Temple Street
CHILDREN'S UNIVERSITY HOSPITAL

Metabolic.ie

National Centre for Inherited Metabolic Disorders

Phenylketonuria

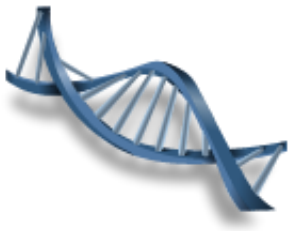
Contents

Introduction

The aim of this guide is to provide information for individuals with PKU or for parents/carers with a child who has PKU. We hope you find it useful. The information in this handbook is correct at the time of printing and the recommendations made are based on current knowledge. We recommend you use this handbook only under full dietary and medical supervision.

Please find further information on our website www.metabolic.ie

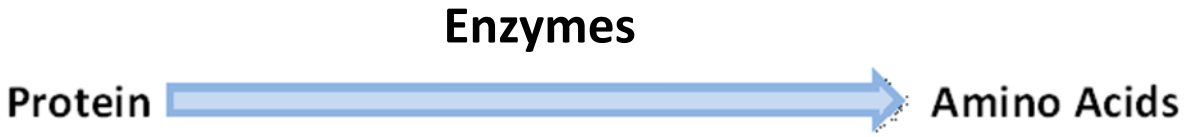
Chapter 1
What is PKU…?

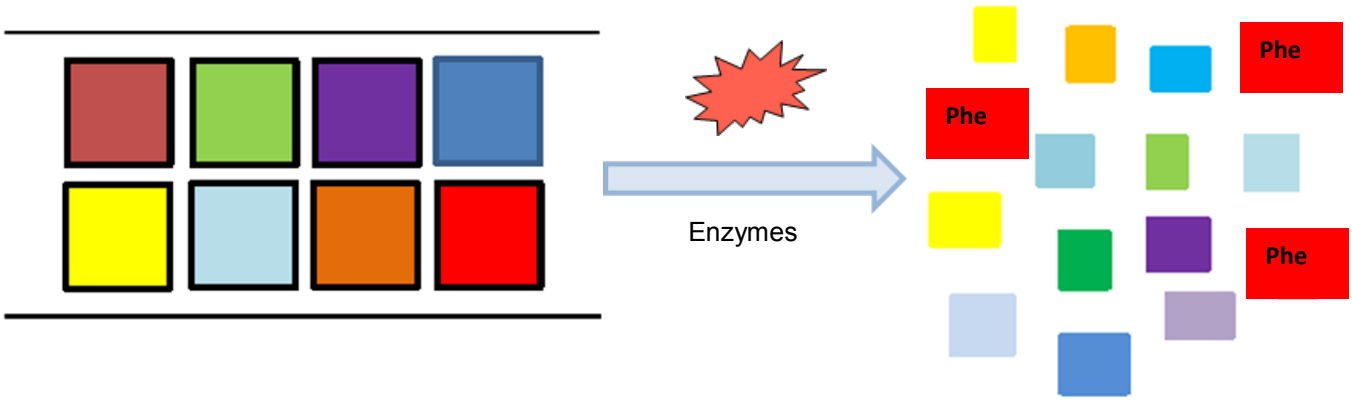


Phenylketonuria (PKU) is an inherited metabolic disorder which affects the breakdown of protein containing foods.

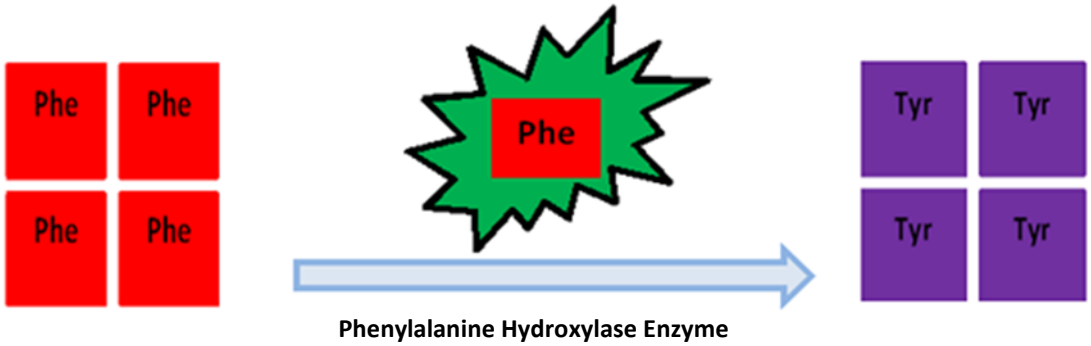
Protein is one of the main nutrients in our diet. It is made up of 20 different building blocks called **amino acids**. When protein is eaten it is broken down by **enzymes** into **amino acids.** These amino acids carry out a wide variety of functions within the body including growth, repairing cells and tissues as well as making other body proteins.

Phenylalanine (Phe) is one of these amino acids. It is used to make another amino acid called tyrosine. People with PKU are born without or with very little of the **enzyme** phenylalanine hydroxylase, which is used to change Phe to tyrosine (Tyr). This causes Phe to be high and the tyrosine to be low in the blood. If left untreated over a period of time, this can cause damage to the brain. Some of the other effects of persistent high Phe levels include behavioural problems, hyperactivity, irritability, restlessness and poor concentration.

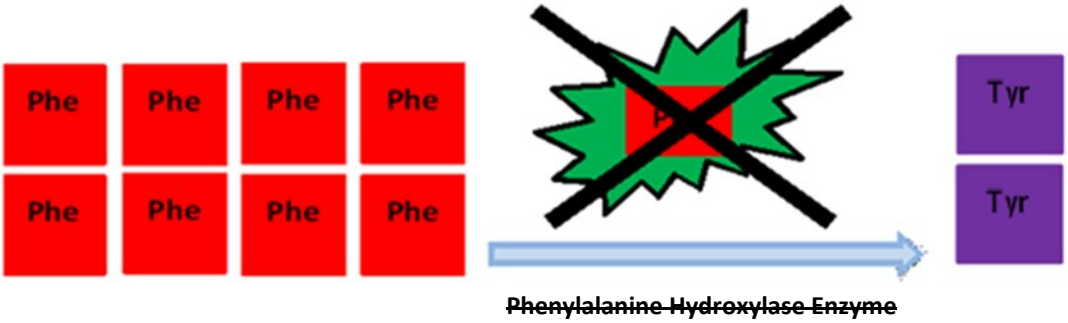




In a person **without PKU**, Phe is converted into another amino acid called tyrosine.



In a person **with PKU,** the conversion of Phe into tyrosine does not happen as it should, causing Phe levels to rise and tyrosine levels to be low.



How is PKU treated?

The main aim of treatment is to ensure acceptable levels of Phe and tyrosine and to provide adequate protein to support normal growth and development. **The treatment of PKU is a low protein diet for life**, **in addition to a Phe-free supplement** (synthetic protein). The synthetic protein contains other amino acids including tyrosine, vitamins and minerals your child may need.

An essential part of the treatment of PKU is to take small blood samples regularly to measure Phe levels in the blood. Tyrosine levels are also checked during these blood tests. These bloods are sent to the Metabolic Lab in Temple Street. **We aim to keep blood Phe levels between 120-360 µmol/l until 12 years of age and 120-600 µmol/l for those over 12 years**. Blood levels need to be sent weekly until 4 years of age.

PKU is a treatable disorder. When detected and treated early, children with PKU are able to reach their full potential. Provided your child's PKU is well controlled, they have every chance of attending university, building a successful career and enjoying a happy family life just like everyone else.

What is Kuvan®?

Kuvan® is a prescription medication that is used to treat people of all ages with PKU. The PAH enzyme is missing or depleted in PKU which causes the Phe levels to build up in the blood. This enzyme converts Phe into tyrosine in the presence of tetrahydrobiopterin (BH4). BH4 is a natural substance found in the body that helps the PAH enzyme reduce Phe to safe levels in the blood. Kuvan is a pharmaceutical version of BH4. Kuvan® works by enhancing the residual enzyme in the liver and hence lowering the phe levels. It is effective in **some,** but not all, patients with PKU.

Kuvan Trial

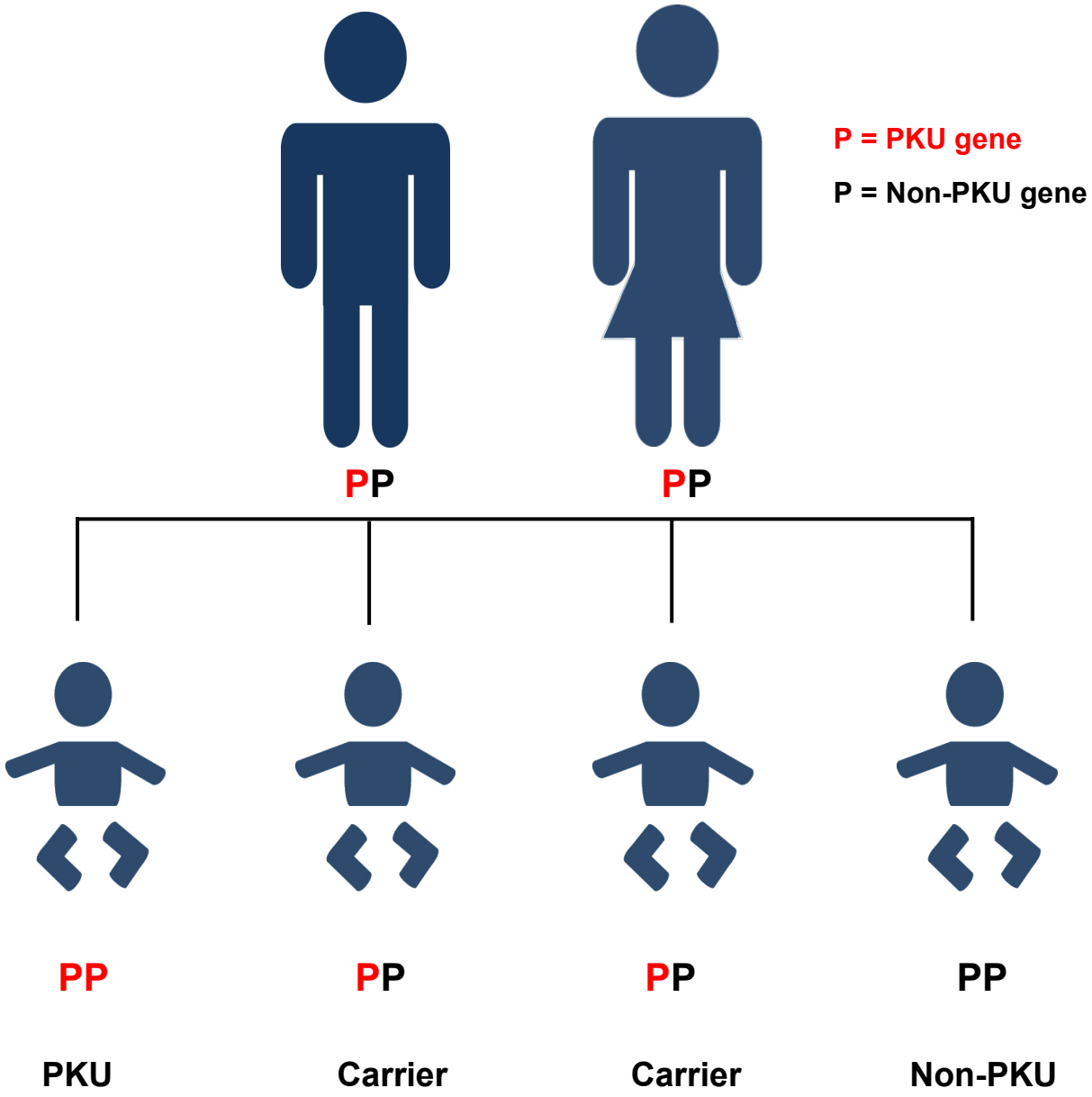
To assess your child's responsiveness to Kuvan, they will be given a 24 hour trial of the drug. If their blood Phe levels drop by 30% or more during this time period, they will be considered to have partially responded/responded to Kuvan.

Diet and Kuvan

This will mean that they may not require as much of a restriction of dietary protein compared to a child who does not respond to Kuvan. The degree of protein restriction will be determined by ongoing blood Phe monitoring. Your child may also need to take synthetic protein. This will also be determined by their natural protein tolerance and blood Phe levels within the recommended target ranges.

How is PKU diagnosed and what causes it?

All babies born in Ireland are screened for PKU by the heel prick test shortly after birth. Around one in 4,500 babies born in Ireland have PKU. PKU is an inherited metabolic disorder. Children inherit two sets of genes, one from each parent. As a parent of a child with PKU, you have one PKU gene and one non-PKU gene. This is known as being a carrier. People who are carriers for PKU do not have PKU themselves. Your child has inherited two PKU genes, one from their mother and one from their father. This means that your child has PKU. **It is important to remember PKU is not something you could have avoided and there is nothing you could have done to prevent this.** There is a 1 in 4 chance that another child born to the same parents will have PKU.


P = PKU gene
P = Non-PKU gene
PP
PP
PP
PP
PP
PP
PKU
Carrier
Carrier
Non-PKU

Chapter 2
Overview of Treatment

The National Centre for Inherited Metabolic Disorders (NCIMD) is where all children diagnosed with PKU are treated in the Republic of Ireland. At present, PKU is treated by diet. PKU should not restrict your child or your family from any normal activities, for example going on holidays, eating out or playing sport.

Successful treatment of PKU includes the following:

Foods to avoid: High protein foods such as meat, fish, chicken, eggs, milk, cheese, yogurts, soya, nuts, bread, pasta and chocolate are generally too high in Phe and are not allowed in the diet. Foods and drinks containing a sweetener called aspartame must also be avoided.

Natural protein/Exchanges: People with PKU still require a small amount of Phe for growth and development; this is what we call their natural protein (exchanges). An exchange is the amount of an allowed food which contains 1 gram of protein. The amount of exchanges allowed will change depending on your child's blood results. Typical exchange foods that are used include measured amounts of potato, beans and cereals.

Daily intake of synthetic protein: This contains amino acids (except Phe) and vitamins and minerals. It can be given in the form of a formula, drink or gel. The synthetic protein should be taken several times throughout the day. The amount required will be calculated specifically for your child's needs.

Protein free foods: Some foods are naturally low in protein and are allowed freely in your child's diet. Examples include most fruit and vegetables, butter/margarine and jam/ honey. There are also a number of specially manufactured low protein foods such as bread, pasta and milk which are available on prescription and can be ordered from your local pharmacy.

Blood levels: Regular monitoring of your child's blood Phe is an essential part of the overall management of PKU. Your metabolic team will let you know how often blood levels need to be done. Depending on your child's age, this can vary from weekly when your child is younger to monthly as they get older.

Keeping supplies up-to-date: It is very important to keep an up-to-date supply of the synthetic protein, low protein foods and supplies for doing bloods.

Clinic visits: When your child is first diagnosed you will attend the NCIMD regularly for information and support. During clinic, you will meet different members of the metabolic team, for example, the dietitian, nurse, doctor, social worker and psychologist. Visits can vary from 6 weekly to 6 monthly. As your child grows older the time between clinic visits will increase, depending on their needs.

Ask yourself:

Do I have enough…..?

- Synthetic Protein ☑
- Low protein foods ☑
- Supplies to do my bloods ☑

Chapter 3
Feeding Your Baby

The first few days

Babies born with PKU will have high Phe levels initially. **The period of time between birth and finding out your child has PKU is too short to cause them any problems**. To reduce Phe levels, your baby will be given a Phe-free formula (PKU infant formula) instead of breastfeeds or standard infant formula for the first few days after diagnosis. This will allow the Phe blood level to come down quickly. Babies usually make the change to the new formula quite easily. Most babies quickly learn to cope with the combination of bottle and breastfeeding or the two different formulas. These should always be given separately and not mixed together.

Breastfeeding

Breast milk is the ideal food for babies. It contains all the nutrients needed for growth and health. It also helps protect babies from bacterial and viral infections.

You can breastfeed your baby and keep the blood Phe at an acceptable level. Breast milk contains less Phe than standard infant formula. However, breast milk **alone** contains too much Phe for babies with PKU. Therefore, PKU infant formula needs to be given in addition to breast milk. Initially, you will need to express regularly to keep up your supply of breast milk. This is only for a few days until breast feeds can be reintroduced. You will be advised by the metabolic team on how much PKU infant formula to give. After the PKU infant formula, alternate the breast you offer first. If your baby is not interested in the second breast, do not worry, offer this one first at the next breastfeed. A measured amount of PKU infant formula is given by bottle first to reduce the amount of Phe taken from breast milk.

A measured amount of PKU infant formula is given first and then breastfeed to appetite.

If your baby is hungry in between feeds you may give extra breastfeeds in addition to the measured amounts of PKU infant formula.

While breastfeeding, you do not need to alter your diet in any way because your baby has PKU. However, the following points may help you during this time:

- Make sure you eat and drink enough to keep up your energy and milk supply
- Try to eat healthy snacks if you can't manage full meals
- Limit your tea, coffee and fizzy drink intake
- Breastfeeding mothers need to drink an extra three to four glasses of fluid a day – water is best for quenching thirst
- Your milk supply will adjust to your baby's feeding demands

How to express breast milk

While your baby is not breastfeeding (during the first two to three days after diagnosis and any other time breastfeeding is interrupted such as illness), you will need to express milk to keep up your supply.

You can express your milk by hand or with a breast pump – using an electric or hand pump. You will need to express approximately every three hours to maintain a good supply of milk.

Please ask your dietitian for a Temple Street Children's University Hospital booklet – **'Information for Parents on Expressing Breast Milk'**. If you have any questions about breastfeeding, go to www.breastfeeding.ie.

Stopping breastfeeding

If you wish to stop breastfeeding, it is best to plan it with the metabolic dietitians and to stop over a two to three week period.

Formula feeding

If you choose not to, or are unable to breastfeed, standard infant formula is an appropriate alternative, along with a PKU infant formula. A measured amount of standard infant formula is given first and then PKU infant formula to appetite.

A measured amount of standard infant formula is given first and then PKU infant formula to appetite.

If your baby is hungry in between feeds you may give extra PKU infant formula in addition to the measured amounts of standard infant formula.

PKU infant formula (synthetic protein)

PKU infant formula contains all the nutrients needed for growth, except phenylalanine. This will be prescribed for your baby.

The amount of PKU infant formula and breast milk or standard infant formula will need to be adjusted from time to time to provide the right amount of protein to meet your baby's needs and keep their blood levels in an acceptable range. Your dietitian will advise you on this as your baby grows.

Unless your dietitian advises you to do so, it is not necessary to add anything to your baby's formula.

Preparing formula (standard infant formula or PKU infant formula)

- Boil fresh water
- Leave to cool for 30 minutes
- Clean surfaces and wash hands
- Pour the required volume of water into a sterile bottle
- Add the appropriate number of scoops using the scoop provided (1 scoop per 30ml/1oz)
- If your baby needs 45ml of formula, add 2 scoops of powder to 60ml/2oz of cooled boiled water and then pour out the extra formula until 45ml is left in the bottle
- Shake well
- Cool quickly under cold running water
- Check temperature
- Throw away unused feed after 2 hours

To store the formula

- Make up each bottle as required. It is safest to prepare a fresh feed each time you need one and to give it to your baby straight away. This is because warm milk provides ideal conditions for bacteria to grow – especially at room temperature.
- If you need to prepare feeds in advance to use later, make up individual bottles, cool them quickly and place in the back of the fridge (5ºC or below). Throw away any feed in the fridge that you have not used within 24 hours.

For additional information please ask your dietitian for the booklet **'How to prepare your baby's bottle feed'** produced by Safefood (www.safefood.eu) and the HSE (www.hse.ie).

Additional information for feeding your baby

Most babies, whether they are breastfed or bottlefed, need five or more feeds every 24 hours until they are four to five months old.

- Provided your baby is well, only the PKU infant formula and breastfeeds or standard infant formula are needed until they are around 17 to 26 weeks of age.
- Babies should not be given herbal or medicinal teas or baby juice. Extra vitamins and minerals are not needed **except for vitamin D**.

Vitamin D and your baby

Whether you choose to breastfeed or formula feed, you should give your baby 5 micrograms (5µg) of vitamin D3 every day, for the first year of life. There are a number of suitable infant vitamin D3 products available to buy in Ireland. The number of drops or amount of liquid required daily is different for each product. Read the product instructions carefully and ask your pharmacist for advice if needed. **Only one dose per day should be given.**

Recording Your Babies Intake

Below is an example of a **guide** of a feeding/daily record plan which may be helpful for you to record your baby's feeding pattern.

For breastfed babies:

Feeds Date……..

Time	Number of measured feed	PKU infant formula **(measured)**	Breastfeed **(to appetite)**
7am	Feed 1	30ml	Right side
10.30am	Feed 2	30ml	Left side
1.30pm	Feed 3	30ml	Right side
2.30pm			Left side
4pm	Feed 4	30ml	Right side
7pm	Feed 5	30ml	Left side
10.30pm	Feed 6	30ml	Right side
1.00am	Feed 7	30ml	Left side
4am	Feed 8	30ml	Right side
5am			Left side

For formula-fed babies:

Feeds Date……..

Time	Number of measured feed	Standard infant formula **(measured)** e.g. Aptamil infant milk	PKU infant formula **(to appetite)**
7am	Feed 1	20ml	60ml
10.30am	Feed 2	20ml	20ml
1.30pm	Feed 3	20ml	30ml
2.30pm			60ml
4pm	Feed 4	20ml	40ml
7pm	Feed 5	20ml	60ml
10.30pm	Feed 6	20ml	10ml
1.00am	Feed 7	20ml	30ml
4am	Feed 8	20ml	50ml
5am			30ml

For breastfed babies:

Feeds Date……..

Time	Number of measured feed	PKU infant formula **(measured)**	Breastfeed **(to appetite)**

For formula-fed babies:

Feeds Date……..

Time	Number of measured feed	Standard infant formula **(measured)**	PKU infant formula **(to appetite)**

Chapter 4
Understanding the Synthetic Protein

Synthetic protein is a specially made protein that contains all amino acids except Phe that your child needs to grow, build and repair tissues. The synthetic protein contains tyrosine which is important for the body. The synthetic protein will also contain all of the vitamins and minerals that your child needs for good health.

The synthetic protein is an essential part of the treatment of PKU. If it is not taken as recommended, Phe levels will rise, which can cause health and growth problems.

The amount of synthetic protein needed by your child will be calculated by your dietitian. This will change throughout your child's life depending on their age, growth and Phe levels.

There is a variety of synthetic protein available in different forms and flavours to suit lifestyles and taste preferences. For example, infant formula for babies, gels/pastes, powders and ready made liquids for older children and adults. These are available on prescription through your local pharmacy. Your dietitian will help you find the right product for your child. The synthetic protein must be taken evenly throughout the day. This helps keep Phe levels within an acceptable range. If your child is struggling to take all of their synthetic protein, please inform your dietitian**. As PKU requires a diet for life, this means that the synthetic protein must also be taken for life.**

What to do if your child is refusing or struggling with their synthetic protein?

- Don't panic
- Contact the metabolic team to explore methods of getting your child to take their synthetic protein
- There are a variety of synthetic protein options which may be more suitable to your child's taste
- **Never mix** the synthetic protein with food. It is very important for your child to accept the taste of the synthetic protein in its true form
- **Never force** your child to take the synthetic protein

If you experience any problems with supply or quality of the synthetic protein, please contact the companies directly. Contact numbers for the companies will be provided.

Chapter 5
Dietary Management

At present, the dietary treatment for PKU is a low protein diet. **This is a diet for life**. The diet prevents a build-up of excess Phe in your child's blood, while at the same time providing enough protein for their normal growth and development. There are four main components of the diet:

1. Synthetic protein (see chapter 4)
2. Foods to avoid
3. Exchange foods
4. Free foods

The PKU diet must be low in Phe and therefore, natural protein must be restricted. The diet is best described by a traffic light system.

RED STOP:

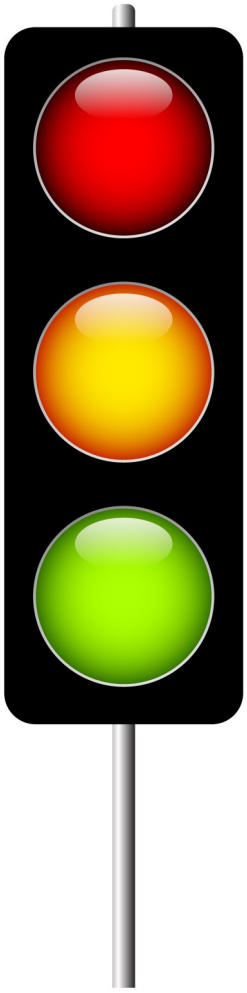
Do not eat these foods as they are too high in natural protein (Phe).

ORANGE GO CAUTIOUSLY:

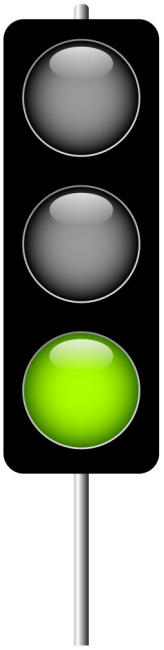
Foods that need to be counted as they contain some natural protein (exchange foods).

GREEN GO:

These foods contain very little natural protein and can be eaten freely.



GREEN GO: These foods can be eaten freely.



These foods are either naturally low in protein or have been specially made to be low in protein and so are 'free from exchanges' or do not have to be measured.

Some foods are naturally low in protein such as most fruit and vegetables, butter, margarine, cooking oils, jam, sugar and honey **(honey is not recommended for any infant under 12 months).** You can cook in all the usual ways (bake, grill, fry) and you can use herbs, spices and flavourings to add interest to foods.

There is a large variety of specially manufactured low protein foods such as bread, pizza bases, pasta, rice, flour, cereals, milk, meat substitutes, cake mixes, biscuits and chocolate. These foods are available on prescription from your local pharmacy. An up-to-date list of these foods will be given to you. All of the above foods should **make up the majority of your child's diet** and they do not need to be counted. There are many recipe books available using these low protein foods. Children with PKU are encouraged not to have too much sugar and fat for the same reasons as other children without PKU.

The following tables give examples of foods that are naturally low in Phe and so are allowed freely in the diet.

Vegetables-fresh, tinned, frozen

Artichokes	Cucumber	Peppers
Asparagus	Curly kale	Pumpkin
Aubergine	Fennel	Radish
Beansprouts	French beans	Rocket
Beetroot	Garlic	Runner beans
Butternut squash	Gherkin	Samphire
Cabbage	Green beans	Scallions
Capers	Leeks	Shallots
Carrots	Lettuce	Spring onion
Cassava	Mangetout	Sugar snap peas
Celeriac	Okra	Swede
Celery	Onions	Tomato
Chayote	Pak choi	Turnip
Courgette	Parsnips	Water chestnuts

Fruit-fresh, tinned, frozen

Apple	Grapes	Passionfruit
Apricots	Guava	Peaches
Blackberries	Jackfruit	Pears
Blackcurrants	Kiwi	Physalis
Blueberries	Kumquat	Pineapple
Cherries	Lemons	Plums
Clementine	Limes	Prunes
Cranberries	Lychees	Raisins
Currants	Mandarins	Raspberries
Damsons	Mangoes	Rhubarb
Dates	Mangosteens	Satsumas
Dragon Fruit	Melon -cantaloupe	Star fruit
Figs	Melon -honeydew	Strawberries
Ginger	Nectarines	Sultanas
Gooseberries	Olives	Tangerines
Grapefruit	Papaya	Watermelon

For the following fruit and vegetables, if the portion identified below is taken **once a day,** this portion can be counted **freely**. However, if **more than this portion** is taken, any additional portions of this fruit or vegetable needs to be counted as **1 exchange.**

For example, 5 mushrooms in the day can be eaten freely, but if 10 mushrooms are eaten, then the additional 5 mushrooms should be counted as 1 exchange. Also, 80g of sweet potato in the day can be taken freely, but if 200g of sweet potato is taken, then the additional 120g should be counted as 1 ½ exchanges.

Fruit and vegetables to be taken in limited amounts

Bananas: 1 banana	Plaintain: 1 plaintain
Broccoli: 8 florets (85g)	Pomegranate: 1 pomegranate
Brussel sprouts: 6 sprouts (90g)	Sharon fruit: 1 sharon fruit
Cauliflower: 9 small florets (90g)	Spinach: 45g (1 blue scoop)
Mushrooms: 5 medium mushrooms (80g)	Sweet potato: 80g (1 ½ blue scoops)
Oranges: 1 orange	Yam: 100g = 1 exchange

Sugar	Sweets	Jams and preserves
Barley sugar	Any chewy sweets*	Chutneys
Brown	Boiled sweets	Golden syrup
Castor	Candyfloss	Honey
Glucose	Iced lollies	Jam-any type
Icing sugar	Lollipops	Marmalade
Jam sugar	Sorbet	Maple syrup
White	Sherbet	Treacle
	Suitable jellies*	
	Other suitable sweets from treats list	

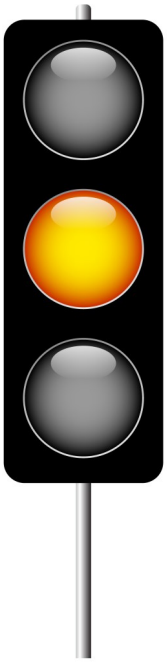
*always check the foods listed above for gelatine

Seasonings	Miscellaneous	Food essences and colourings
Chilli	Baking powder	Almond essence
Curry paste	Bicarbonate of soda	Cochineal
Curry powder	Buttercream icing	Peppermint
French dressing	Cornflour	Vanilla
Herbs	Cream of tartar	
Mint sauce and jelly	Pasta, noodles or rice made from konjac (Asian root vegetable)	
Mustard	Sprinkles	
Pepper	Some custard powders	
Salt	Sago	
Spices	Tapioca	
Vinegar		

Fats and oils	Drinks	Sauces
Butter	Some coconut milks	Apple sauce
Coconut oil	Hemp milk	Balsamic vinegar
Duck Fat	High energy drinks	Brown sauce
Fry light spray	Minerals	Cranberry sauce
Grapeseed oil	Pure fruit juice	Chutney
Garlic oil	Rice milk (not suitable in children under 5 years or pregnant/ lactating women)	Horseradish sauce
Goose fat	Squashes/cordial*	Ketchup
Lard	Tea/coffee/herbal tea	Most mayonnaises, salad creams
Margarine	Water/ soda water/mineral water	Mustard
Olive oil		Pasta sauces, cook-in sauces
Rapeseed oil		Piri Piri sauce
Suitable low fat spread	See chapter 13 for suitable alcoholic drinks	Relish
Sunflower oil		Salad dressing
Sesame oil		Soya sauce (check label)
Vegetable oil		Sweet chilli sauce
		Tomato salsa
		Tabasco sauce

* Remember to check for aspartame

ORANGE GO CAUTIOUSLY: These foods can be eaten in measured amounts and are known as exchanges.



Your child requires a small amount of Phe for growth, development and repair. Foods such as cereals, rice, beans, peas and potatoes contain a small amount of protein and Phe. These are given in small **measured quantities** called exchanges. Every child will have a set number of exchanges per day. The number of exchanges your child is allowed will change depending on their Phe levels. This is often referred to as natural protein. Your dietitian will educate you on counting exchanges and give you a booklet containing commonly used exchange foods.

An exchange is the amount of any food that contains 1 gram of protein.

1 exchange = 1 gram (g) protein

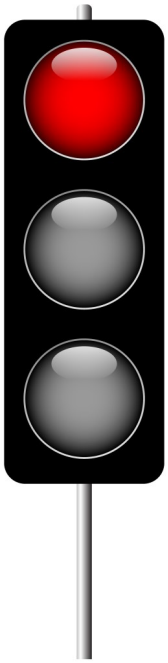
Examples of foods that contain 1 exchange include:

- 1 ½ level blue scoop of mashed potato
- 2 tablespoons of peas
- 1 level blue scoop of sweetcorn
- 1 blue scoop of boiled rice
- 1 tablespoon of baked beans
- 1 tablespoon of chickpeas

These foods are spread out between the day's meals to provide small amounts of essential Phe. It is important to be able to read food labels so that you can count exchanges for your child. This will be discussed in detail later in this chapter.

RED STOP: Do not eat these foods.

These foods are high in protein and Phe and so are not allowed.



Meat: Beef, lamb, pork, ham, bacon, chicken, turkey, duck and game.

Offal: Liver, tongue and kidney.

Processed meat: Sausages, rashers, pudding, corned beef, chorizo, meat products e.g. burgers, meat paste, meat pate and meat pies.

Fish: All kinds including shellfish, frozen or tinned. Also processed fish such as fish fingers and fish cakes.

Eggs: All varieties.

Milk and milk products: Milk (including cows, goats, soya), yogurt, yogurt drinks, cream, ice cream and chocolate.

Cheese: All varieties (*some vegan cheese and cheese spreads may be suitable*).

Soya: Foods made from soya such as TVP, Quorn and Tofu.

Vegetarian products: Vegetarian meals, burgers and sausages.

Nuts and seeds: Including peanut butter and nut spreads.

Bread: White and brown bread, soda, pitta, naan, bagels, ciabatta, wraps, croissants and scones.

Flour based foods: Pasta, cakes and biscuits.

Gelatine: Jelly, jellies and sweets containing gelatine.

Aspartame (E951/E962) is an artificial sweetener that contains Phe. It is therefore **not allowed in the diet.** Many foods and drinks contain aspartame, particularly fizzy drinks, cordials, alcoholic drinks, puddings, crisps and chewing gums. It can be found in some table top sweeteners. All other table top sweeteners are suitable e.g. saccharin and sucralose. It is important to double check all labels on a regular basis as ingredients can change.

For every food that your child cannot have, there are suitable low protein alternatives available.

Eating the wrong foods will not make your child immediately sick but will cause their Phe levels to go high.

Giving unsuitable foods can result in the child developing a taste for these foods and make the diet more difficult for the child to stick to.

Label Reading

Reading food labels plays an important role in helping you decide whether a food is suitable or not. Reading food labels can be confusing but you will have support from the dietitian to help you with this. Once you know how to read food labels, this can increase the variety of foods in your child's diet. It is important to check labels regularly as ingredients and protein content can change.

Tips on Reading Food Labels:

- Firstly look at the ingredients to see what the product contains.
- Sometimes, the nutritional information states that some protein is present within the product. However, if you look at the ingredients you may see that all the ingredients present in that product are foods allowed freely. In this situation, the product can be given freely e.g. tinned tomatoes (4g of protein in a can of tinned tomatoes according to the nutritional information). *This product is not counted because tomato is a free food.*
- If the product contains exchange containing foods e.g. rice, potato, peas or cream, then the protein should be counted as described in the nutrition label. For example, a jar of curry sauce contains cream, so therefore it must be counted.
- Another example of this would be a soup which contains peas. This product contains peas which is an exchange food; therefore, the protein content needs to be counted as described in the label. For example, if there is 1.0g of protein per carton of soup then this product needs to be counted as 1 exchange.

Remember to look out for **Aspartame** (E951/E962).
It is not permitted as in contains a source of Phe.

Some foods or drinks will already have the protein content per portion written on the food label. Use the guide below to help you.

Protein content per portion	Number of Exchanges
0 – 0.3g	Free
0.4g – 0.7g	½
0.8g – 1.2g	1
1.3g – 1.7g	1 ½
1.8g – 2.2g	2

However, this is not always the case. Sometimes, you will have to calculate the protein content per portion yourself.

To do this you will need to know two things:

1. The weight/amount of the food to be eaten
2. The protein content per 100g of the food

This is how it is done:

$$\frac{\text{Weight of food to be eaten} \times \text{Protein content per 100g}}{100}$$

Example 1: Tinned Ratatouille Provençale

390g carton

Ingredients: Tomato, Courgette, Tomato Purêe, Aubergine, Peppers, Onion, Vegetable Oil, Sugar, Water, Salt, Pepper Extract, Garlic Extract, Thyme Extract, Lemon Concentrate

Nutritional Information

Typical Values	Per 100g	Per 195g (½ carton)
Energy (kJ)	179	350
Energy (kcal)	43	85
Fat	2.0	3.9
(of which saturates)	0.3	0.6
Carbohydrate	4.2	8.2
(of which sugars)	3.0	5.9
Fibre	1.9	3.7
Protein	1.1	2.1
Salt	0.5	1.0

Step 1: Look at the ingredients. All the ingredients are ‘free’ foods so you do not need to use the nutritional label to check the protein content.

Example 2: Tomato Pasta Sauce

150g carton

Ingredients: Tomatoes, **Cream**, Lemon Juice, **Mascarpone Cheese**, Cornflour, Sugar, Tomato Paste, Salt, Fat Powder, Basil, Garlic, Spices, Herbs

Nutritional Information

Typical Values	Per 100g	Per 150g carton
Energy (kJ)	237	356
Energy (kcal)	57	86
Fat	2.6	3.9
(of which saturates)	1.7	2.6
Carbohydrate	6.6	9.9
(of which sugars)	3.8	5.7
Fibre	1.9	2.9
Protein	1.2	1.8
Salt	0.55	0.83

Step 1: Look at the ingredients. **Cream** and **mascarpone cheese** are both 'exchange'/protein containing foods. Therefore, you need to use the nutritional label to work out the protein content.

Step 2: Look at the nutritional label

1 carton = 1.8g protein

Therefore, 1 carton = 2 exchanges

Example 3: Garden Vegetable Soup

400g tub

Ingredients: Water, Carrot, Turnip, Onion, Celery, **Potato**, Parsnip, Leek, **Sweetcorn, Red Lentils** (2%), **Peas**, Vegetable Stock (Salt, Onions, Celery, Carrots, Turmeric, Parsley), Salt, Tomato Purée, Black Pepper

Nutritional Information

Typical Values	Per 100g
Energy (kJ)	105
Energy (kcal)	25
Fat	0.2
(of which saturates)	<0.1
Carbohydrate	7.3
(of which sugars)	2.1
Fibre	1.7
Protein	1.6
Salt	0.6

Step 1: Look at the ingredients. **Potatoes, sweetcorn, red lentils** and **peas** are all exchange foods. Therefore, you need to use the nutritional label to work out the protein content.

Step 2: Look at the nutritional label

Protein per 100g = 1.6g protein. However, ½ tub = 200g

Step 3: Use the formula to work out how many exchanges in the ½ tub.

$$\frac{\text{Protein content per 100 g (1.6g)} \times \text{Weight of product to be eaten (½ tub = 200g)}}{100}$$

200g (½ tub) = 3.2g protein

Therefore, ½ tub = 3 exchanges

Example 4: Rice Cakes

Ingredients: Wholegrain Rice (100 %)

Nutritional Information

Typical Values	Per 100g	Per cake (6.7g)
Energy (kJ)	1639	110
Energy (kcal)	387	26
Fat	3.7	0.2
(of which saturates)	0.7	<0.1
Carbohydrate	78	5.2
(of which sugars)	0.7	0.1
Fibre	3.4	0.2
Protein	8.5	0.6
Salt	0.01	<0.01

Step 1: Look at the ingredients. Rice is an exchange food. Therefore, you need to use the nutritional label to work out the protein content.

Step 2: Look at the nutritional label

1 cake = 0.6g protein = ½ exchange

2 cakes = 1.2g protein = 1 exchange

Additional Information on Reading Food Labels

For foods that are not on our exchange lists, you may need to work out the amount of food that contains 1 exchange e.g. breakfast cereals

- Weight of product that is 1 exchange:

$$\frac{1 \times 100}{\text{Protein content per 100g}}$$

- Weight of product for your required number of exchanges:

$$\frac{\text{No. of Exchanges} \times 100}{\text{Protein content per 100g}}$$

The following example shows you how to use this information.

Example 5: Oat Flakes

Ingredients: Wholegrain **Oats** (84%), Maltodextrin, Sugar, Malted **Barley** Extract, Potassium Chloride, Salt, Niacin, Iron, Pantothenic Acid (B5), Thiamin (B1), Riboflavin (B2), Vitamin B6, Folic Acid, Vitamin B12

Nutritional Information

Typical Values	Per 100g
Energy (kJ)	1618
Energy (kcal)	387
Fat	5.5
(of which saturates)	1.1
Carbohydrate	73
(of which sugars)	1.3
Fibre	6.1
Protein	11.1
Salt	trace

Step 1: Look at the ingredients. Oats are an exchange food. Therefore, you need to use the nutritional label to work out the protein content.

Step 2: Look at the nutritional label

Protein per 100g = 11.1g protein

Step 3: Use the formula to work out how many grams of these oat flakes are 1 exchange.

$$\frac{1 \times 100}{11.1} = 9\text{g of oatflakes for 1 exchange}$$

$$\frac{2 \times 100}{11.1} = 18\text{g of oatflakes for 2 exchanges}$$

Preparing a low protein meal & a family meal at the same time

Below are some tips on how to make the low protein meal similar to family meals. It is important that your child feels included in the family meal. They should eat at the same time as the rest of the family. Try to include them in the cooking as often as possible.

Family Meal	Low Protein Meal
Roast meat with roast vegetables and gravy	Roast vegetables served with: • Gravy and potato (count exchanges) • Low protein stuffing • Low protein burger/ sausage mix (count exchanges) • Low protein Yorkshire pudding
Quiche or Pie	Grated vegetables (courgette, parsnip, carrot, butternut squash, sweet potato (May need to count exchanges) • Baked as a pie with herbs and white lasagne sauce using low protein pastry • With pasta sauce and a low protein bread crumb topping • Piled into a vol-au-vent shell (count exchanges)
Grilled meat and steamed vegetables/ salad	Fried or baked aubergine or courgette in low protein breadcrumbs with steamed vegetables/salad
Stir fry with rice or noodles *(cook meat separately and use the vegetable stir fry for everyone)*	Stir fry vegetables with suitable soy sauce, ginger and garlic or use a suitable stir-fry sauce Cook low protein spaghetti and toss into stir fried vegetables to resemble noodles Use low protein flat noodles, konjac noodles, rice noodles (count exchanges if applicable)

Family Meal	Low Protein Meal
Sausage, tomato sauce, mash or chips and vegetables	Use sausage/burger mix and make into sausage shape (count exchanges) with potatoes (count exchanges) or sweet potato chips (may need to count exchanges) or parsnip chips
Burger and chips	Use burger mix to make a burger shape (count exchanges) and serve between a toasted low protein roll with salad and sweet potato or potato chips/wedges (count exchanges if applicable)
Macaroni Cheese	Use white lasagne sauce, herbs and low protein macaroni pasta and vegan cheese
Pizza	Low protein bread or pizza base – spread with tomato purêe and top with vegetables and low protein cheese (count exchanges if applicable)
Barbeque	Vegetable skewers and baked potato (count exchanges) with coleslaw. Or make up burger mix (count exchanges) to a burger shape and serve in a low protein roll with coleslaw
Spaghetti Bolognese	Low protein spaghetti, fry off desired vegetables and tomato based bolognese sauce. Use burger mix to make meatballs if desired (count exchanges)
Curry	Use a curry paste with tinned tomatoes and tomato paste for flavour and add to fried vegetables of choice. Add a small amount of coconut milk if desired (may need to count as exchanges). Serve with low protein rice

Chapter 6
Monitoring

Monitoring blood Phe is an important part of managing PKU. Keeping blood Phe levels under control reduces the risk of problems. The ideal blood range for Phe is 120-360µmol/l for children under 12 years and 120-600µmol/l for those 12 years and older.

PKU requires frequent bloods to monitor the effectiveness of treatment. The results of these bloods are used to adjust the diet as required. Additional bloods may be required depending on blood control and other factors such as pregnancy. The metabolic team will decide on this and inform you what is necessary at a given time.

Blood Phe is measured using a small blood sample taken from the heel of babies or from the fingertip of children. In the first few days after your baby is diagnosed, you will be taught how to collect these blood samples. Nursing staff will take the blood samples until you feel confident enough to do it yourself. Grandparents and other carers may also be taught how to take blood. Once your baby is at home, you will need to take these blood samples and send them to the Metabolic Laboratory in Temple Street for analysis. You will then need to contact the metabolic dietitians to obtain the results roughly two working days later. The metabolic nurses will provide you with cards and other supplies needed for sending the blood samples.

If Phe levels are too high or too low, your metabolic dietitian will advise you appropriately on changes to make. A number of factors cause blood Phe levels to increase or decrease outside of the target range.

Phe levels may increase when:

- Phe intake (exchanges) is too high
- Intake of synthetic protein is too low which causes the body to break down its own muscle tissue for nutrients. This releases Phe into the blood
- A child's rate of growth has slowed meaning less Phe (exchanges) is being used to build new proteins
- Illness, vaccinations and teething may make it difficult to eat or take the synthetic protein. This can cause higher Phe levels from the breakdown of the body's own muscle tissue
- Insufficient calorie intake results in the body breaking down its own muscle tissue for calories and causes the release of Phe into the blood
- Accidental intake of products containing aspartame

Phe levels may decrease when:

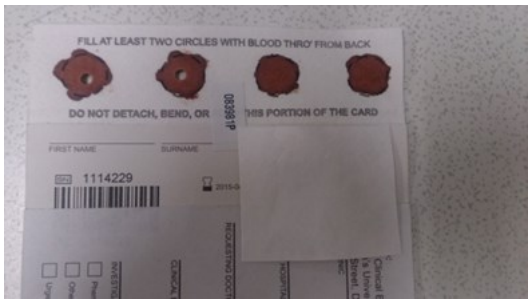
- Intake of Phe (exchanges) is lower than recommended
- Growth in a child causes levels to fall as Phe is used to build new body tissues
- Sometimes following illness when the body is rebuilding body protein

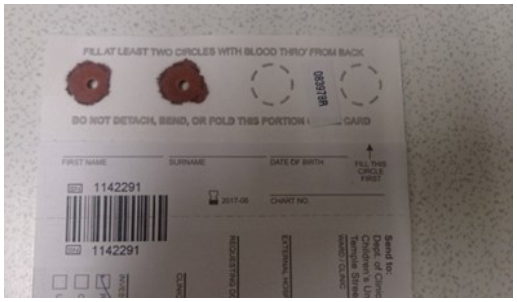
It is really important to inform your dietitian of any issues that may have arisen in your child during the days prior to levels being taken e.g. over exchanging, illness, etc.

Steps to taking blood Phe samples

1. Check the expiry date on screening card and if this is in date proceed to tear off the top sheet of the screening card; affix the patient name labels as previously demonstrated. Do not forget to write the date of the sample on the card also. If the date is not written on the card, the blood sample cannot be analysed.
2. Ensure your child's heel/toe (if walking)/finger (if over 1 year) is warm before proceeding to obtain the blood sample with your lancing device.
3. When a blood droplet forms, gently blot the blood onto the screening card within the circles outlined. Blood should be blotted onto the card from the back (as indicated on the card). **Two** full circles are sufficient so there is no need to fill all four circles. It is important to check that the blood has soaked through to the other side.
4. Allow the blood spots to dry completely before placing the card in the special envelope. It is important to attach the pre-addressed label to the outside of the envelope as well as a stamp otherwise the sample will not reach the laboratory for analysis.
5. Post the sample early in the week and telephone the metabolic dietitians for a result approximately two days later. A dietitian will give you the result and inform you of any necessary dietary changes.

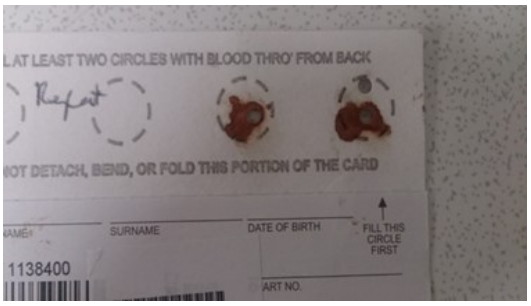
Here are some examples of **well filled** cards:



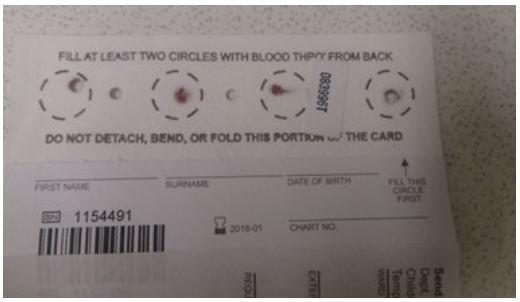


Here are some examples of **not well filled** cards that the Metabolic Lab **will not be able to analyse**:

Smudged card



Too little blood



Useful tips for doing bloods

- Keep all the equipment for blood tests together and contact the metabolic nurses when you require new supplies.
- Stock up on necessary supplies at your clinic visit.
- Have a stock of postage stamps available for the blood sample envelopes.
- Remember to dispose of the used needles/lancets in a safe manner.

NOTE: If you experience any problems taking a blood sample or need supplies of envelopes, cards or stickers, please do not hesitate to contact the nursing staff on 01 8784409 or contact the Metabolic Unit on 01 8784317.

Frequency of monitoring blood levels

How often your child's blood is monitored depends on their age and Phe levels. This is determined by the metabolic team. Some general guidelines are below:

Age	Recommended Frequency of Samples
First few weeks of life	1-2 times per week
0-4 years of age	Weekly
4-10 years of age	Fortnightly
10-16 years	Fortnightly to monthly
>16 years	Monthly
Planning pregnancy	Weekly
Pregnancy	Twice weekly

Ringing for levels

It is really important to call the metabolic dietitians for your child's results. The contact information for the dietitians is as follows:

01-8784317 and press 2 for dietitians

If the phone is not answered, please leave a message and we will return your call as soon as possible. The laboratory in Temple Street usually analyse bloods on Tuesdays and Wednesdays with results available the following day. The voicemails are checked daily Monday to Friday. To provide an efficient service please ring between the following times:

Monday, Tuesday, Thursday and Friday: 2-4pm

Wednesday and Thursday: 10.30am-12.30pm

In case of emergency, please contact the metabolic nurse or doctor on-call via switch 01-8784200.

Texting system

A texting system has been introduced by the metabolic team for sending blood results. This system is suitable for children over the age of 2 years. One parent or guardian will usually receive a text message on a Wednesday or Thursday (or depending on when the blood results are available).

The texting system serves as a reminder to contact us in the event of abnormal levels and it also means you do not have to contact us if the levels are normal. You can contact us however if you have any queries.

If the level is within range (120-360 for the under 12's and 120-600 for those 12 years and older) you will receive the following message, which means there is no need for you to contact the metabolic dietitians:

> **NCIMD: Your PHE result received on** *(date)* **for** *(hospital number)* **is** *(level)* **and is within the target range. No changes are necessary.**

If the level is high or lower than the range for your child's age group you will receive this text :

> **NCIMD: Your PHE result received on** *(date)* **for** (*hospital number)* **is not within normal range. Please Phone 01 8784317 to speak to a dietitian.**

If there was a problem with your child's sample then you get this message:

> **NCIMD: Your Phe sample for** *(hospital number)* **received on** *(date)* **was insufficient. Please send another sample. Any queries ring 01 8784317.**

We can also inform you of any delays on getting results:

> **NCIMD: Phe results are delayed this week. You will receive a text when results are available. Apologies for any inconvenience caused.**

Once a child is over 18 years old, they can start to receive the text message themselves which encourages independence with the management of their PKU. It also encourages them to start ringing for their own levels.

If for any reason you change your number or you want to change the person receiving the text, it is really important to inform the metabolic dieitians so that we can change your details. If for some reason you do not receive a text message please contact the metabolic dietitians.

Chapter 7
Weaning

Weaning is an exciting time in your baby's life when you begin to introduce some solid foods. It is recommended that weaning begins no earlier than 17 weeks (4 months) of age. This is because your baby's tummy is not ready for solid food before this. We recommend not to delay weaning beyond 26 weeks of age as there is a 'window of opportunity' to allow your baby to develop the skills to eat and accept new tastes and textures. It will also ensure your baby eats a balanced diet. Protein 'free' foods are started first.

Your dietitian will give you a copy of **'Weaning your baby on a PKU diet'** before the time comes to wean and will discuss weaning in detail with you.

This booklet can also be downloaded from:
http://metabolic.ie/patient-family-information/metabolic-conditions/phenylketonuria/

Chapter 8
Preparing for
Childcare, Pre-School & School

Preplanning

Children with PKU can easily make the transition to childcare and school with preparation. There is no reason why their condition cannot remain well controlled. There needs to be ongoing support and communication between the family, school and the metabolic team.

The following suggestions are a guide for planning and discussing your child's PKU with the childcare or school.

Discussing PKU with childcare and school

Who do you need to inform?

It is important that teachers and others at your child's childcare or school understand why your child needs a special diet and why it needs careful supervision.

You should inform the following people about your child's metabolic disorder:

- The principal and admissions staff (when you are planning your child's enrolment)
- The class teacher
- The supervisor of after school care
- The tuck-shop or canteen staff

What to discuss with staff

Firstly, they will need a basic understanding of PKU and the importance of the low protein diet.

You are probably familiar with the terminology and concepts of the condition, but explaining it simply to others is sometimes difficult.

Anyone caring for your child needs to know that:

- Your child's condition is inherited and non-contagious
- Children with this condition cannot break down protein in food
- All children need a certain amount of protein for growth and repair of the body, but in your child's condition the extra protein can cause problems
- Staying on a protein restricted diet keeps the child healthy and supports development
- Eating the wrong foods will not make them immediately sick, but may have a detrimental effect over the day or longer-term
- Your child is on a very specific diet that is calculated by a specialist metabolic team, with portions/exchanges measured out daily by the child's family – so it is important that there is a system in place at school to supervise the type and amount of food they eat during the day
- You must be informed if the child has eaten food that is not allowed or does not eat foods that are sent from home

You can request a letter or photocopy relevant sections of this handbook for the childcare or school to refer to as needed.

Food at school

Each childcare facility or school will vary in what they provide in terms of meals or canteen food.

You will need to decide whether to:

- Provide all the food your child will eat at school
- Provide the main meals but use the school canteen or preschool meals for snacks, such as fruit or salad plates – you could also give the school a list of foods that are 'free' (contain minimal or no protein) or a list of exchange foods (foods allowed in measurable quantities)
- Use only the school facilities – by either pre-ordering the selected foods from the menu, or providing low protein food for the school canteen to make into meals

Taking synthetic protein in school

Your child is likely to be happy to take the synthetic protein at the usual times at school. Label it with their name and store it in the fridge or at room temperature depending on their preference. Encourage your child to take the synthetic protein to school to help spread it throughout the day.

Parties and cooking days

Prepare for these ahead of time by keeping a supply of pre-made food which has the protein measured or low protein treats in the freezer.

If the class is learning about food or cooking, your child can share food using the low protein equivalents, so they are not excluded. Other children can also sample the food.

What to discuss with your child

Talking to your child is an important part of preparing for childcare and school.
While the school staff will do their best to supervise, you will feel more secure if your child understands and is able to manage their diet appropriately.

Things to talk to your child about include:

- Knowing which foods are free and which to count
- Bringing home uneaten food in the lunch box so that you can calculate exchanges
- Buying only suitable food from the canteen
- Not swapping lunches with friends
- Deciding when to drink the synthetic protein
- How to explain the different diet to other children
- What to do if being teased

Offer encouragement to your child by:

- Giving positive messages about the foods they can have
- Discussing this way of eating keeps them healthy and helps them to grow
- Reassuring them that other people are on a special diet so your child knows they are not the only one

Letters for childcare/school are available from the metabolic team or on our website www.metabolic.ie

Chapter 9
Encouraging Independence

When your child is very young you have the main responsibility for managing their condition. Treat your child like other children and try not to be over sympathetic or protective. Never say anything negative about the food or synthetic protein to your child. While managing PKU will be part of both you and your child's life, it will be just that: a part of life. A positive and an encouraging attitude by all the family is much more likely to lead to their acceptance of the treatment as they grow.

It is important to start sharing this responsibility with your child as they grow older. Looking after their diet needs to be incorporated into their care, as much as learning to wash their hands after using the toilet or tidying up their toys.

Fostering independence in managing their PKU has a number of advantages. It:

- Encourages **acceptance**
- Helps them develop a good **understanding** of PKU
- Increases their **confidence** in controlling their condition
- Helps you and your child **work together** better as a team

By allowing your child to take charge as they mature, some of the more difficult aspects of their treatment become a shared responsibility. When children with PKU are able to accept and take some responsibility for managing their condition in everyday life, their long-term adjustment and dietary control tend to be better. This helps them prepare for adult life when they will need to take full responsibility for their PKU. As your child grows your role will change from managing the diet to supervising it. Later, you will observe and support your child as *they* make the important decisions.

What to expect of your child at different stages

Toddlers: 2–5 years

- Are aware that they have a special diet
- Know they need to take their synthetic protein
- Know they need to have blood tests
- Know to check new foods with their parents
- Watch you prepare the synthetic protein

Toddlers: 2–5 years continued

- Start to learn about yes/no foods
- Are aware of exchange foods that need to be counted

Early childhood: 5–8 years

- Understand that they have a condition called PKU
- Understand that they cannot eat high protein foods
- Have a basic knowledge of why the synthetic protein is important
- Can assist in taking their own blood samples
- Know what is considered a high Phe level
- Help you prepare the synthetic protein
- Practice using scoops to measure foods and count exchanges
- Have ideas about meal choices
- Learn to select low protein foods with assistance in social situations

Late childhood: 8–12 years

- Have a basic understanding of PKU, Phe and their diet
- Are able to prepare their own synthetic protein
- Understand that taking synthetic protein helps provide nutrition and helps ensure good blood levels
- Can measure foods accurately and count exchanges
- Can read food labels with assistance
- Are able to make appropriate meal choices
- Are able to take their own blood samples with assistance
- Know how frequently they need to take blood samples

Early teens: 12–14 years

- Have an understanding of what is PKU, Phe, tyrosine and amino acids
- Understand the risks and effects of high Phe levels on their health
- Prepare their own synthetic protein all the time
- Take their own blood samples and know the recommended range and frequency for blood Phe levels
- Make an effort to expand low protein diet choices
- Are able to make appropriate meal choices and prepare some basic meals for themselves

Early teens: 12–14 years continued

- Are able to keep a food diary to record their protein intake
- Read and calculate exchanges from product labels without assistance. Check labels for aspartame
- For girls: Are aware of the pregnancy risks associated with maternal PKU
- Are able to engage in clinic appointments

Late teens: 14–18 years

- Have a thorough understanding of PKU and its effect on their body
- Prepare their own synthetic protein
- Can measure exchanges accurately
- Are able to make and prepare appropriate meal choices
- Remember to take their own blood samples and make phone call with parents to the metabolic dietitian for their level results
- Are able to make their own dietary changes depending on the exchanges recommended
- For girls: Are aware of the pregnancy risks associated with maternal PKU
- Know how often clinic visits are needed and take responsibility for making their own appointments
- Are able to engage in clinic appointments
- Know how to contact their metabolic team when required

Talking about PKU with your child

Talking to your child about PKU and its day-to-day management is beneficial even at an early age. The metabolic team will support you in this, answering any questions you may have, and helping you increase your child's knowledge and independence. By the time children start school or have meals at friends' houses, they need to have an understanding of their diet and the foods they are allowed to eat. Help your child to make decisions about aspects of their diet – such as taking their synthetic protein to school, or if to take food to a party or sleepover, or choose food they are allowed from what is provided.

Having PKU, they will be asked questions about their diet.

Frequently asked questions include:	Here are some possible answers:
Why do you not eat meat?	I am on a special diet. I do not mind not eating meat – I have never had it and do not miss it.
Are you vegetarian?	My diet is like a vegan diet but even stricter.
Are you sick?	No, I am not sick – I am healthy and my diet keeps me healthy.
Is it contagious?	It is no more contagious than being a vegetarian. You cannot catch PKU, you have to be born with it.
How can you drink your synthetic protein?	I have taken a synthetic protein since I was a baby, so I'm used to it. I am used to having synthetic protein. It is like medicine – I need it to be healthy.

Rehearsing answers and scenarios with your child beforehand may help them avoid being tongue-tied or embarrassed when they are confronted with questions.

Games

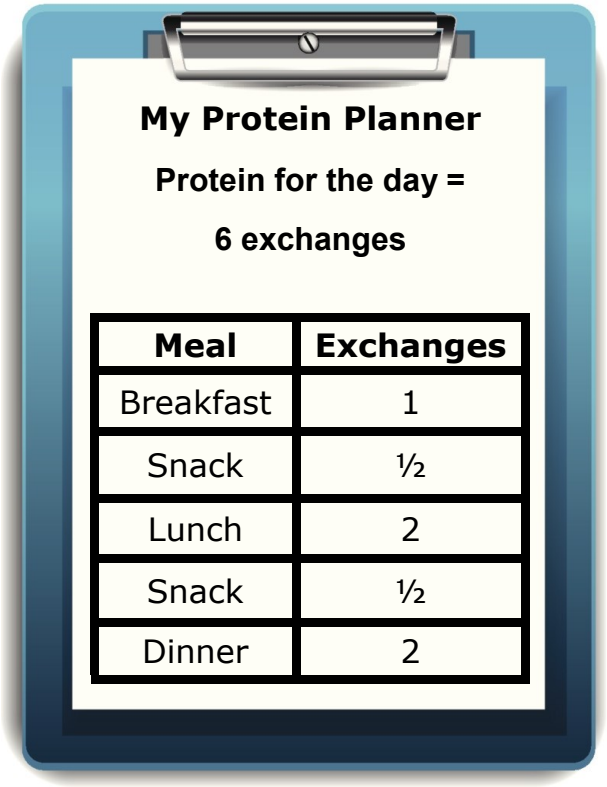
Regular discussions about PKU with your child will help them feel more comfortable talking about PKU and give them the confidence to ask you questions and share their concerns. You can help your child to understand PKU in a variety of ways from an early age. Many families use games, such as the following, or invent their own:

- Placing different foods into 'yes' and 'no' categories
- Cutting out different foods from magazines and pasting them into a PKU Scrapbook
- Letting your child rate new foods or meals from 1–5 to encourage them to try new foods
- Involve your child in cooking low protein recipes to take to school or share with the family. Children as young as 2 or 3 years of age can help you pour ingredients into a bowl and stir. School age children can help peel and cut vegetables and measure ingredients. As children get older they can plan a meal and follow a simple recipe
- A star chart/reward system for drinking their synthetic protein, managing their diet and doing their blood tests

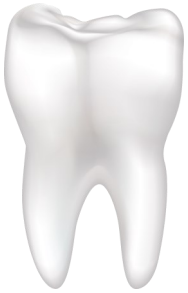
The more PKU can become just another part of life, the easier their life and yours will be.

Daily exchange planner

Why not count exchanges together? Attach a magnetic board to the wall and write in the amount of exchanges you will offer at each meal. For each exchange, place a magnet on the board. As the exchanges are eaten during the day, your child removes magnets from the board until they are all gone. Another option is to laminate a chart and stick it to the fridge. Write on it with a whiteboard marker and keep a tally that way. If your child is on limited exchanges it would be helpful to use different coloured magnets to indicate 1 exchange and ½ exchange as some foods may be measured in ½ exchanges. Beads on a string can also be used to demonstrate counting exchanges.



Chapter 10
Dental Care



Tooth decay and dental erosion can affect the teeth of any child. Children and adults with PKU are at greater risk and need to take good care of their teeth. Sugary foods and acidic drinks may be consumed more often as the diet must be low in protein and high in carbohydrate, which increases the risk of tooth decay. The synthetic protein is acidic and may be sweetened, but remains an **essential part** of your child's diet.

Suggestions for reducing dental problems include:

- Encourage your child to finish their synthetic protein in one go rather than sipping over a long period of time
- Drinking the synthetic protein with a straw may be beneficial (if applicable) as it reduces exposure of the teeth to sugar and acid
- It is best to consume synthetic protein at meal times. Give some water after each synthetic protein
- Water is the best drink to have apart from the synthetic protein. Encourage often throughout the day
- After the age of 6 months, encourage your baby to start using a beaker. The main reason for this is that bottle feeding can be harmful to teeth
- Only put infant formula and water in an infant's bottle
- Do not let your child sleep with a bottle or beaker in their mouth
- If sweets, juices or fizzy drinks are taken, it is best to take them with meals, rather than between meals
- Visit your dentist regularly, at least once every year. It might be worth discussing with your dentist preventative measures to reduce decay such as fissure sealants
- Do not use toothpaste until your child reaches 2 years of age. Use a soft toothbrush and water only once their teeth appear
- From age 2-7 years use a small pea size amount of fluoride toothpaste
- We recommend to assist your child with brushing their teeth up to the age of 8 years
- Ensure regular brushing of teeth twice per day. Brushing teeth should last for 3 minutes

See: http://www.dentalhealth.ie for further information on dental health.

Chapter 11
Illness

Children with PKU will have the same number of coughs, colds and other illnesses as a child without PKU. Illness may cause a temporary rise in your child's Phe levels, **but no long-term harm is done.** During illness, the body starts to break down its own tissues, releasing Phe into the bloodstream, which can cause Phe levels to rise. Despite illness, please ensure you still do blood levels as normal and let the metabolic team know they have been unwell.

When your baby/child is sick

Contact your GP if your baby/child is unwell just as you would if they did not have PKU. Inform the GP that they have PKU to ensure appropriate medication is given that does not contain **aspartame**. Your pharmacist will be able to guide you with this. If a medication is required urgently and contains aspartame give it until you can get an alternative or discuss with your metabolic team.

Maintaining your child's fluid intake is important during illness. Here are some practical guidelines:

Babies

- Offer feeds more frequently than usual.
- If your baby has a poor appetite and is feeding poorly from the breast you may have to express to keep up your supply.
- Give any medications recommended by your doctor.
- Encourage adequate fluid intake by offering drinks every one to two hours. Fewer wet nappies or strong coloured urine is a sign your baby may not be drinking enough.
- If your baby has gastroenteritis see your GP as it is important to avoid dehydration.

Older children and adults

- Encourage small frequent meals and regular fluids as tolerated.
- Your metabolic team may advise you to cut back on protein exchanges if Phe levels are raised. In many cases this will have happened naturally as appetite may be reduced during illness.

Older children and adults continued

- Ensure adequate fluids are taken especially if symptoms include vomiting and/or diarrhoea. It may be useful to use Dioralyte or an equivalent electrolyte replacement powder to help prevent or correct dehydration.
- Try to ensure the synthetic protein is taken. However do not force it especially if your child is vomiting. If forced, this may lead to problems at a later stage.
- It can be useful to try to dilute the synthetic protein further with water.
- It is also **very important** not to mix any medications such as antibiotics through the synthetic protein as this will alter the taste and may lead to problems at a later stage.

If your child is admitted to a local hospital, inform the metabolic team so that they can liaise with the relevant staff. Remember to bring your child's synthetic protein and low protein foods as they will not be stocked in the local hospital.

Chapter 12
Teenagers

During the teenage years, many questions may arise as your teenager becomes more independent with their diet. There is a move of responsibility of managing the diet from the parent to the young adult. This chapter looks at some of the issues that occur during the teenage years. Your teenager may want to meet others their own age with PKU. This can be arranged through the metabolic team. There are also a number of online apps available to help compliance. Diet and blood levels can be monitored on the move to fit in with your teenager's busy lifestyle. These apps can provide reminders to take their synthetic protein, do blood levels and record their exchanges.

Staying on diet

When PKU was first discovered, the low protein diet was stopped at the end of childhood as it was thought that raised Phe levels could not cause further damage to the brain and nervous system. Little was known about the benefits of continuing the treatment into adulthood. Studies show that staying on diet into adult life is beneficial. Some people with PKU who have gone off their diet and then start it again say that when they are back on diet they:

- Feel better
- Look better, especially their skin
- Can think more clearly
- Are less moody
- Find it easier to get along with others
- Feel less tired and have more energy
- Can concentrate
- Can think clearly to study and do exams
- Can complete assignments more easily
- Think it is a must for doing final year school exams
- Can work things out better, e.g. when trying to think strategically such as in team sports or playing games or at work.

We strongly advise that the PKU diet is for life.

High Phe levels may mean, among other things, that they are not able to make judgements as well as they should – such as when driving a car, operating machinery or making decisions.

Some people with PKU who are not on diet or stop taking their synthetic protein develop problems such as:

- Tremors (the shakes)
- Nervous system problems such as behaving inappropriately, having mood swings or being confused about reality – not seeing things as they really are
- Stiff or weak legs
- Headaches
- Nutritional deficiencies which can cause severe problems, such as lack of vitamin B12, iron and calcium
- Eczema, skin and hair problems

Exercise

Having PKU does not limit their ability to participate in exercise or sport. Regular physical activity is an important part of a healthy lifestyle and we encourage it. The Irish recommendations for exercise are moderate activity for at least 60 minutes every day for teenagers. For example; cycling, running, boxing, martial arts, hurling/ camogie, brisk walking, football, skate boarding, dancing, skipping, rugby, tennis, swimming and basketball.

After exercise, their body needs fluid, carbohydrate and protein to recover. Drink plenty of fluid, especially water and eat some carbohydrate foods like low protein bread or pasta. The type, intensity and length of time exercising will determine how much protein they require. Taking their synthetic protein as prescribed should provide their body with enough protein. 'Protein powders' that promise to increase muscle bulk are **not appropriate** for a person with PKU as they will increase their blood Phe levels. Ask the dietitian about suitable synthetic protein alternatives and if it is required.

Eating out and alcohol

These topics will be discussed in chapter 13.

Pregnancy

Babies born to a mother who has consistently high Phe levels during pregnancy are at a high risk of having reduced mental ability, heart defects, low birth weight, small head and brain. To prevent these problems women with PKU must plan their pregnancy and aim for low Phe levels (120-360µmol/l) before they conceive. Additional support from the metabolic team will be provided at this time. We have had many successful pregnancies in the NCIMD for women with PKU. See later chapter 15 for more information.

Chapter 13
Eating Out & Alcohol

Eating Out

Eating out with PKU can be enjoyable and easy if you take the time to prepare in advance. Many restaurants have menus online so you can view these in advance. It is a good idea to phone ahead and explain a little about the diet in order to find out if the restaurant has suitable dishes already on their menu. Some menu items may need slight modification such as leave out the cheese topping. Find out if they would be willing to cook low protein pasta, bread or pizza bases.

When eating out there are two options you can take:

1. Stick to your usual pattern of spreading your exchanges throughout the day, meaning you have only a few left for dinner.
2. Save up your exchanges for the meal if it is a special occasion, have all low protein meals during the day. **This should not be done on a regular basis.**

The following are ideas for eating out in different locations:

Barbeques

- Homemade vegetable and fruit skewers
- Vegetable kebabs marinated in garlic and honey sauce or other low protein marinades
- Salads*
- Hash browns*
- Vegetable fingers*
- Vegetable burgers*
- Corn on the cob*
- Mushrooms*
- Aubergine slices
- Dips, e.g. salsa, chutney, guacamole*

**May need to count exchanges*

Cafes

- Mixed salad with a suitable dressing and coleslaw
- Fruit salad/fruit juice
- Chips*
- Jacket potato* with low protein filling – tomato, coleslaw, sweetcorn*, guacamole* and sweet chilli sauce
- Vegetable soup (not based on milk, cream, lentils or other beans, e.g. kidney or white beans or chickpeas)
- Mashed potato* and vegetables with gravy
- Latte, cappuccino, milkshake, smoothies made on coconut milk or rice milk ***(Note: Rice milk is not suitable for children under 5 years and pregnant or lactating women)***

Italian restaurants

- Pasta** with tomato based sauces
- Garlic mushrooms* on garlic bread**
- Garlic bread**
- Bruschetta**
- Vegetarian antipasto – artichokes, olives, tomatoes, roasted peppers, tapenade
- Pizza**
- Gluten free pizza base* (check with the restaurant) with low protein cheese**
- Fruit
- Sorbet

Chinese restaurants

- Boiled rice*, plain fried rice*, noodles*
- Vegetable dishes, e.g. stir fried vegetables
- Prawn crackers*
- Vegetable spring rolls*
- Vegetable Chinese soup*

**May need to count exchanges*

***Bring your low protein product for the restaurant to prepare*

Thai/ Malaysian restaurants

- Boiled rice*, plain fried rice*, noodles*
- Vegetable dishes, e.g. green or red curry (avoid dishes with nuts and cream based sauces, e.g. panang curry)
- Fresh fruit

Indian restaurants

- Boiled rice*
- Dishes made with sago*
- Vegetable dishes without legumes (lentils, dahl, red or white beans, chickpeas) or paneer (Indian cheese)
- Mango chutney and pickle, lime pickle
- Poppadums*

Greek restaurants

- Rice*
- Dips – hummous*, baba ganoush (check ingredients)*
- Olives, cucumber, red onion and tomato in a vinaigrette
- Vegetable dishes, e.g. vegetable kebab
- Salad

Mexican restaurants

- Sweet potato wedges*
- Tortilla chips*
- Corn chips*
- Taco shells*
- Salsa
- Guacamole*
- Stir fried seasoned vegetables

**May need to count exchanges*

Alcohol

Having PKU does not stop you from drinking alcohol. The minimum age alcohol can be legally consumed in Ireland is 18 years. The current guidelines in Ireland recommend a maximum of 17 units of alcohol for men and 11 units for women per week. It is recommended to have at least two alcohol free days each week. Drinks should be spaced out over the week and not consumed in one sitting. Drinking more than the recommended amount may cause harm.

What is a unit?

Small glass of wine (100ml)
1 pub measure of spirit (35.5ml)
½ pint of beer, ale, cider, Guinness
1 alcopop (275ml)

There are many suitable alcoholic drinks for people with PKU. However, some alcoholic drinks contain natural protein and therefore should be restricted. *Also some drinks contain aspartame and should be avoided.*

Suitable alcoholic drinks for PKU

- Cider-dry, sweet and vintage for example Ritz, Bulmers
- Glass of red/white/rose wine (need to count if taking more than 2 large glasses of wine) *Note: 1 bottle (750ml) of wine is approximately 1 exchange*
- Fortified wine-port and sherry
- Liqueurs-cherry brandy and curacao
- Vermouth-dry and sweet
- Spirits-whiskey, gin, vodka, rum, brandy, pimms and martini
- Alcopops *
- Cocktails including suitable alcohol

**Remember to check for aspartame*

Chapter 14
Overseas Travel

Being on a low protein diet does not mean that holidays should be restricted to locations that are close to home. There is no reason that anyone with PKU with some forward planning cannot travel anywhere in the world and have a great holiday.

When you have PKU, travelling requires careful planning to ensure you have your low protein foods, synthetic protein and other equipment to hand at all times**.** The more often you travel the easier this planning becomes and the more confident you become.

Dietetic customs letter

If you are travelling overseas you must take a dietetic customs letter. The customs letter will list the name of the synthetic protein and low protein foods that you will be carrying in your luggage. If you do not have this letter you may have difficulties in taking your necessary products into another country. **Ensure to give the metabolic dietitian plenty of notice to arrange this letter.**

Travel insurance

Ensure you take out travel insurance.

Packing your suitcase

Ensure you keep the synthetic protein in its original sealed packaging. Some people find it useful to change to a powder version of their synthetic protein to reduce the overall weight of luggage. You may be able to ship your product to your destination in advance. Contact the nutrition company who makes the product for further details on this.

If you are flying, you can contact the airline to enquire about extra baggage that may be required as some airlines allow you to bring it free of charge.

Lost luggage

It is a good idea to carry extra synthetic protein to cover the possibility of your luggage being lost. Divide it between your suitcase and hand luggage. Make sure you have enough synthetic protein and low protein foods to last a few days in case you arrive at your destination before your main luggage.

Arrange to have someone on standby to post any items you may have forgotten.

Eating on the flight

Contact your airline to see if suitable food options are available. Take low protein foods and exchange foods for the flight. Be prepared for flight delays. It is a good idea to have a plan for the first meal when you arrive at your destination. Have your synthetic protein within easy access.

Booking accommodation

When booking your holiday, it is important to look at your accommodation options.

- Hotel accommodation – before travelling, enquire about the type of meals the hotel provides. A buffet style meal is preferred as it offers a wide variety of options. Contact the accommodation in advance and ask them if they will be willing to prepare your low protein foods. It may be a good idea to provide them with some simple recipe ideas.
- Self-catering may be an option. Check the cooking facilities and local supermarkets that are available.

Label reading

Food labels may be different in foreign countries. Take a dictionary to help with translation. Drinks abroad may not be labelled as containing aspartame. Look out for the E numbers, E951 and E962.

Useful phrases

You may wish to print off some useful phrases and explain PKU and dietary requirements in the language of the country you are travelling to. These could be shown in restaurants and hotels.

Travelling for long periods

If travel is planned for long periods, for example, a few months or longer this will need to be organised at least 6 weeks in advance. You will need to contact the manufacturer of both your low protein foods and synthetic protein to see what services are available. There is no guarantee that your synthetic protein and low protein foods will be sent to your required destination free of charge. This is why forward planning is essential.

If you are planning to stay in one destination for your trip you can be linked in with the nearest metabolic centre, if there is one, to have your bloods done and arrange any outpatient follow up if required.

Holiday Checklist

1. Dietetic customs letter
2. Synthetic protein including some extra in case of emergencies
3. Low protein foods
4. Blood testing equipment (if required)
5. Mixing container and scoop measures
6. Dictionary
7. Diet information for example exchange booklet, calculator, recipes
8. Hospital contact number in Dublin
9. Passport :)

Chapter 15
Pregnancy

Women with PKU have to take particular care before and during pregnancy. This is because high Phe levels during pregnancy can affect your baby.

Consistently high Phe levels may result in the following complications for your baby:

- Heart defects
- Microcephaly (small head and brain)
- Learning difficulties
- Increased risk of miscarriage

We therefore recommend that all women with PKU should plan their pregnancies carefully. Safe Phe levels for a woman planning a pregnancy are lower than normal acceptable levels. We recommend that all women with PKU who are planning a pregnancy or are pregnant need to maintain **blood Phe levels of 120-360µmol/l**.

If you are thinking about **planning a pregnancy**, talk to the metabolic team about the issues for you and your pregnancy. Also discuss the risk of your baby having PKU. Some changes and precautions will be necessary. You should start sending **weekly Phe levels**. The metabolic dietitians will advise you on ways to get Phe levels within the acceptable range. Once Phe levels are within the acceptable range for a number of weeks in a row you can try to conceive.

If Phe levels are well controlled before and during pregnancy, these is no reason why any woman with PKU should not be able to have a healthy baby. Unfortunately, there are other reasons that can cause birth defects that cannot be controlled even if the mother does not have PKU.

> We recommend that **all women with PKU should ideally plan their pregnancies**. However if the pregnancy is unplanned, contact your metabolic team immediately.

It is important to:

- Be healthy before you get pregnant. Have your PKU in good control and try to be in a healthy weight range.
- Check with your metabolic dietitian that your intake of vitamins and minerals is adequate before you get pregnant as well as during the pregnancy.
- All women planning pregnancy should take a 400µg folic acid supplement for three months prior to conception and for the first 12 weeks of pregnancy.
- You may also require DHA (docosahexanoic acid), a special type of omega 3 fat, before and during your pregnancy.
- Maintain close contact with the metabolic team before and during pregnancy. Aim to do weekly levels prior to conception. Let the team know as soon as you know you are pregnant.
- You will need more frequent blood tests-**twice weekly**. Changes to your diet and amount of synthetic protein will be necessary as your pregnancy progresses. You will need more natural protein in your diet as your baby grows and you may need to eat some foods you normally exclude to achieve this. You may also need to start taking a tyrosine supplement.
- The team will work with your obstetrician (pregnancy doctor) to provide the best possible care for you and your baby.
- If you are suffering from morning sickness, inform the metabolic team. Your metabolic doctor and dietitian will work together with you to come up with a plan to ensure you get enough synthetic protein and energy to help maintain good blood Phe levels.

- Gain sufficient weight during the pregnancy. Discuss this with your dietitian. You will need to use additional low protein foods and may need energy supplements. Adequate energy intake is important during pregnancy to keep good blood Phe levels and also for your baby's growth.
- If breastfeeding, regular blood monitoring will continue in the first few weeks to check that your intake of protein and energy is adequate.

Obstetric care during the pregnancy

Your obstetric care before the birth is similar to women without PKU. The difference is you will be in regular contact with your metabolic team. Your metabolic team will advise your obstetrician, GP or midwife about the treatment for your PKU during the pregnancy. They will also liaise closely with the maternity hospital on the plan around the time of the birth.

Like all babies born in the Republic of Ireland, your baby will be tested for many different metabolic disorders and other conditions soon after birth. The metabolic team will advise you and the hospital when the tests should be done.

Contact the maternity hospital where you will be having your baby well before your due date to plan your diet during the admission. The metabolic dietitian can help you plan your meals with the dietitian in the maternity hospital.

Breastfeeding

Breastfeeding is ideal for babies and has many health benefits for both you and the baby. You will most likely need to eat more natural protein and energy than before you were pregnant. Regular blood monitoring will check that your intake of protein and energy is adequate.
If you are planning to breastfeed, please discuss this with your metabolic dietitian.

Further information is available in the **'A Guide to planning for your pregnancy with PKU'** booklet which can be obtained from the metabolic team or downloaded from:
http://metabolic.ie/patient-family-information/metabolic-conditions/phenylketonuria/

Chapter16
Health Related
Benefits & Entitlements

All individuals diagnosed with PKU are eligible to get their synthetic protein and an approved list of manufactured low protein foods **free of charge.** These are provided under the Long Term Illness Scheme. The Long Term Illness Scheme does not depend on your income or other circumstances. A medical social worker will meet with all newly diagnosed PKU patients and their families. They will assist you in applying for a Long Term Illness card (LTI). The medical social worker can be contacted on 01 8784212.

Chapter 17
Glossary

Amino acid

Amino acids are the basic building blocks of proteins. The body makes many amino acids, and others must be obtained from food.

Aspartame

This is an artificial sweetener which contains phenylalanine and should be avoided by people with PKU. It may be listed on food labels as E951 or E962.

Calorie

A calorie is a measure of energy released when a food is eaten.

Energy

Energy is the capacity of the body to do work. The body obtains its energy from the carbohydrate, fat and protein in food.

Enzyme

An enzyme is a protein that facilitates a specific chemical reaction. Enzymes are sometimes described as helpers.

Exchange

An exchange system is used to count protein in the diet. One exchange = 1 gram of protein. Also known as natural protein. People with PKU will be on a set amount of exchanges to provide the body with enough phenylalanine for growth and development. This will vary depending on blood results.

Folic acid

One of the B vitamins. It is recommended for all pregnant women to help prevent birth defects (also called folate).

Gene

Genes carry hereditary information for bodily processes and traits, such as blood group and hair colour, and instructions for producing chemicals.

Natural protein

Also known as exchanges. People with PKU will be on a set amount of natural protein to provide the body with enough phenylalanine for growth and development. This will vary depending on blood results.

Phe

The abbreviation for phenylalanine.

Phe-free formula

Also called the synthetic protein. This is taken by people with PKU to replace the protein in their diet. It contains all essential amino acids (except phenylalanine), plus vitamins, minerals and extra tyrosine. It is vital for people with PKU.

Phenylalanine

An essential amino acid found in protein foods. It is normally converted to tyrosine in the body. It also plays an important role in the formation of brain chemicals or neurotransmitters. People with PKU cannot break down phenylalanine in foods in the usual way, so they must avoid foods containing protein (and therefore phenylalanine). The small amount of phenylalanine they need comes from carefully measured amounts of foods. If PKU is not treated, phenylalanine builds up in the blood causing brain damage.

Phenylketonuria (PKU)

An inherited condition where the body lacks the enzyme phenylalanine hydroxylase needed to break down phenylalanine in foods. The condition is treated from soon after birth with a special diet. It is recommended that this diet be followed for life.

Protein

Protein is made up of amino acids. It is needed by the body for growth and repair. Many foods contain protein. Foods such as meat, fish, chicken, eggs, milk, cheese, yogurts, soya, nuts, bread, pasta and chocolate are rich in protein and are not suitable for people with PKU. A synthetic protein makes up for the protein they are unable to eat in food.

Synthetic protein

Also called the Phe-free formula. This is given to people with PKU to replace the protein in their diet. It contains all essential amino acids (except phenylalanine), plus vitamins, minerals and extra tyrosine. It is an essential part of the treatment for PKU.

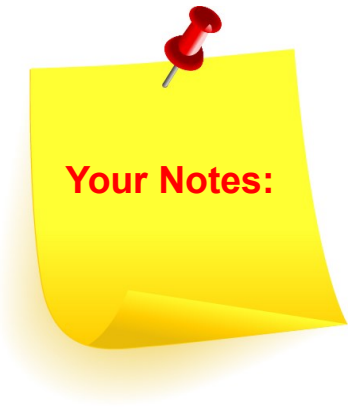
Tyrosine

Tyrosine is an amino acid that is partly obtained from phenylalanine. It is used by the body to produce hormones, skin and hair pigment. It is also considered vital to normal mental functioning. People with PKU cannot convert phenylalanine to tyrosine. They obtain tyrosine from the synthetic protein.

Acknowledgements:

All members of the National Centre for Inherited Metabolic Disorders (NCIMD) team who contributed to this guide.

ASIEM (Australasian Society for Inborn Errors of Metabolism)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Version: 2

Approval date: August 2021

Review date: January 2019