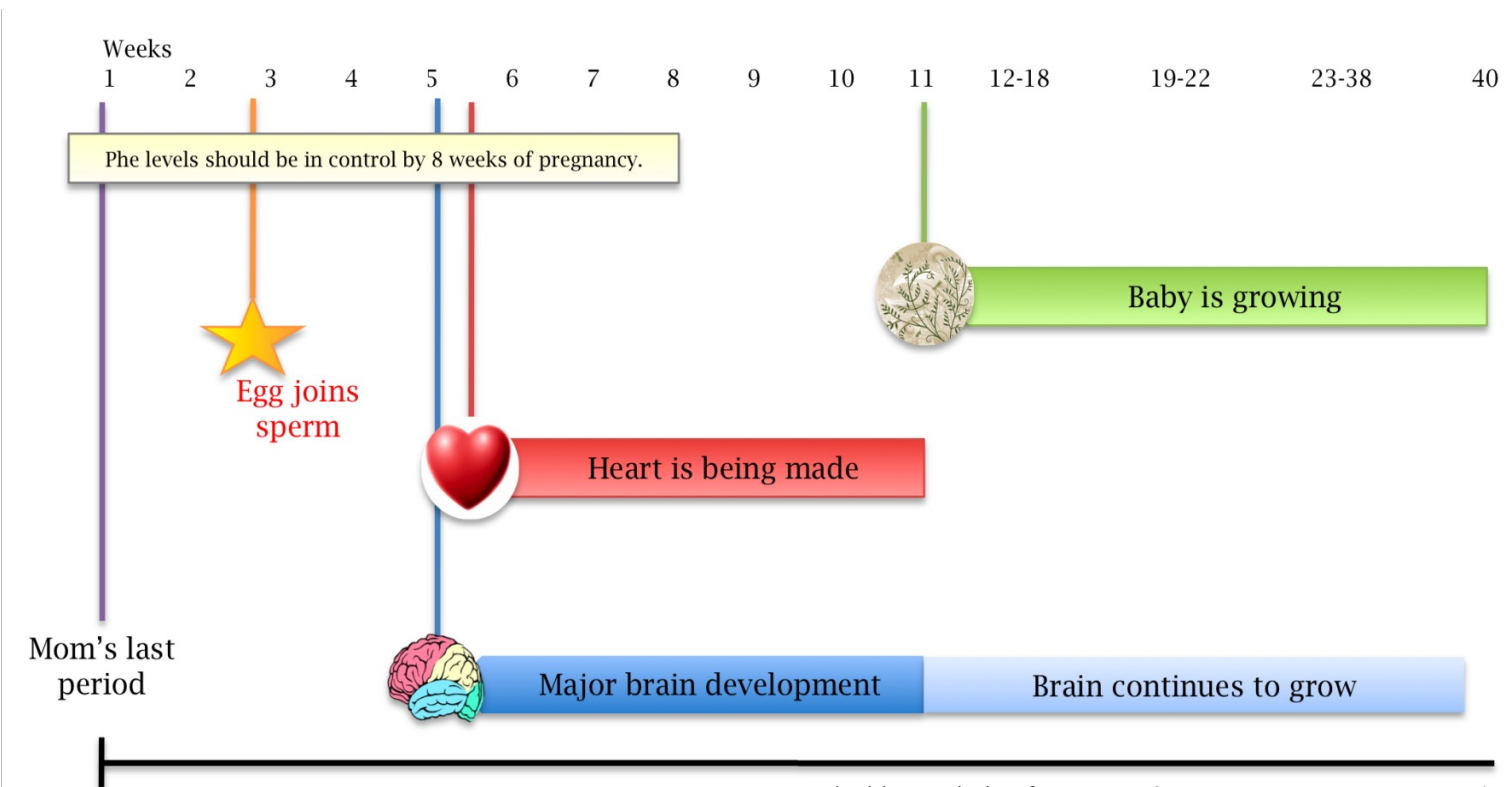
360 µmol/L

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



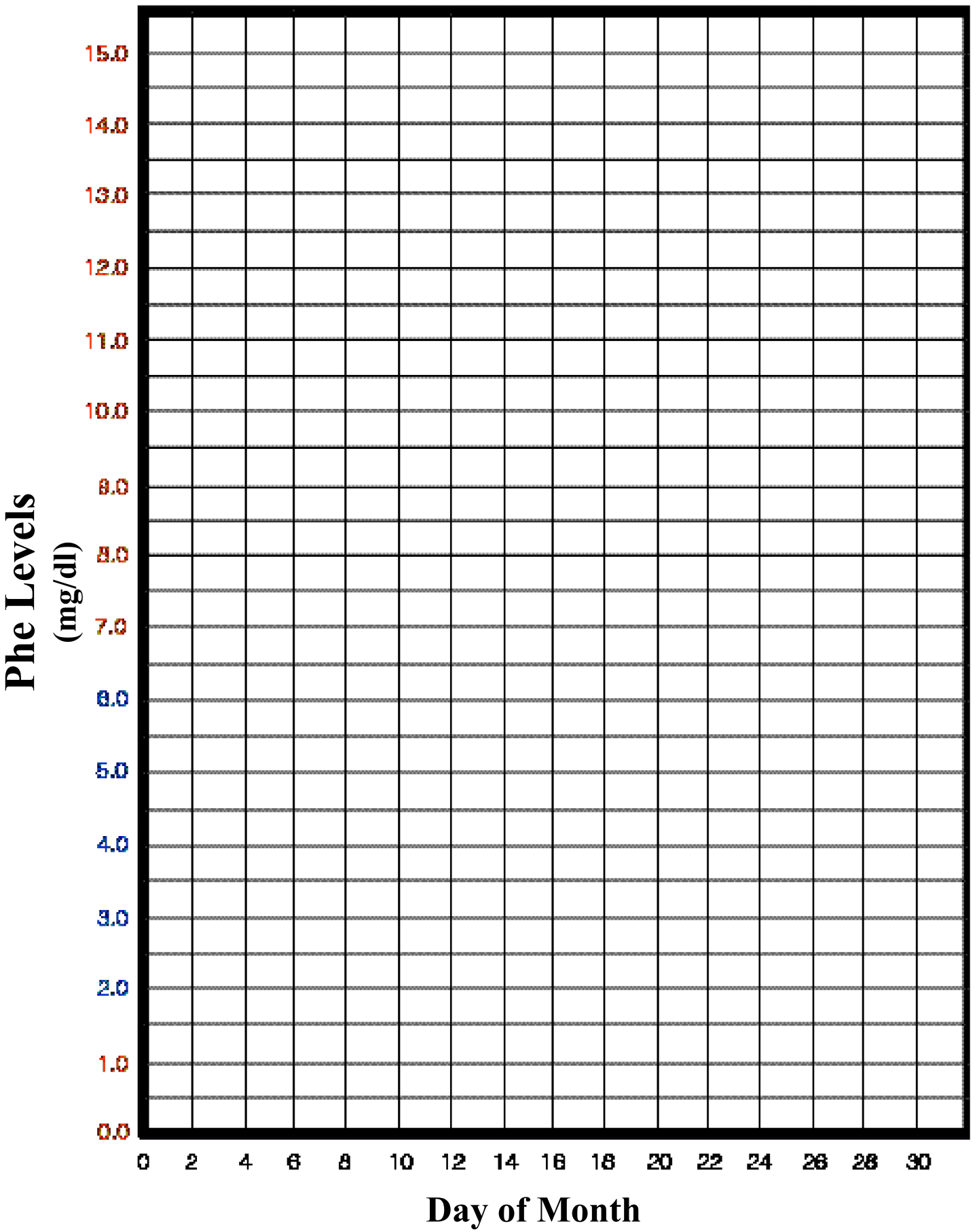
Used with permission from **PKU & Pregnancy, For Women With PKU**

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			**By 22 Weeks of Gestation:** • Fetus weighs approximately ½ pound and is 10 inches long • Eyebrows and scalp hair have begun to form • Skin is covered with vernix (a protective coating) and lanugo (fine hair)
16			
17			
18			
19			
20			
21			
22			
23			
24			

PREGNANCY CALENDAR

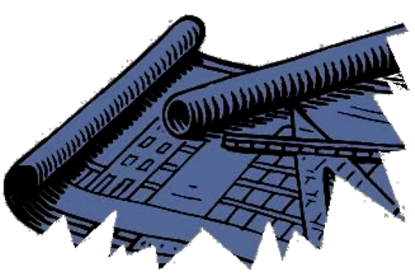
THIRD TRIMESTER

Week	Phe Level	Tyr Level	Description of Development
25			• 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. • 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. • 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.
26			
27			
28			
29			
30			
31			
32			
33			
34			
35			
36			
37			
38			
39			
40			

DNA BLUEPRINT

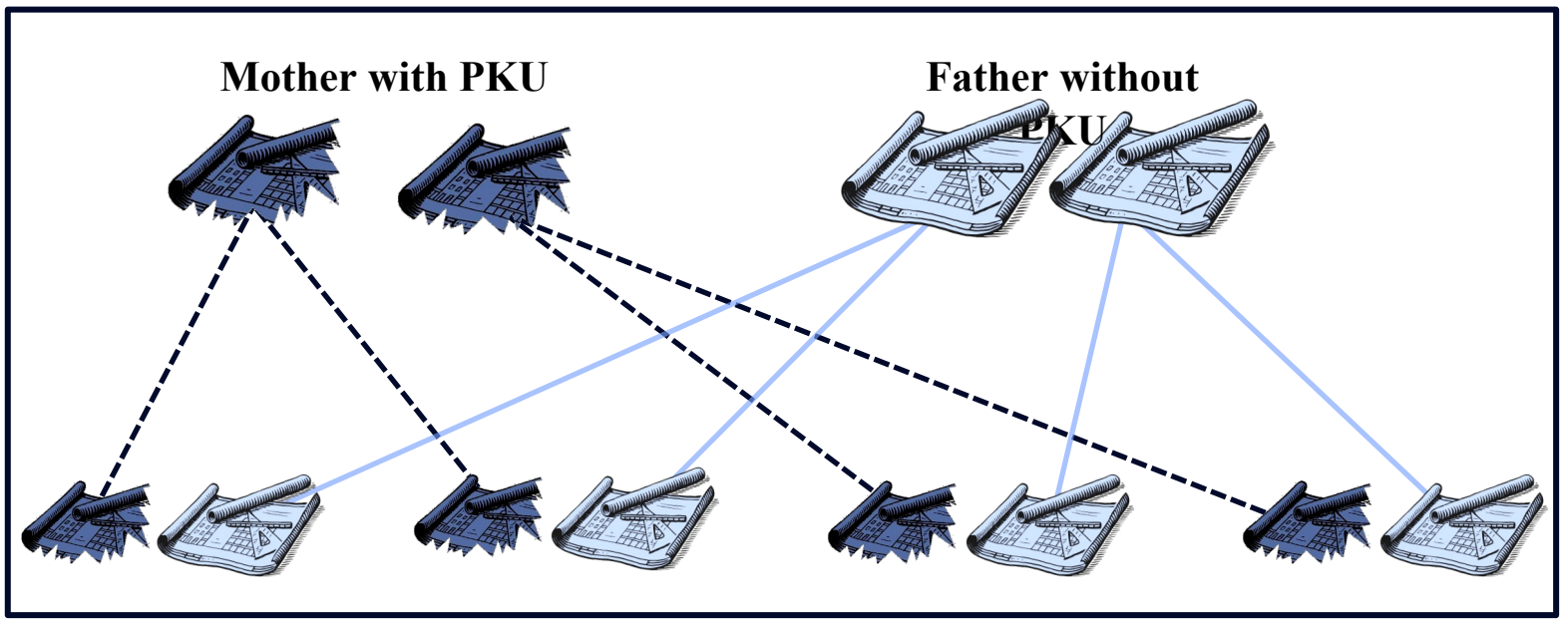
Instructions are normal. Phenylalanine hydroxylase (PAH) is made. Works efficiently.

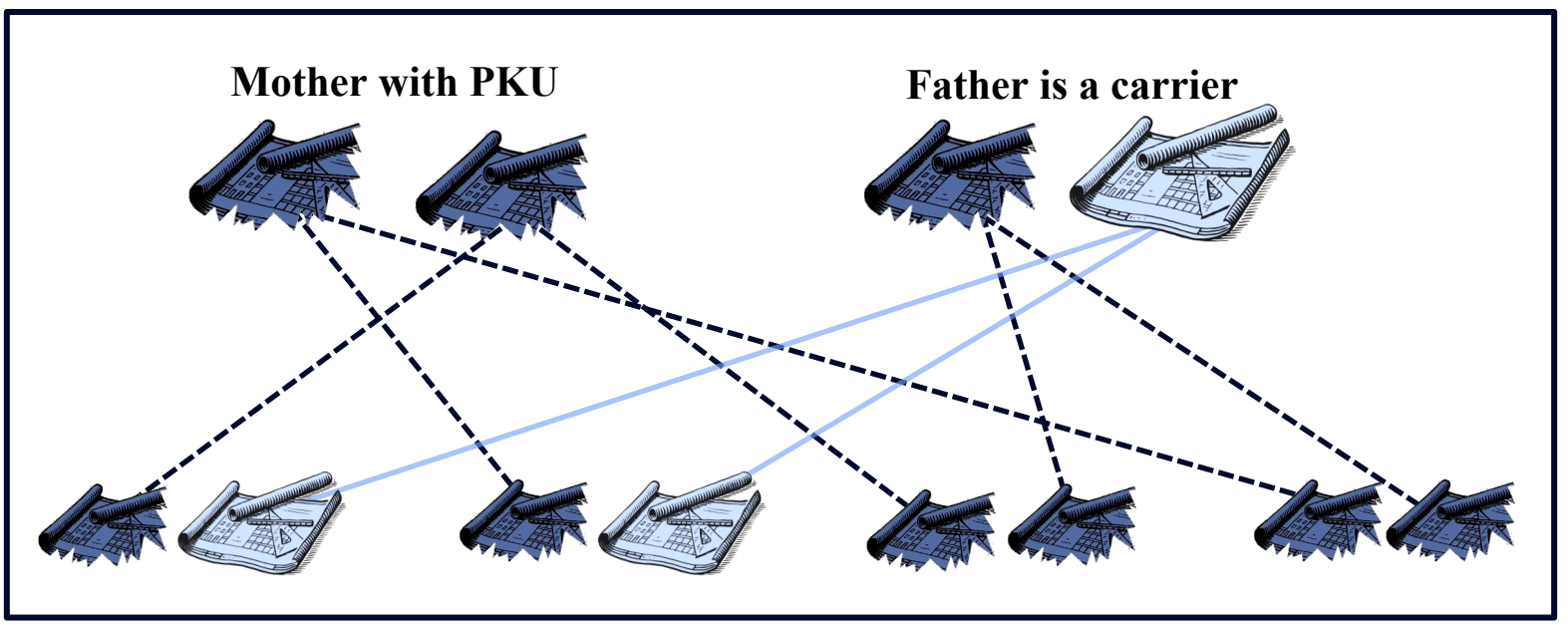
Instructions have an error. Phenylalanine hydroxylase (PAH) is made, however it does not work as well.



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

AUTOSOMAL RECESSIVE INHERITANCE

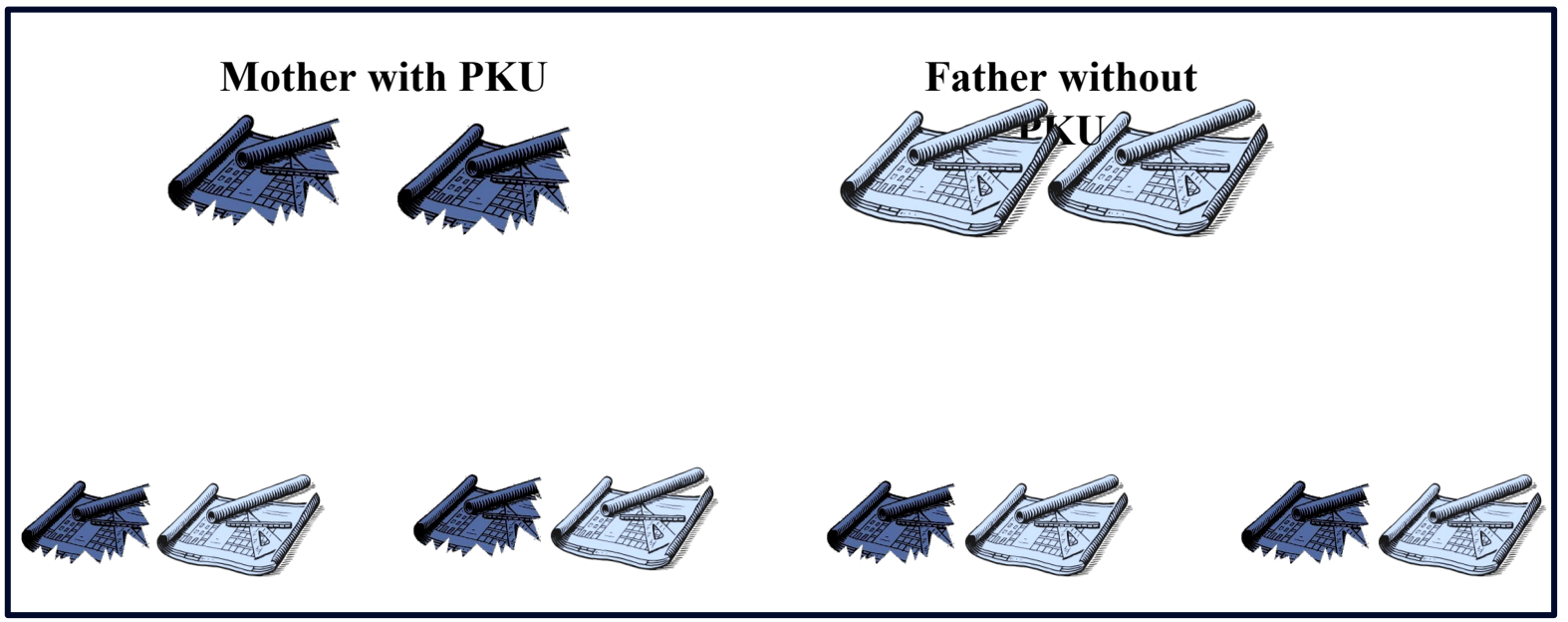


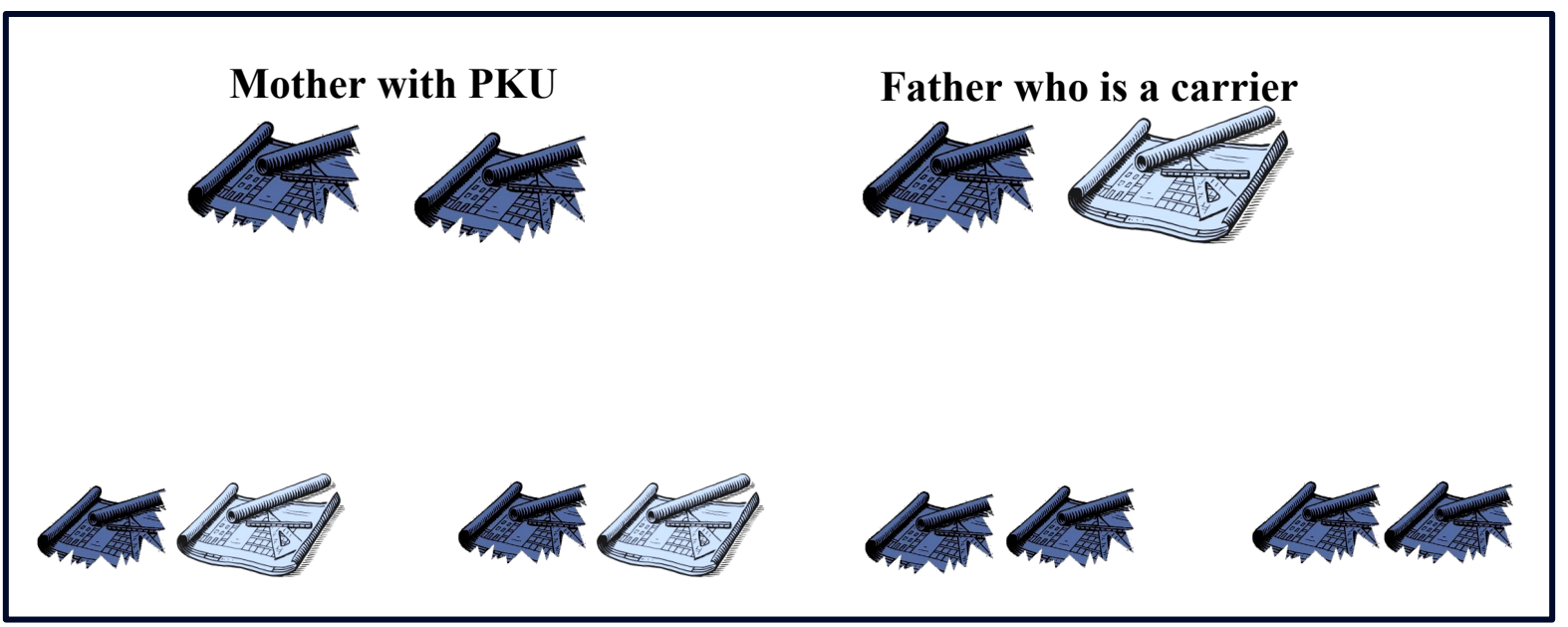


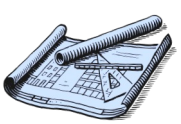

Gene for normal or full phenylalanine hydroxylase (PAH) activity


Gene for altered phenylalanine hydroxylase (PAH) activity

AUTOSOMAL RECESSIVE INHERITANCE







Gene for normal or full phenylalanine hydroxylase (PAH) activity

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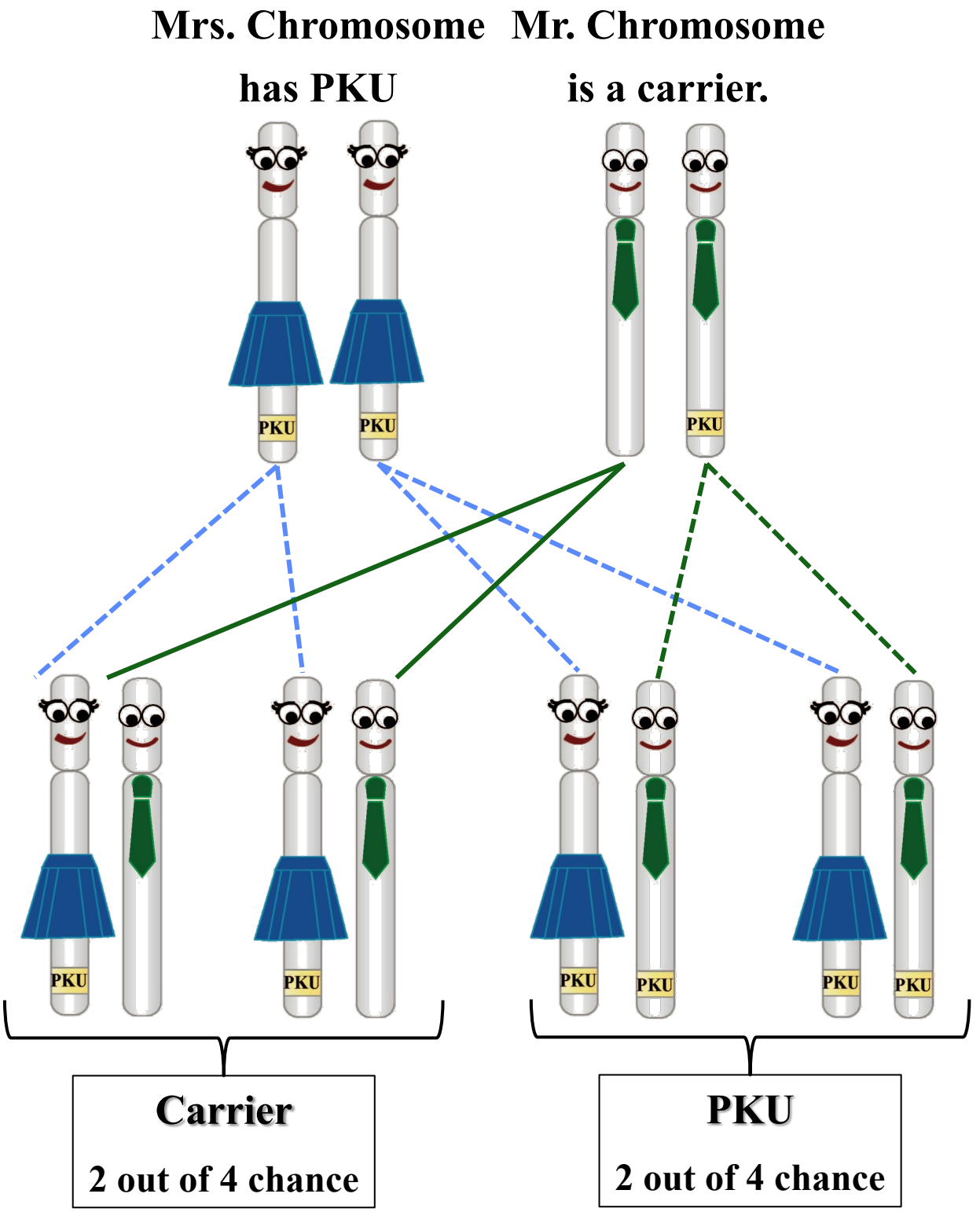
AUTOSOMAL RECESSIVE INHERITANCE

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.



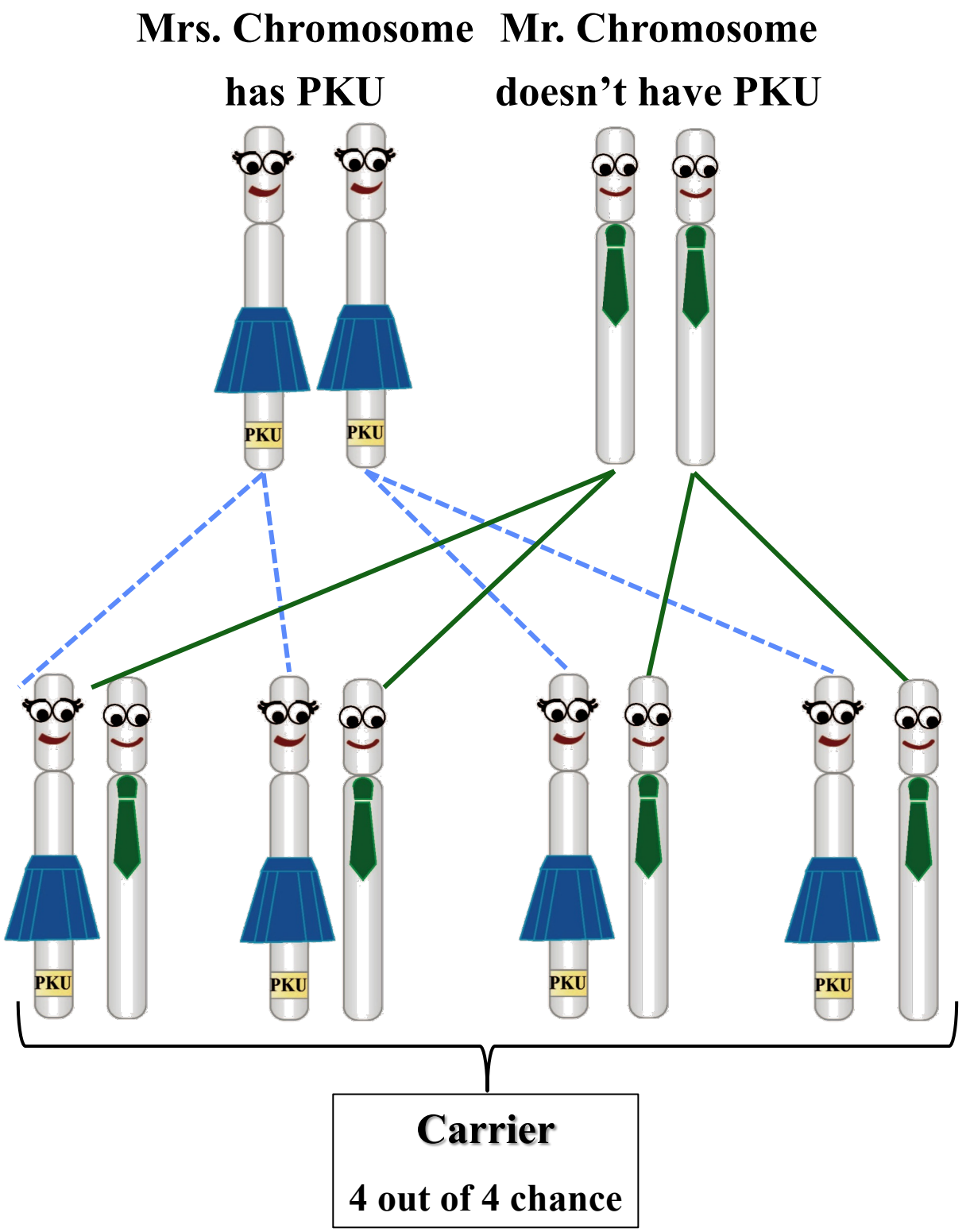
Images by Janine M. Gessner

AUTOSOMAL RECESSIVE INHERITANCE

Chromosome #12 and the PKU Gene

Our genes are organized on our body on Chromosomes.

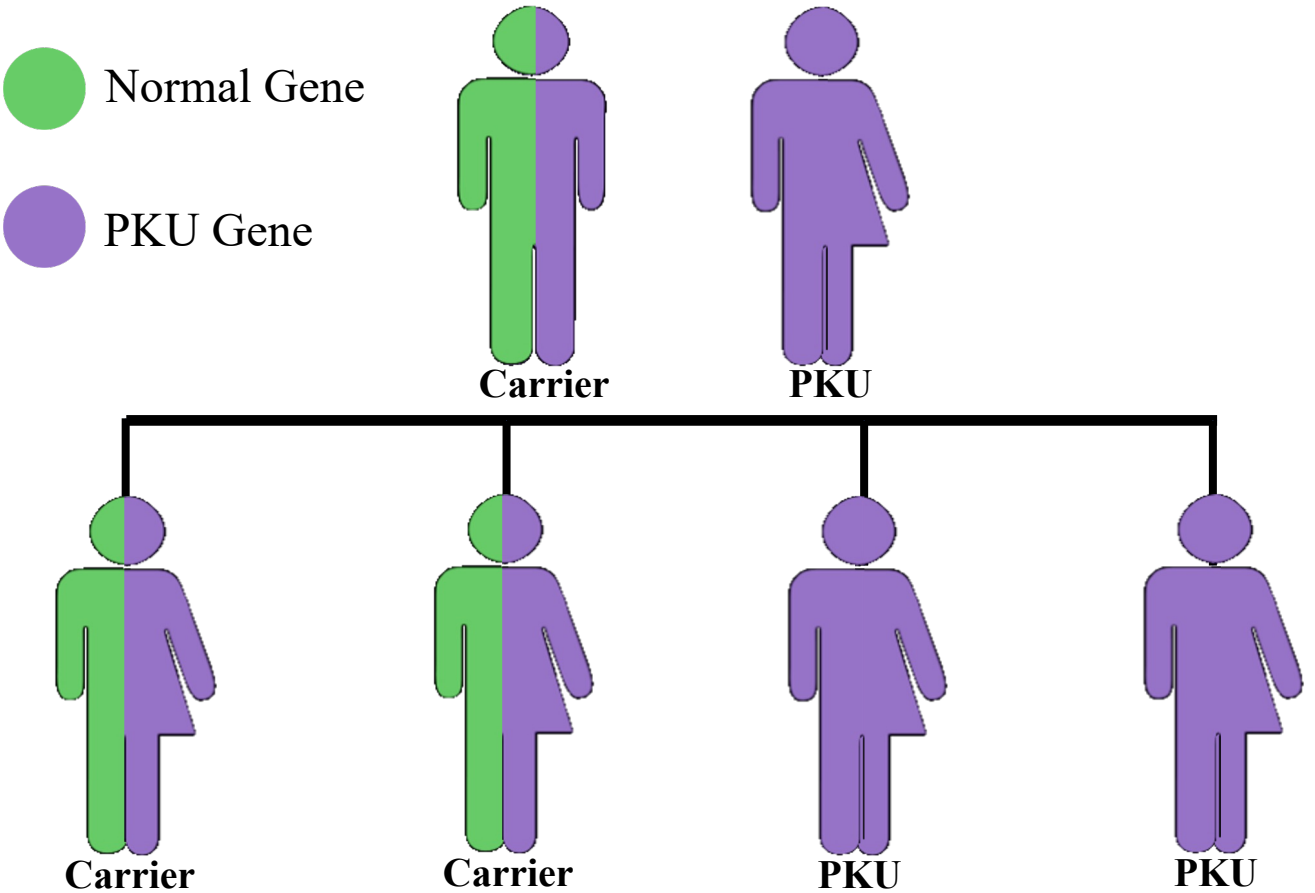
Mrs. Chromosome has PKU and Mr. Chromosome doesn't; see their chances of having PKU based on Autosomal Recessive Inheritance.



Images by Janine M. Gessner

Autosomal Recessive Inheritance

Dad is a carrier. Mom has PKU.



What Does Autosomal Recessive Inheritance Really Mean?

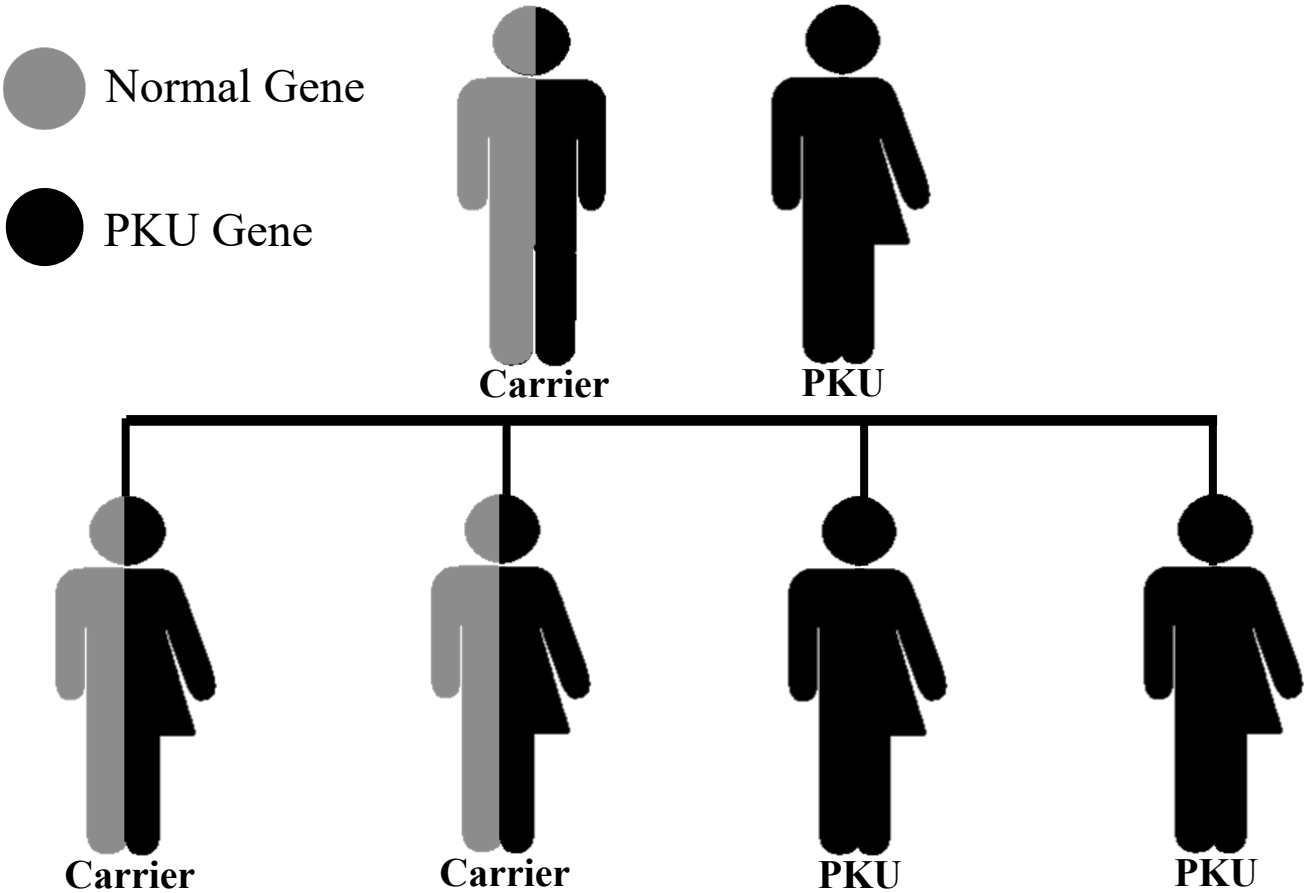
Autosomal recessive inheritance means you have to receive two copies of an altered gene, one from each parent, to have the condition. Individuals who have only one copy of an altered gene are called carriers and do not have the condition.

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.



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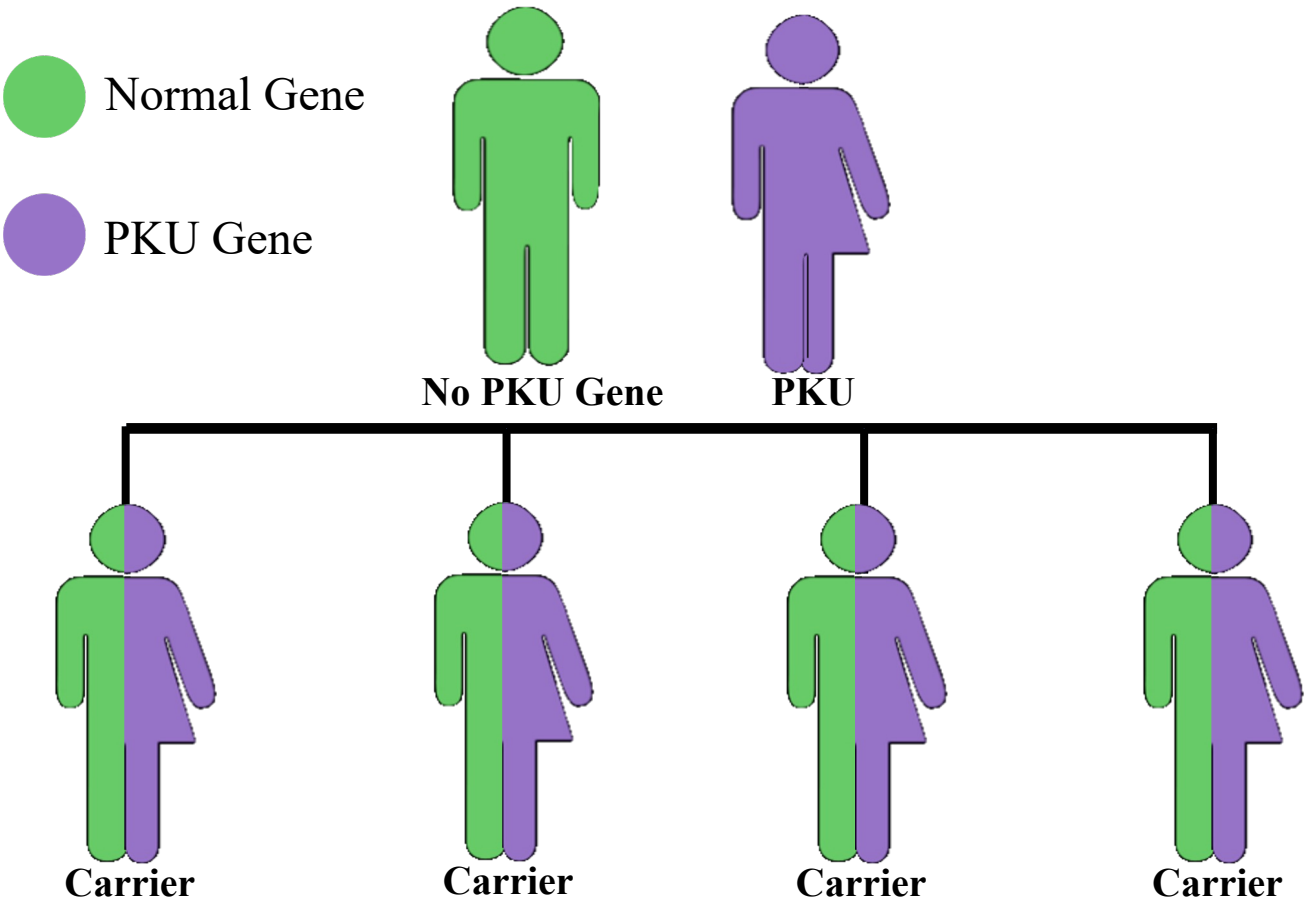
Autosomal Recessive Inheritance

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



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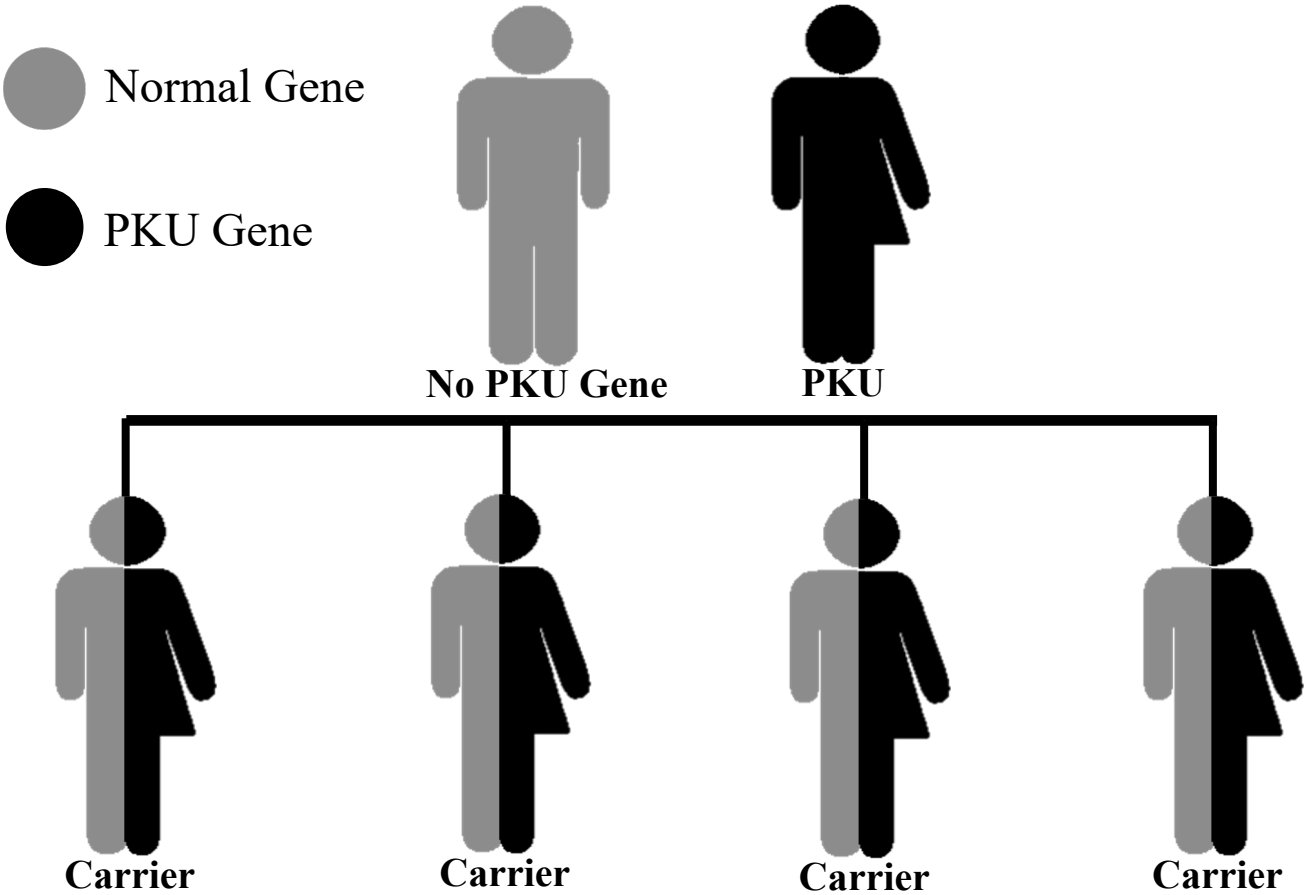
Autosomal recessive inheritance means you have to receive two copies of an altered gene, one from each parent, to have the condition. Individuals who have only one copy are called carriers. They do not have the condition.

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.



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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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