

# EAT RIGHT



# STAY BRIGHT<sup>TM</sup>

## Guide For Hyperphenylalaninemia



The Children's Hospital  
Affiliated with  
University of Colorado Denver

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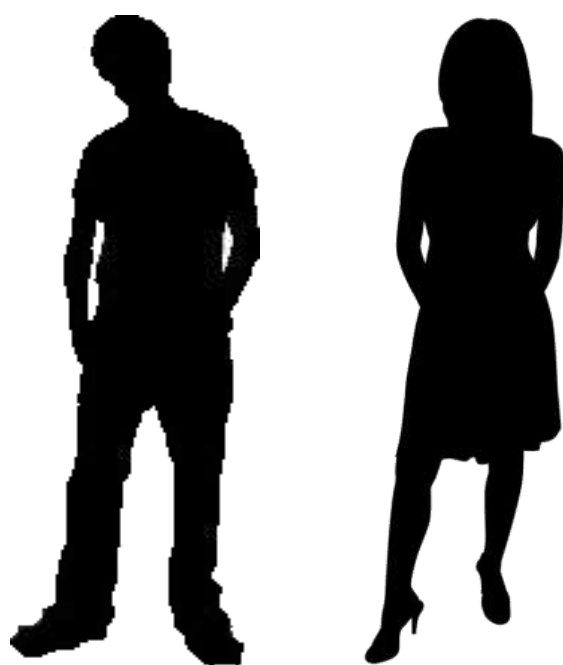
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# CHAPTER THREE



## 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.

# TABLE OF CONTENTS

<u>Title</u>	<u>Page</u>
<b>Clinic Encounter Check Lists</b>	
Introduction	1
Early Adolescence & Parents	2 – 3
Middle Adolescence	4 – 5
Late Adolescence	6 – 7
<b>Experience and Thoughts</b>	8 – 9
<b>Principles of Diet Prescription</b>	
Throughout a Lifecycle	
Male	10
Female	11
8 Years Old Through Adulthood	12
Medical Food Options	13
<b>Daily Diet Prescription</b>	
8 Years Old Through Early Adolescence	14
Middle Adolescence	15
Late Adolescence	16
24 Hour Diet Diary	17
Large Neutral Amino Acids (LNAA)	18 – 19
Glycomacropeptide (GMP)	20
Kuvan	21
<b>Teaching Aids and Handouts</b>	
<b>PKU and Diet</b>	
The Altitude Of Your Attitude	22 – 23
How Do You Feel?	24 – 25
Self Talk	26
Personal Ship	27
How Well Do You Like Yourself?	28
Scale For Self Esteem	29
Do You Hear Yourself?	30
Who Are You?	31 – 32
Problem Solving Tool Kit	33
Problem Solving Strategies	34
Problem Solving Worksheet	35
I Messages	36

<u>Title</u>	<u>Page</u>
<b>Teaching Aids and Handouts...</b>	
<b>PKU and Diet...</b>	
Put Yourself In The Parent Seat	37 – 38
PKU Adventure Game	39
Tic Tac KNOW	40
PKU Twister	41 – 42
“Yes” “No” BINGO	43
BINGO Game Pieces & Boards	44 – 49
PKU Jeopardy	50 – 51
The Golden Key	52 – 53
Polly Ate A Cracker	54
The Naked Egg	55
Weighing & Measuring	56
Weighing & Measuring References	57
Blood Drawing	58 – 59
Risky Business	60 – 61
Management Contracts	62
Guidelines For Negotiation	63
Sample Contracts	64 – 66
<b>Genetics</b>	
DNA Extraction	67 – 68
Unraveling Your Genes	69
Exotic Tea Recipe	70
Mutations	71 – 72
GO! Speed Racers	73
Recessive Traits	74
Recessive Traits Portrait	75
PKU Bracelet	76 – 77
Random Chance	78
PKU Punnet Squares	79 – 80
Enzyme Bill	81 – 83
<b>References</b>	84
<b>Resources</b>	85 – 86

# 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 hyperphenylalaninemia, 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 adolescents to make consistently healthy choices.**

# CHECKLIST: Early Adolescence & Parents

## Planning For The Future<sup>1</sup>

- ☐ Short term goals
- ☐ Long term goals

## Genetics

- ☐ Revisit autosomal recessive inheritance<sup>2</sup>
- ☐ 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<sup>3</sup>

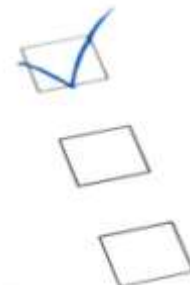
## Phe Levels, Growth Charts, Interim History

- ☐ 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<sup>4</sup> (pg 58 – 59)
  - Time of draw
- ☐ Setting<sup>5</sup>
  - School
  - Home
  - Work<sup>6</sup>

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





# CHECKLIST: Early Adolescence & Parents

## Psychosocial Issues

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

## Nutrition Intervention

- ☐ Formula consumption
  - B-12 deficiency
  - Folate deficiency
- ☐ Use of the following items<sup>17</sup>
  - 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<sup>1</sup>

- ☐ Short term goals
- ☐ Long term goals
- ☐ Achieving goals

## Self Management Strategies

- ☐ Goal setting
- ☐ Monitoring
- ☐ Self administering consequences
- ☐ Management contracts<sup>18</sup> (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<sup>3</sup>

## Daily Living Routine

- ☐ Cooking/recipes
- ☐ Availability of low protein foods
- ☐ Diet records
- ☐ Blood draws
- ☐ Setting<sup>5</sup>
  - School
  - Home
  - Work<sup>6</sup>



# CHECKLIST: Middle Adolescence

## Psychosocial Issues

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

## Nutrition Intervention

- ☐ Formula consumption
  - B-12 deficiency
  - Folate deficiency
- ☐ Use of the following items<sup>17</sup>
  - 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<sup>18</sup> (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<sup>3</sup>

## Daily Living Routine

- ☐ Cooking/recipes
- ☐ Availability of low protein foods
- ☐ Diet records
- ☐ Blood draws (pg 58 – 59)
- ☐ Setting<sup>22</sup>
  - School
  - Home
  - Work



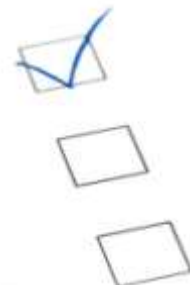
# CHECKLIST: Late Adolescence

## Psychosocial Issues

- ☐ Problem solving skills<sup>19</sup>
- ☐ Family<sup>9</sup>
- ☐ Finances<sup>16</sup>
- ☐ School/vocation
- ☐ Autonomy

## Nutrition Intervention

- ☐ Formula consumption
  - B-12 deficiency
  - Folate deficiency
- ☐ Use of the following items<sup>17</sup>
  - 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

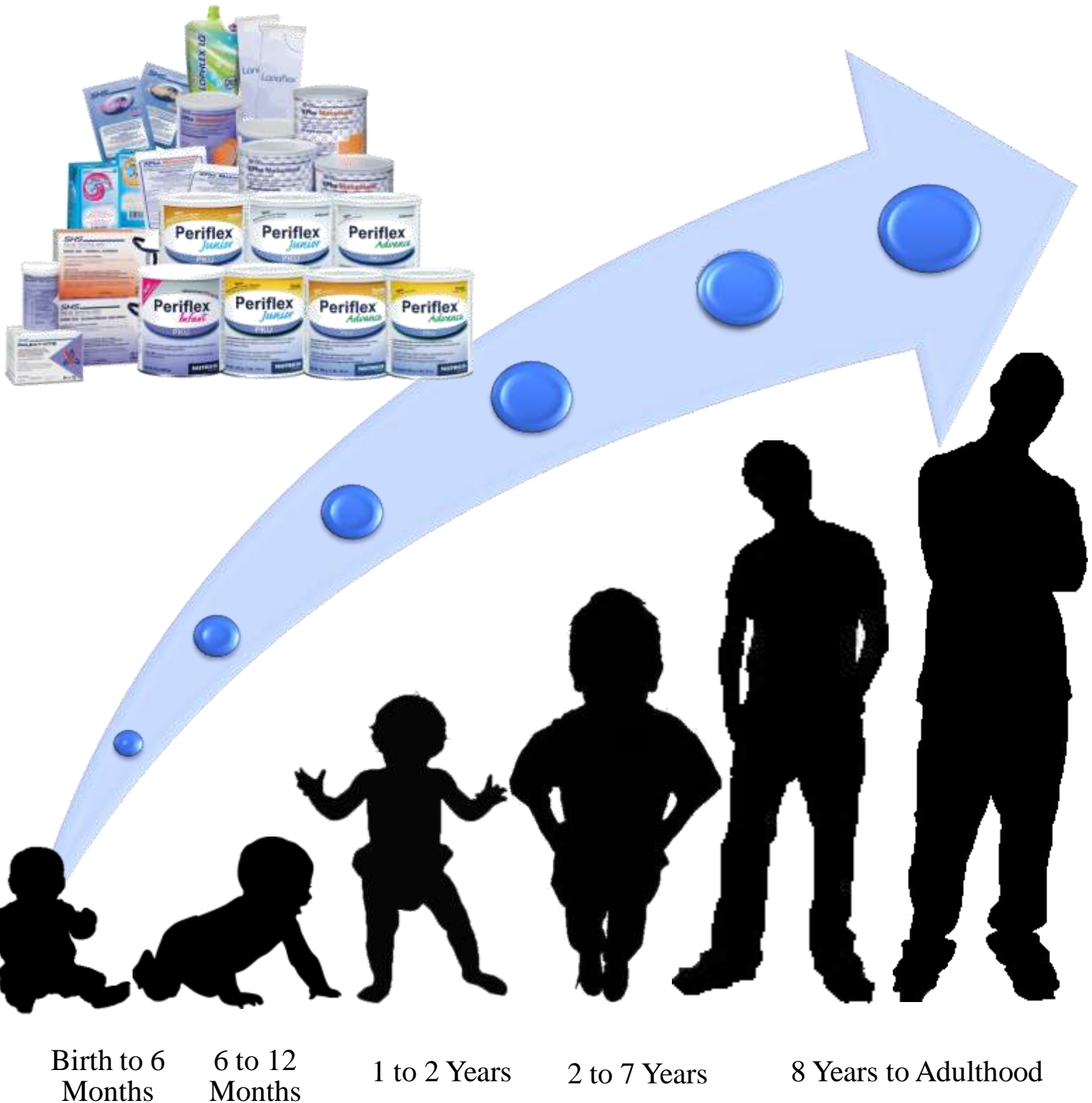
1. It has been our experience that having both the adolescent and parents verbalize and discuss goals on an ongoing basis leads to realistic expectations and facilitates future planning.
2. Revisiting autosomal recessive inheritance may be critical in a family where there are unaffected siblings. Understanding 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

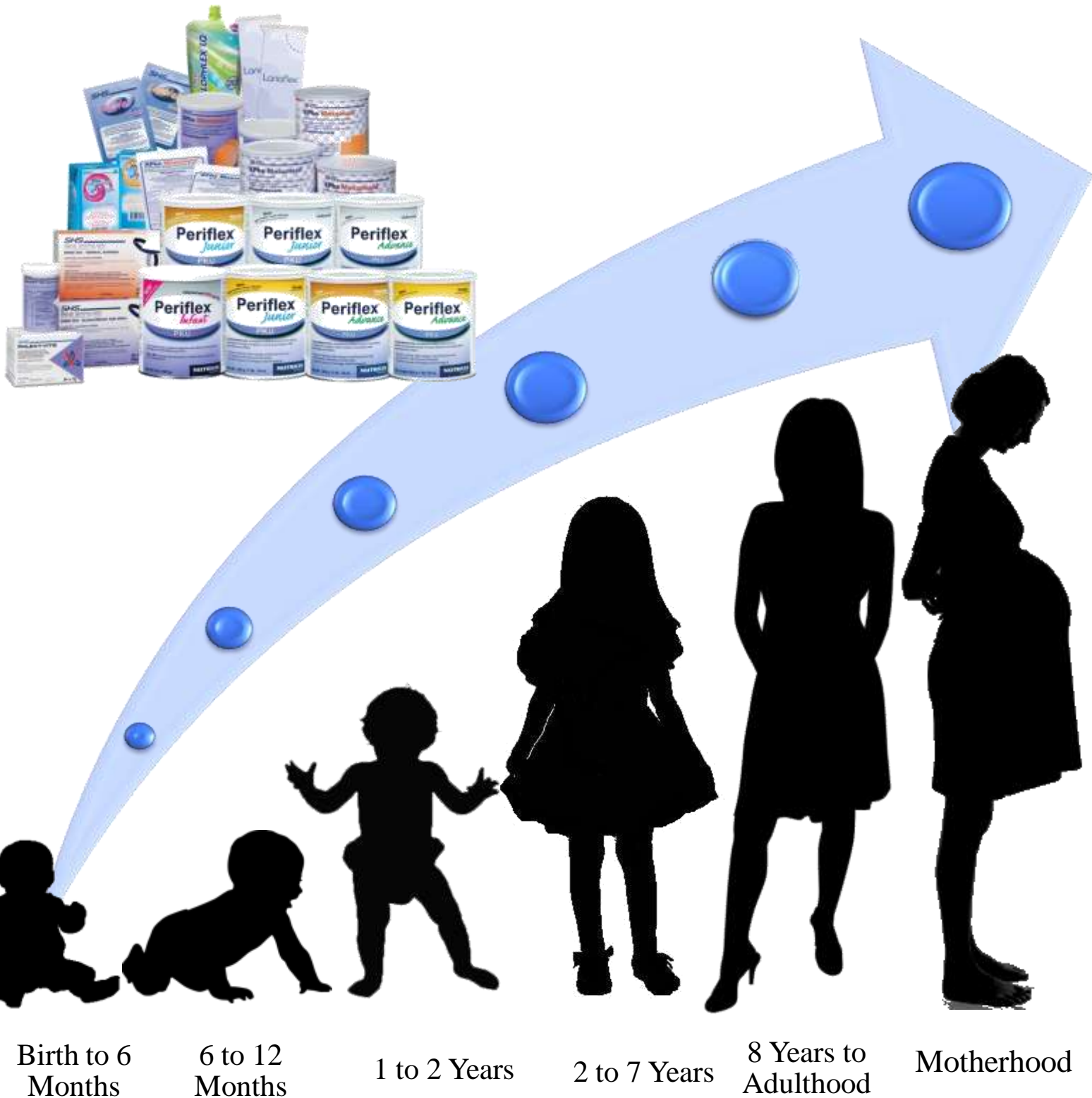


**At Every Age, Medical Food Is An Important Part Of Your Daily Nutrition!**



# PRINCIPLES OF DIET PRESCRIPTION

## Throughout a Lifecycle



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

# PRINCIPLES OF DIET PRESCRIPTION

## 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



**Food**



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



Food



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



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



Food



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)
<b>Totals</b>					

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 18 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 and SHS Int. support “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 18 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](http://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 its use as an ingredient for nutritional formulations and health foods over the last 15 – 20 years. Because whey 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 your favorite foods

**More information available at**  
**[www.cambrookefoods.com](http://www.cambrookefoods.com)**



### 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.

### 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.

### What is KUVAN?

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

### Ingredients of KUVAN

**Active Ingredient:** sapropterin dihydrochloride.

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



*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](http://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.



### 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 Needed**  
The Altitude of Your Attitude

# **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 you 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?

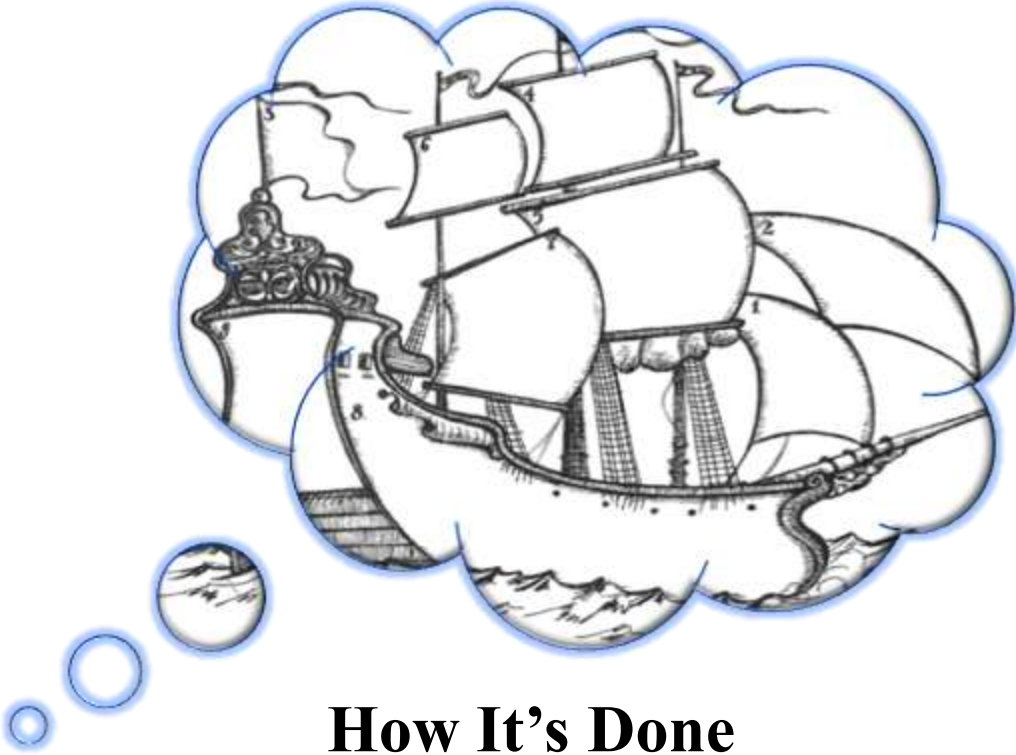




# TEACHING AID

## 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

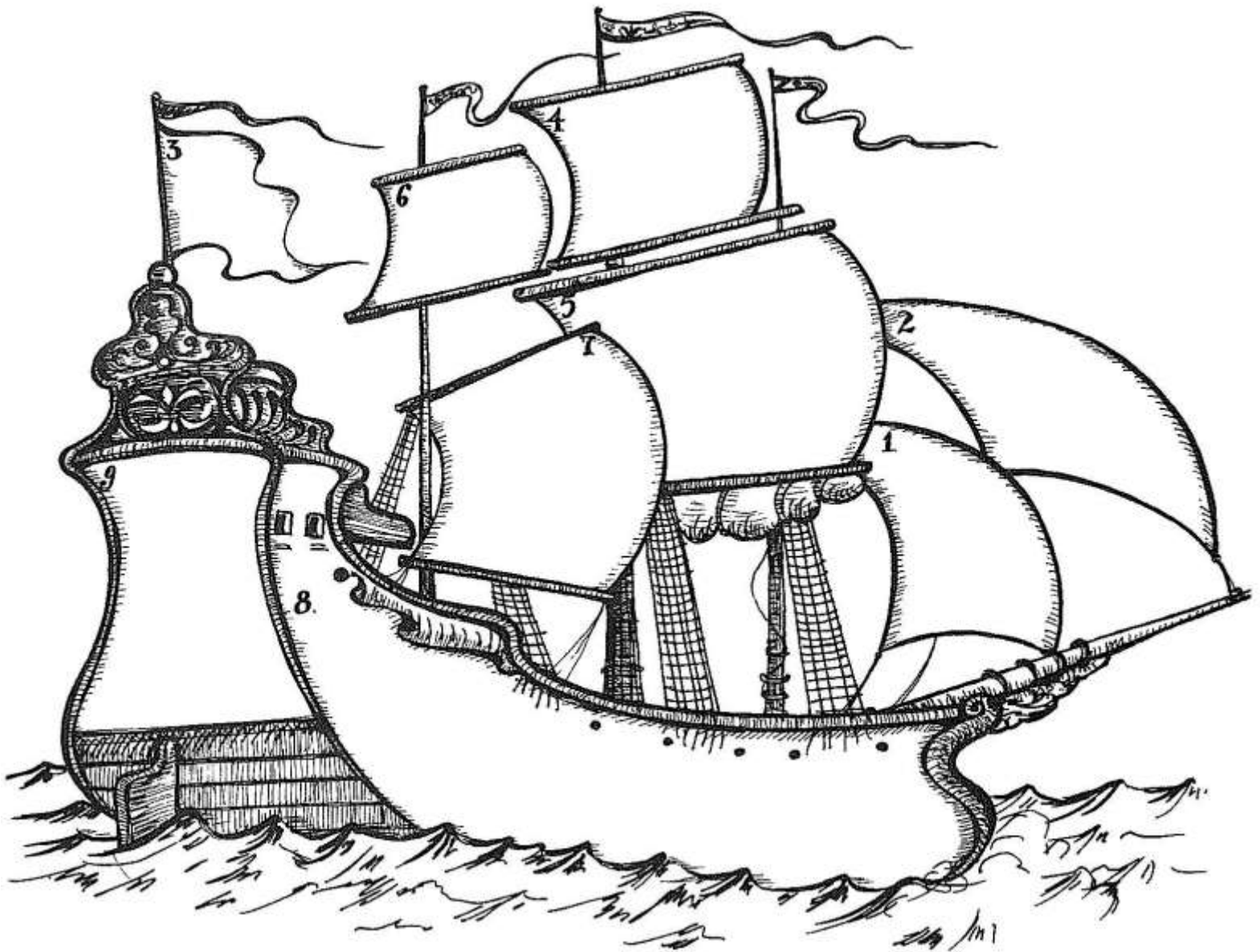
Adapted from M. Halter and B. Fierro Lang Making Choices



# 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 from
9. Three words I would like people to use to describe me



# How Well Do You Like Yourself?

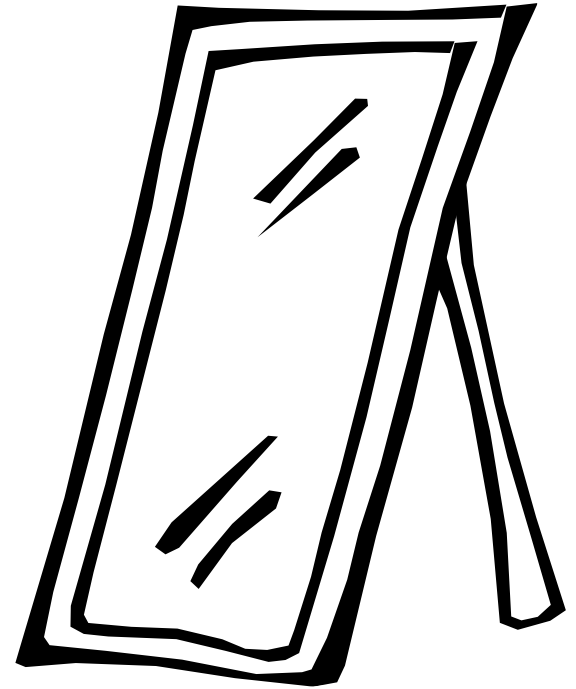
Complete the survey below using the following scores:

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

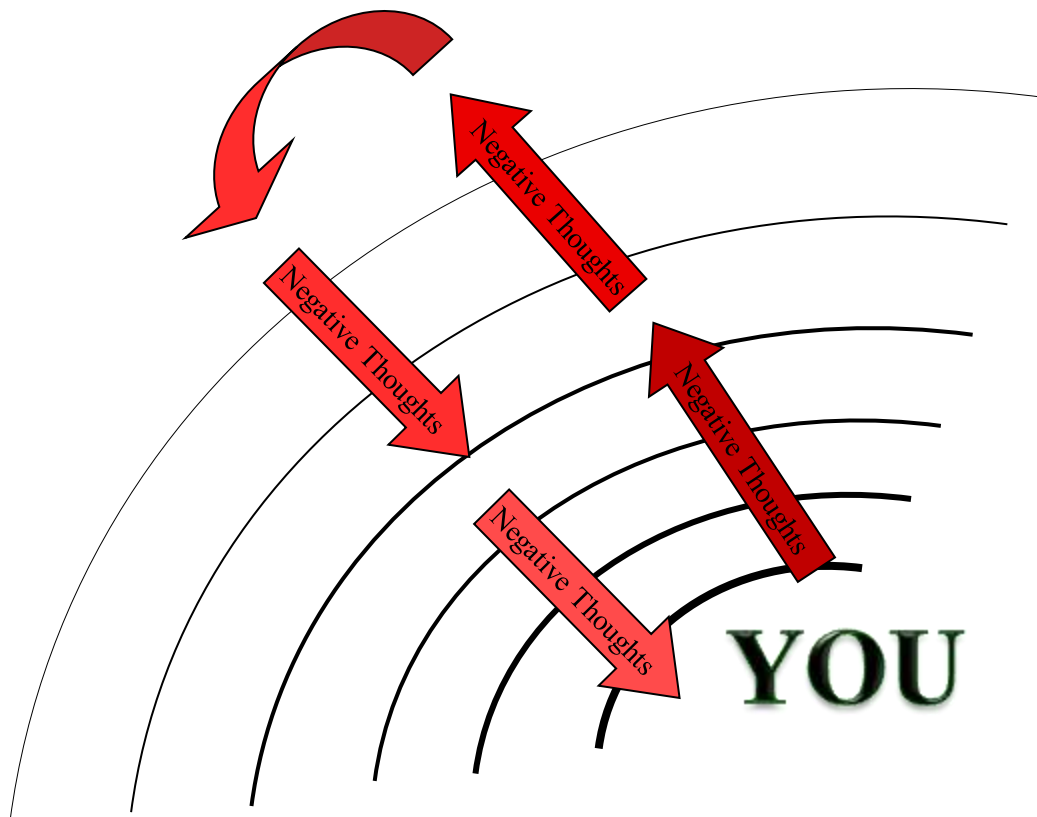
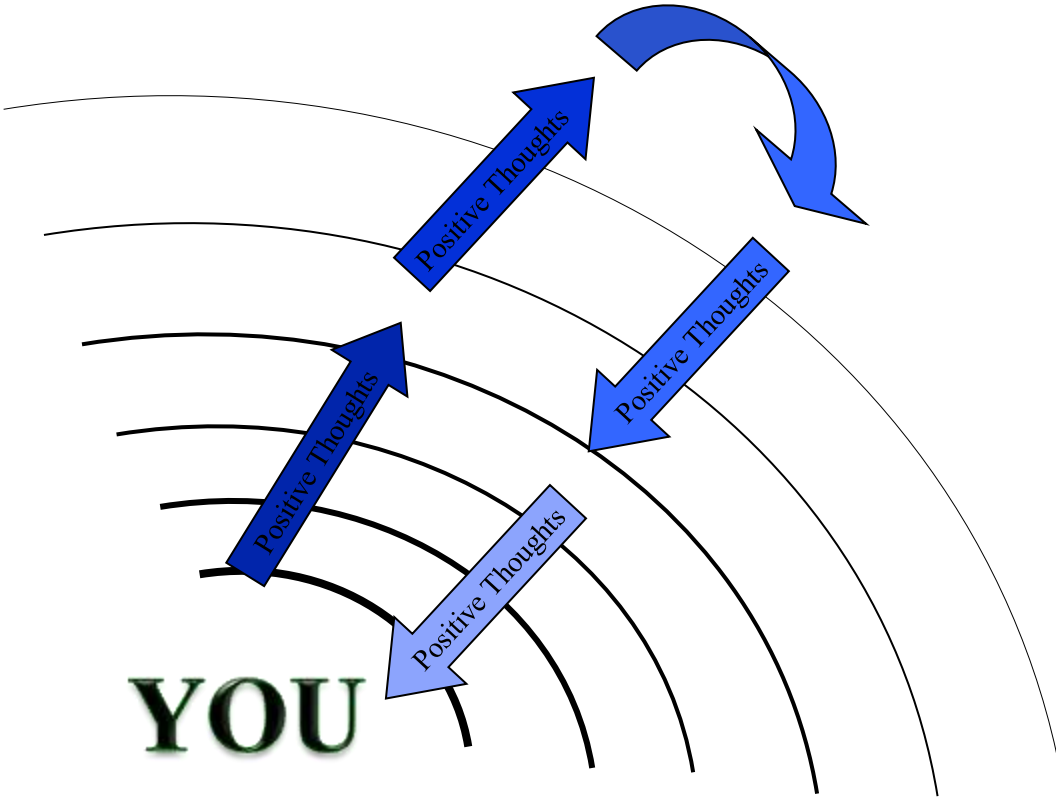
Total Score	What Your Score Means
200-225	Very high self esteem, positive self-worth. You like yourself.
120-195	Self-esteem and self worth are good. You accept yourself and your life. Self image could be improved with conscious effort.
45 -115	Self esteem is so-so. Acceptance of yourself and your self 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!**

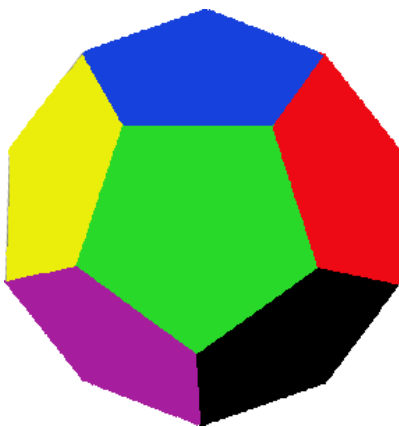
# Do You Hear Yourself? Are You Listening?



# 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.



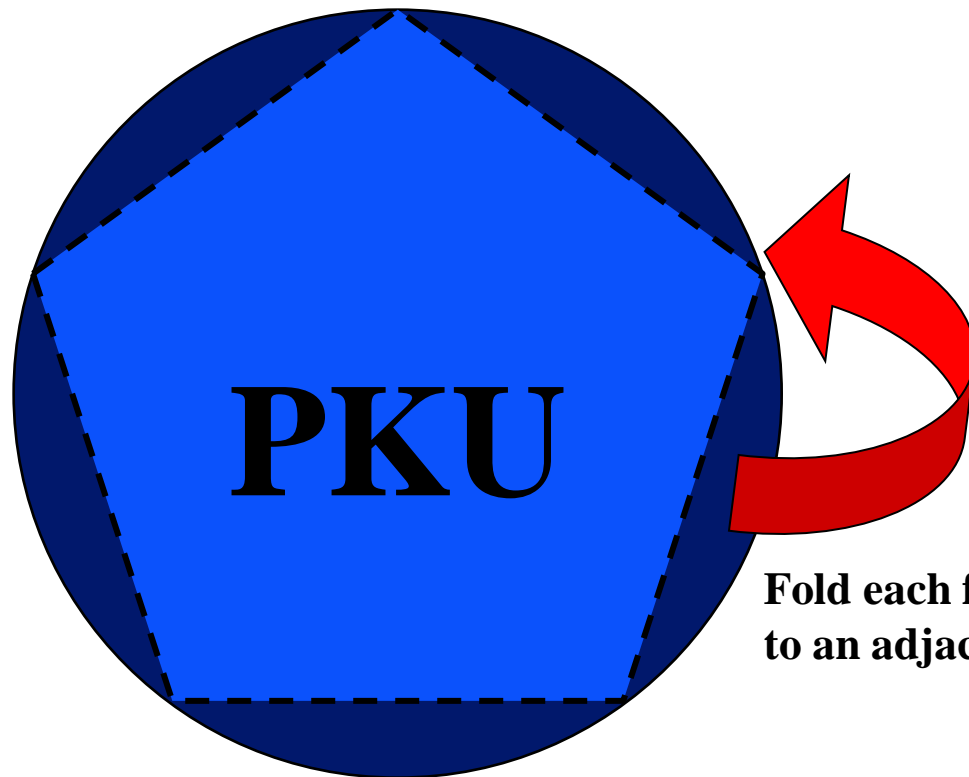
## How It's Done

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  $\frac{1}{4}$  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?

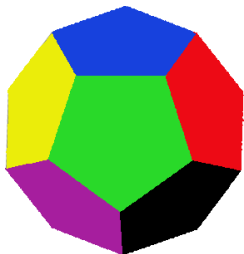
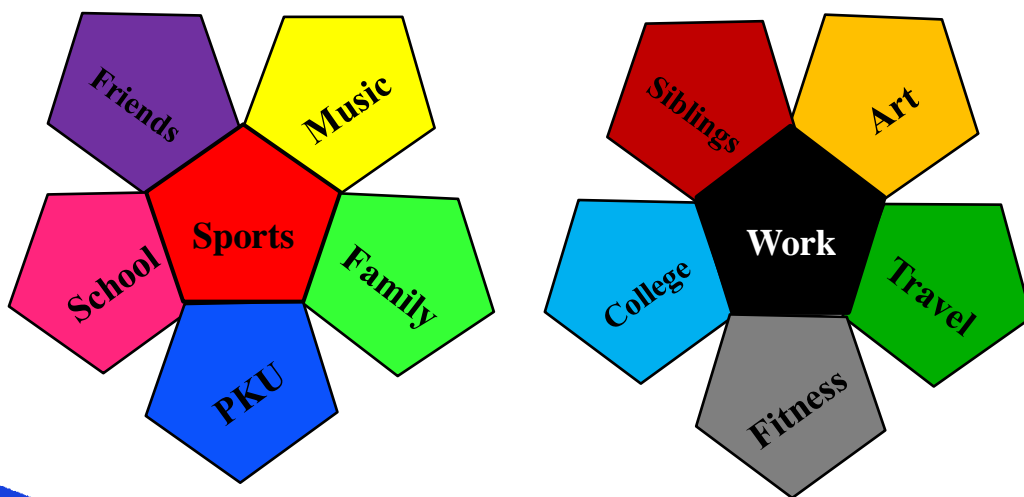
# Who Are You?



**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

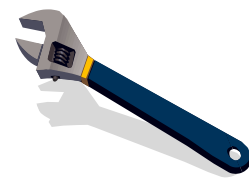
- ❖ Problem Solving Strategies
- ❖ Problem Solving Worksheet
- ❖ I Messages

# 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 Adherence to Pediatric Medical Regimens**



# Problem Solving Worksheet

1. Identify the problems with compliance:

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

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

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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:

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

## *Example*

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 5<sup>th</sup> 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 Adherence to Pediatric Medical Regimens

# 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.

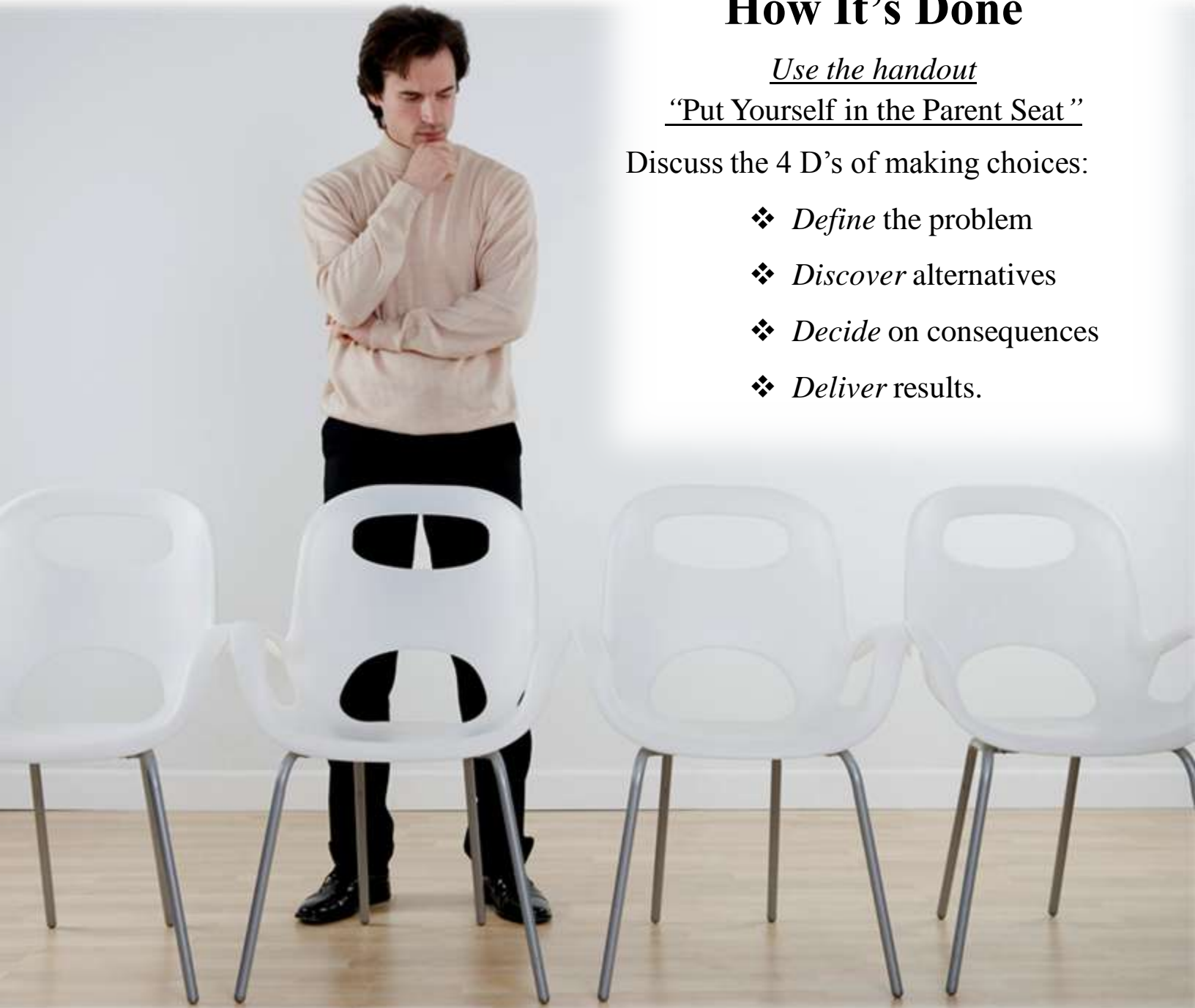
### How It's Done

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

Chapter Three Teaching Aid: PUT YOURSELF IN THE PARENT SEAT

# 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?

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**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!

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**Deliver** results: Will the consequences above accomplish the desired change in behavior?

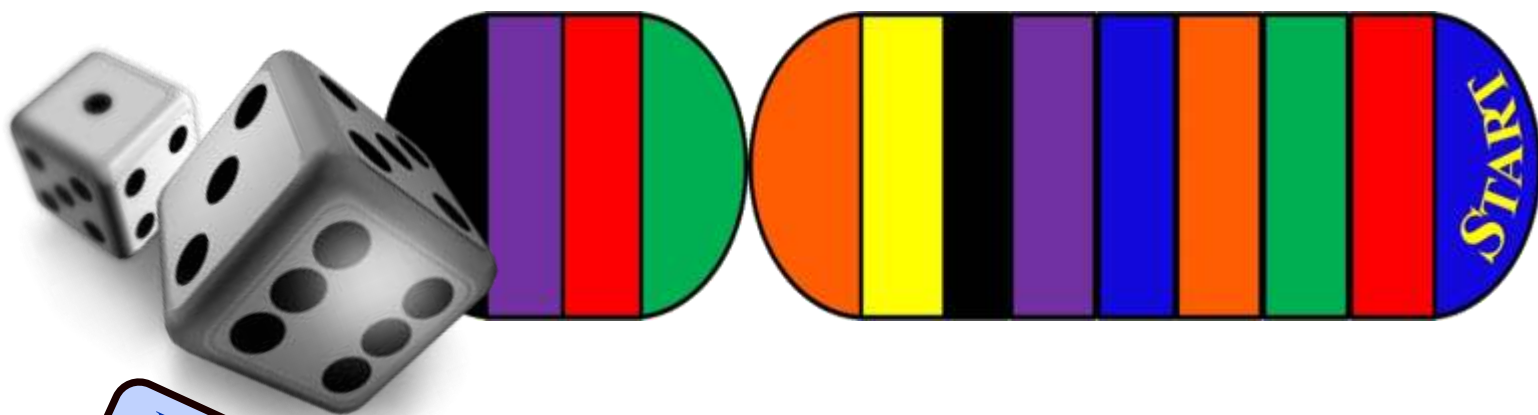
Consider the following questions:

- ❖ 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 set so 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.



### PKU Question Card

What is the name of your formula?

### PKU Question Card

What does PKU stand for?

### PKU Question Card

How much Phe do you get from food each day?

### Materials Needed

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

## How It's Done

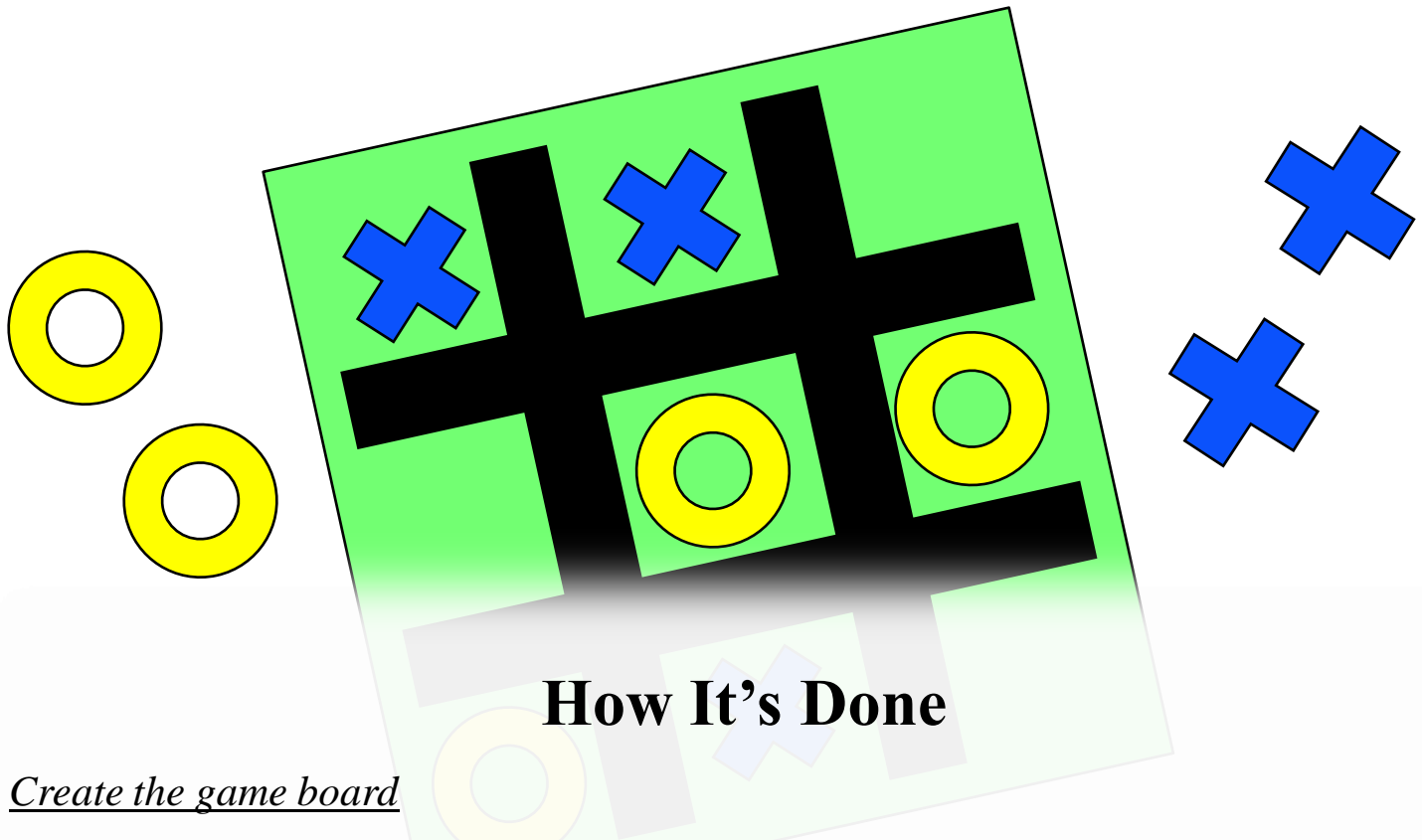
### Set up a board game

- ❖ 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 roll 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 use a large piece of poster board. Using an additional piece of poster board, create 10 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.

### How It's Done

#### Using a Twister Game

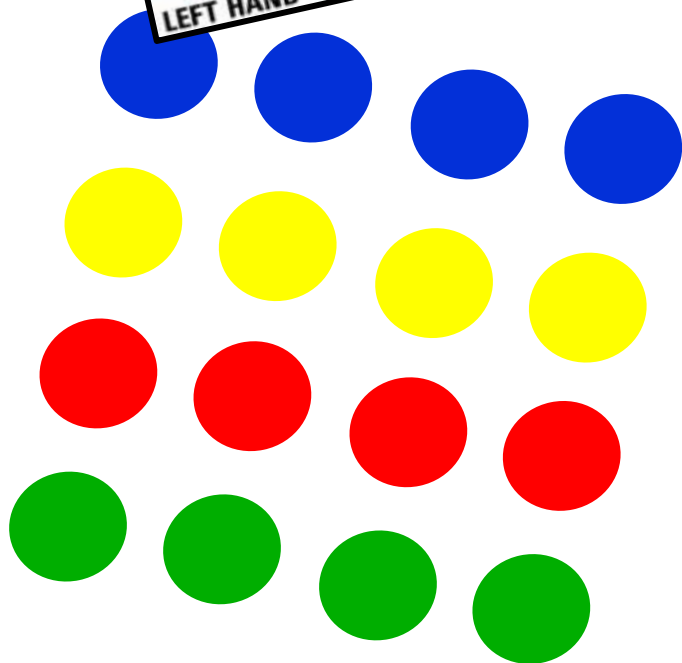
- ❖ Mark each color on the twister spinner board with a different topic; Phenylalanine, PAH, Tyrosine, Neurotransmitters (or make up other topics you want to discuss).
- ❖ When the spinner lands on a color/topic a question from that category will be asked (see Twister Questions Handout). The participants 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 individuals.



#### **Materials Needed**

- ❖ Twister Game

#### **Handout:**

Twister Questions



# 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.*

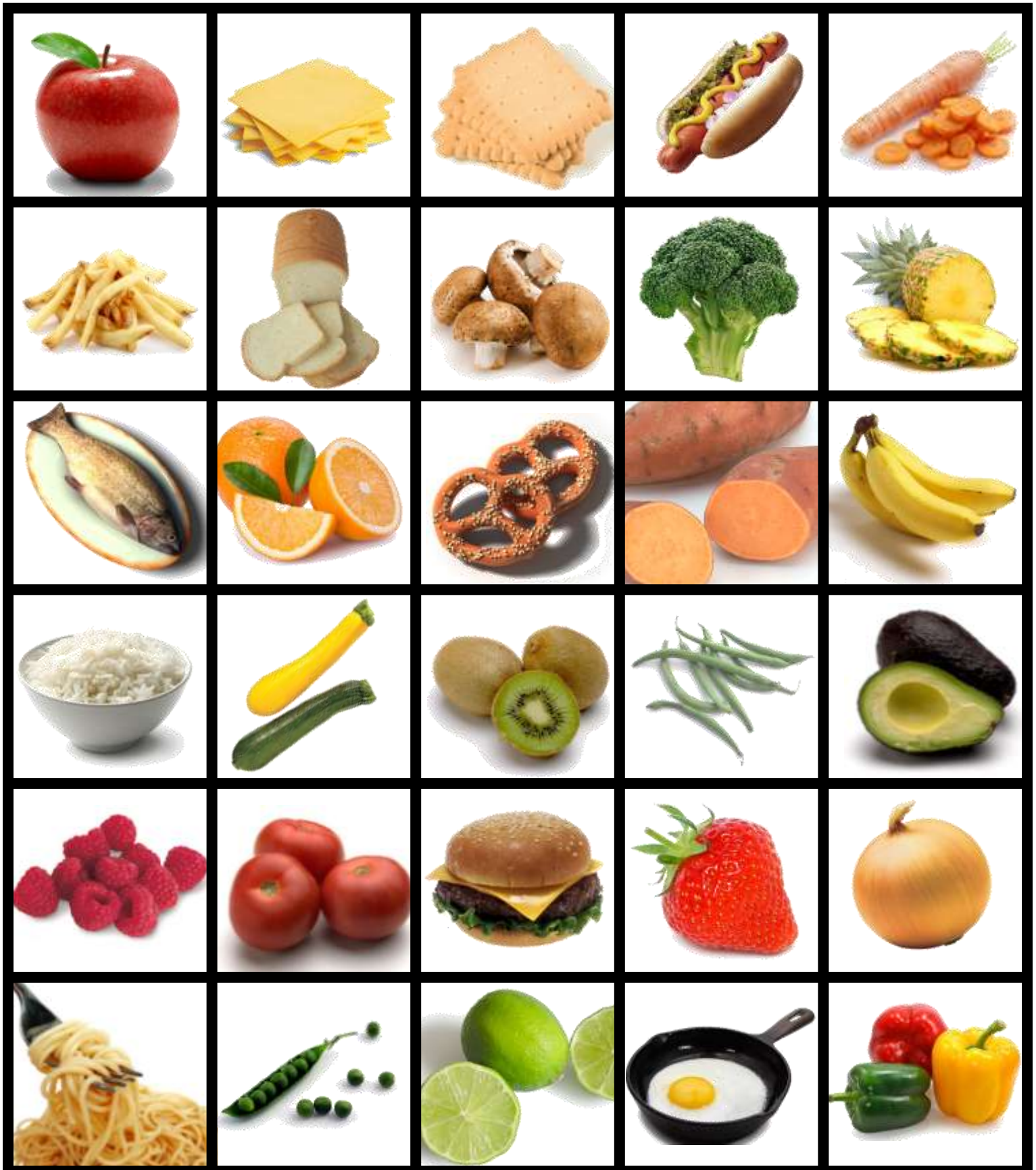
- ❖ Using the cut out game pieces mixed in an envelope, draw one square and call the name of the food out loud. Have participants mark the appropriate square with a piece of candy, and have the participants say if it is a yes, no, or sometimes food (for older participant, begin to discuss what a typical 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

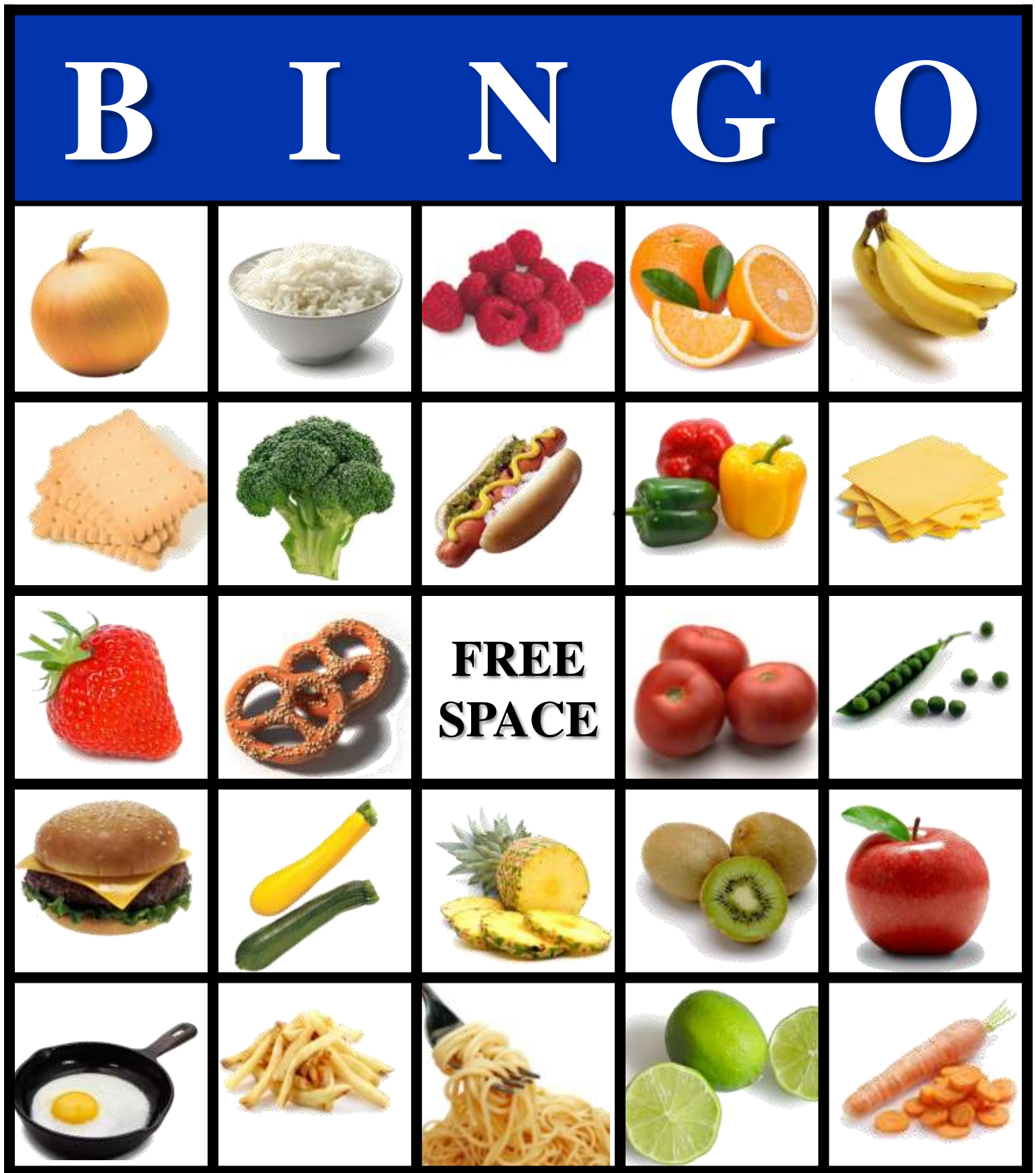
Cut out these game pieces. As you draw them, have the participants place candy on the appropriate square and discuss each food until someone gets 5 in a row. Discuss that some of these foods are only “yes” foods if they are a low protein option (i.e. rice, bread, pasta).





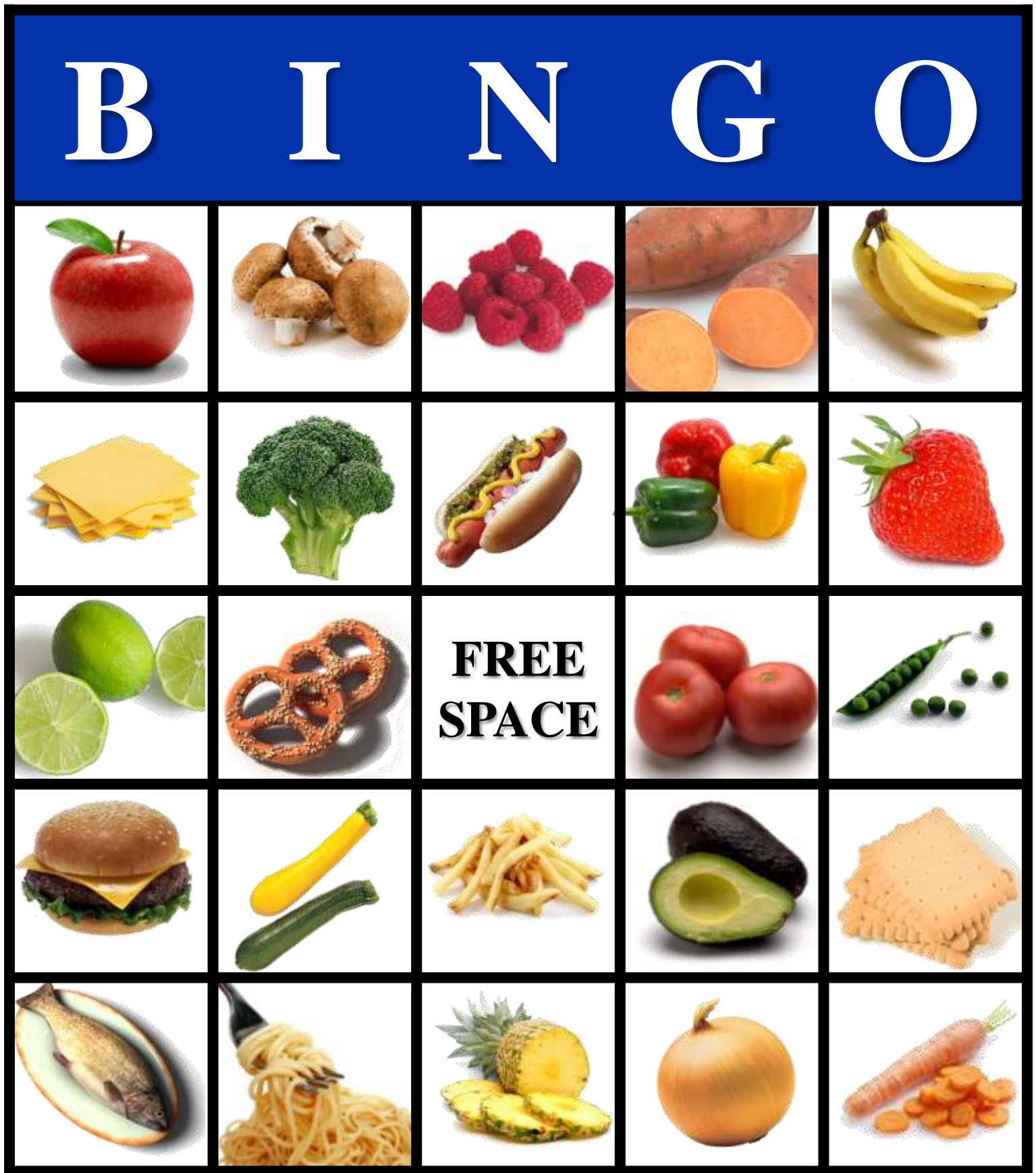
# BINGO GAME BOARD

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



# BINGO GAME BOARD

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





# 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

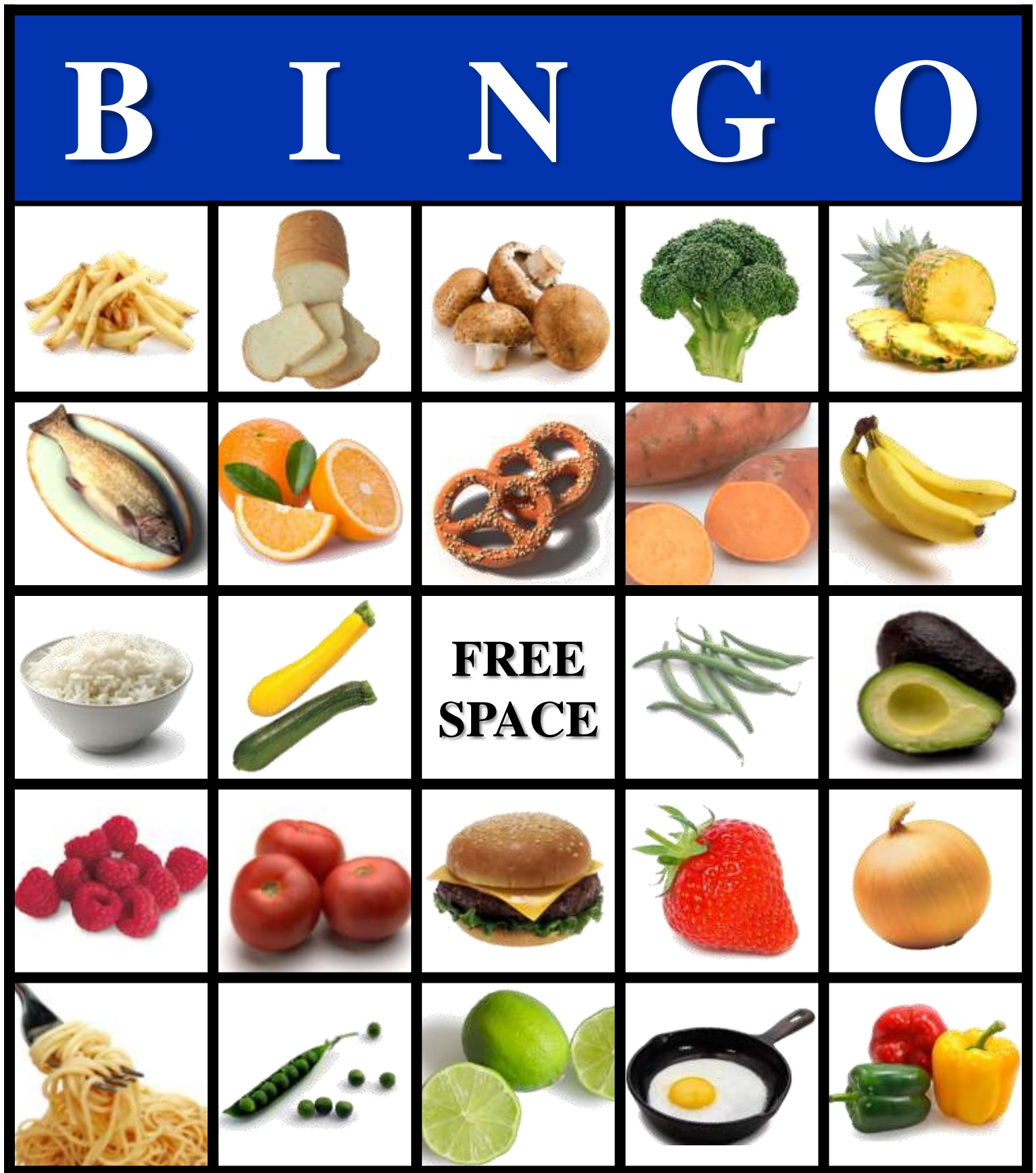
Place candy on the appropriate square and discuss each food until you get five in a row.





# BINGO GAME BOARD

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



# TEACHING AID

## PKU Jeopardy

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

# JEOPARDY

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.

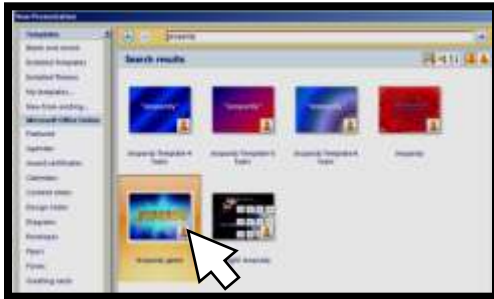
#### Materials Needed

- ❖ 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).*

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

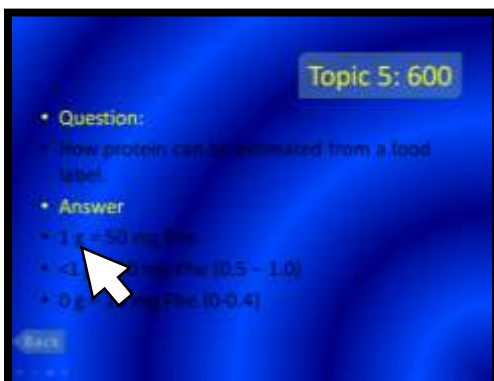
**Q:** What is the enzyme that is broken in PKU?

**A:** Phenylalanine Hydroxylase (PAH)

The question would be phrased like:

**Q:** The enzyme that is broken in PKU?

**A:** *What is* Phenylalanine Hydroxylase (PAH)



## Step #5: Play The Game!

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

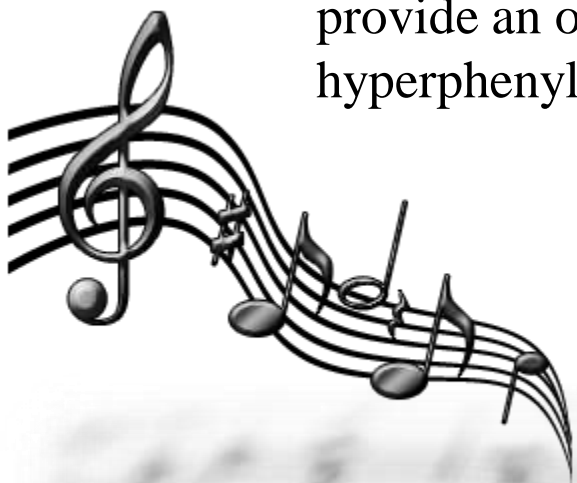
A sample Jeopardy Power Point Game is included as a separate file. Be sure to add questions about information covered in your clinic.



# 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.



### 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.

**Handout Needed**

The Golden Key Song Sheet



# 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.

### How It's Done

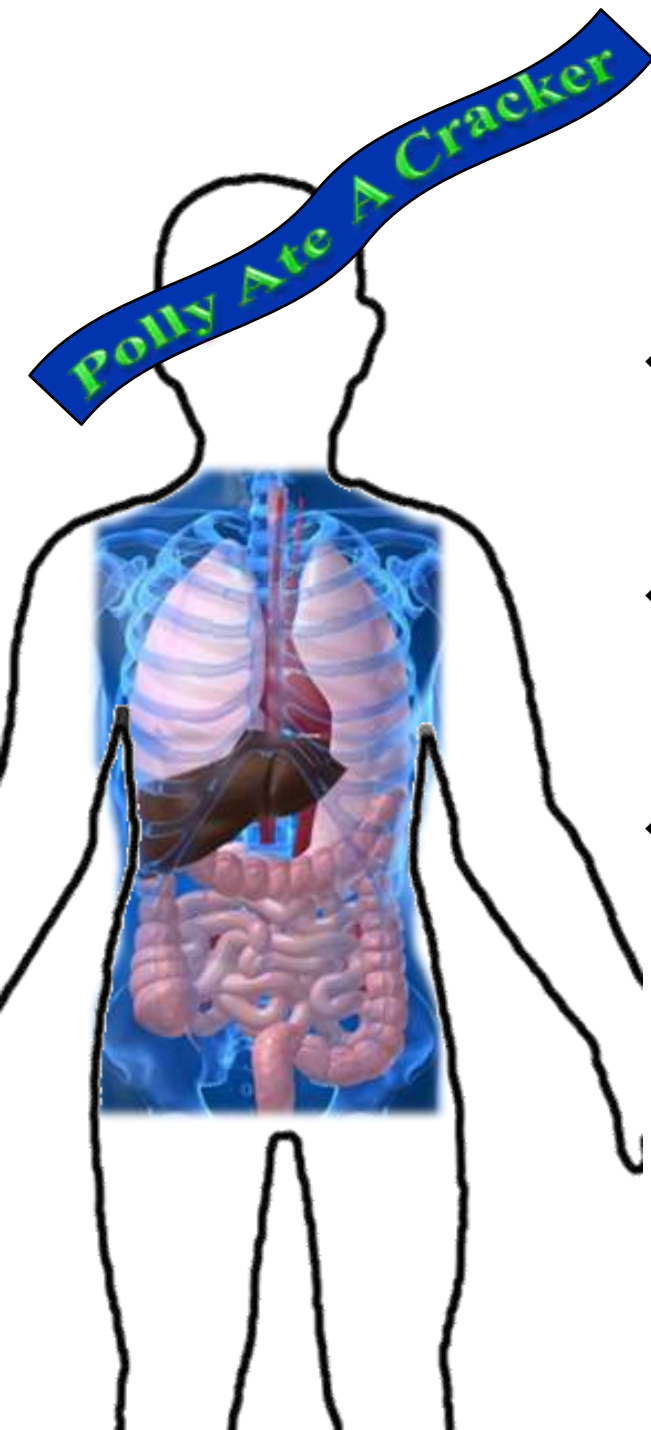
Begin with a discussion of Autosomal Recessive Inheritance.

- ❖ 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.”

### Materials Needed

- ❖ Butcher Paper
- ❖ Markers



# TEACHING AID

## The Naked Egg

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

### 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.



#### Materials Needed

- ❖ Jars
- ❖ Eggs
- ❖ Vinegar
- ❖ Wire
- ❖ Beads

# 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

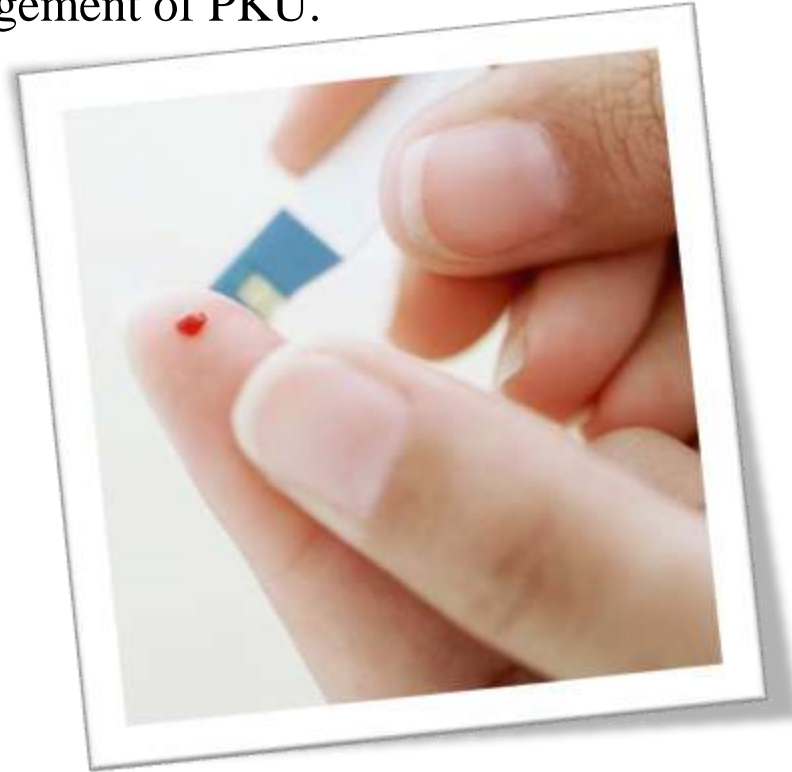
## Metric vs. US Measurements

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

# Risky Business

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

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 not 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 be 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 be 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<sub>12</sub> 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.

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



# 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.*

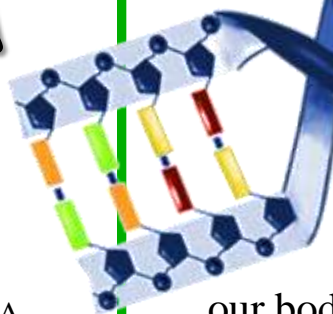
## DNA

DeoxyriboNucleic Acid

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

# MUTATIONS

## *Original Message:*

Wash the dishes after dinner.

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.

### 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.



**Lime  
Racer**



**Nitro  
Muscle**



**Blue  
Bullet**

### Materials Needed

- ❖ 3 Lego® racing vehicles
- ❖ 3 sets of instructions



# TEACHING AID

## Recessive Traits

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

### 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.



#### Materials Needed

- ❖ Coins (2 per team)

#### Handout:

Recessive Traits Portrait

# 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

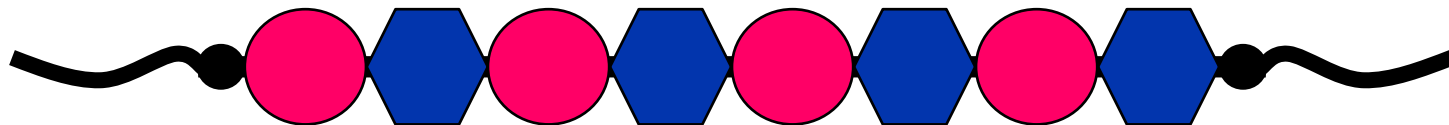
**Draw a picture of what your child will look like.**

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

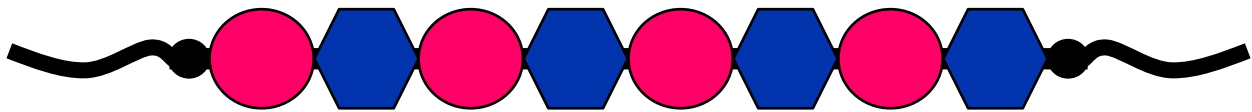
### Materials Needed

- ❖ 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 its 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.

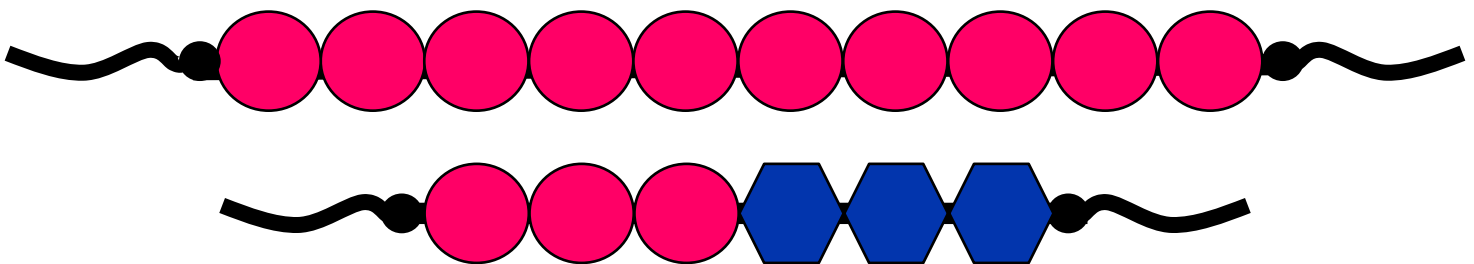
**This is an example of what a PAH enzyme would look like if the PAH gene has no errors:**



This enzyme can breakdown Phe.

**Make two bracelets that look 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 genotypes 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 homozygous 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 genotype, for example one carrier parent, no carrier 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 genotype. 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	<b>PAH/PAH</b>	<b>PAH/PAH</b>
PAH	<b>PAH/PAH</b>	<b>PAH/PAH</b>

**Risk of PKU 0%**

	PAH	pah
PAH	<b>PAH/PAH</b>	<b>PAH/pah</b>
pah	<b>PAH/pah</b>	<b>pah/pah</b>

**Risk of PKU 25%**

	PAH	pah
PAH	<b>PAH/PAH</b>	<b>PAH/pah</b>
PAH	<b>PAH/PAH</b>	<b>PAH/pah</b>

**Risk of PKU 0%**

	pah	pah
PAH	<b>PAH/pah</b>	<b>PAH/pah</b>
pah	<b>pah/pah</b>	<b>pah/pah</b>

**Risk of PKU 50%**

	pah	pah
pah	<b>pah/pah</b>	<b>pah/pah</b>
pah	<b>pah/pah</b>	<b>pah/pah</b>

**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 conversion 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

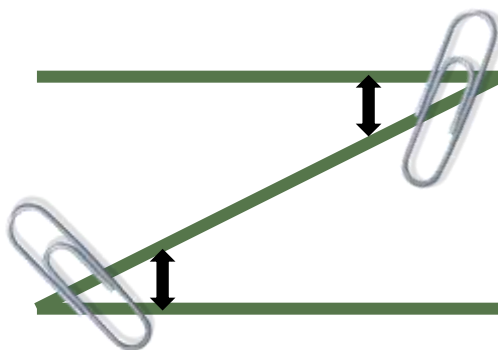
1. Begin with a dollar bill.



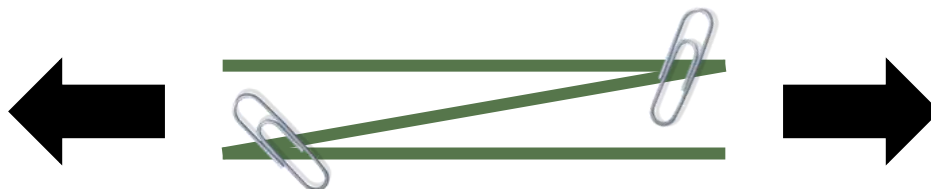
2. Fold the dollar bill into thirds, so it forms a “Z” shape. This is the necessary conformation of the enzyme.



1. Attach paper clips to the outer end and opposing inside fold. Do not 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





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