



Mater Misericordiae University Hospital

**A GUIDE TO
PLANNING FOR YOUR
PREGNANCY
WITH
PKU**



Metabolic.ie

National Centre for Inherited Metabolic Disorders

Why Should I Think About This Now?



- ◆ Even if you are not planning a pregnancy, good diet control now will help prevent serious problems in the case of an unexpected pregnancy. Do not forget over half of all pregnancies are unplanned.
- ◆ If you are sexually active and can't control your diet at present, it is important to avoid becoming pregnant until a time that your levels are under control. This can be discussed at your clinic visit.
- ◆ Even if you are not planning a pregnancy, it is easier to **“stay on”** the diet now than going **“back on”** the diet if you do become pregnant in the future. Also, good diet control is better for your health now, and in the future.
- ◆ You can contact the metabolic service at any stage if you have questions. Ring the number of the service you attend and ask to speak to the dietitian and/or the nurse.

What Kind of Problems Can Occur?

If a woman with phenylketonuria (PKU) becomes pregnant and her diet is not well controlled, there is a very high chance that her baby will have serious problems. Examples of these problems include spontaneous abortion, miscarriage, heart problems, microcephaly (small head), intellectual disabilities and behavioural problems.

Why Do These Problems Occur?

This is because there is a doubling of the phenylalanine (phe) concentration across the placenta. Therefore, the foetus is exposed to a higher concentration of phe than that in the mother's blood. Phe levels that are safe for adults can be harmful to the foetus.

For example:

**If mother's level is 300mmol/l then baby's level is approx.
600mmol/l**

The higher the phe concentration at conception, the greater the degree of problems for the foetus. *Any woman who has phe levels > 360mmol/l at conception and regularly during pregnancy may be have a higher risk of problems for her baby.*

Can Any Problems Be Reversed?

Problems that occur during pregnancy as a result of persistently high phenylalanine levels in the mother cannot be reversed or helped by a special diet. These problems are permanent.

How Can These Problems be Prevented?

The good news is that women with PKU who follow their diet to lower their phenylalanine levels **before** and **during** pregnancy will protect their baby from the effects of high levels. However, unfortunately there are other reasons that can cause birth defects in babies that cannot be controlled even if the mother does not have PKU.

This means ideally getting your levels to between 120mmol/l-360mmol/L **before** and **during** pregnancy. Phe levels within normal range at the time of conception and throughout pregnancy are important to your baby's development. **It is very important to plan your pregnancy and maintain strict dietary control before and during pregnancy.**

Pre-Pregnancy Planning and Counselling



The best management begins months before conception. You and your partner will be seen in outpatients. This gives you a chance to discuss the issues, to ensure you both understand the risks involved with raised phe levels and the treatment required. Bloods may be taken at this visit as part of pregnancy planning and assessment to ensure there are no vitamin or mineral deficiencies.

Support from the woman's partner/family member/friend is important as the diet can be a challenge to adhere to in pregnancy as it requires emotional and physical commitment.



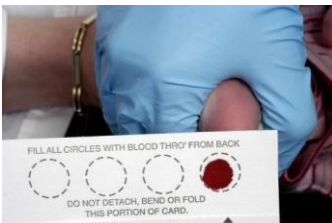
Dietary Management in Pregnancy

The aim of the dietary management is to reduce the risk of foetal abnormalities by:

- Controlling blood Phenylalanine (Phe) concentrations within the recommended range for PKU pregnancy of 120-360 mmol/l
- Maintaining adequate nutrition
- Ensuring appropriate pregnancy weight gain
- Supplementation of Tyrosine
- DHA Supplementation (essential fats)

Controlling blood Phenylalanine (Phe) concentrations within the recommended range for PKU pregnancy

Levels recommended for pregnancy are between 120-360



mmol/l. Levels should be maintained at this level when planning to become pregnant. Levels should be done weekly at this stage for three months prior to conception.

Your dietitian/nurse will advise you which method to use.

At this point taking your prescribed volume of synthetic drink is very important. If you are having any difficulties contact your dietitian. During pregnancy the prescribed volume may change. The volume can be increased for instance if levels rise and reduced depending on the number of exchanges which are tolerated as the pregnancy progresses.

It can be a challenge to be on reduced exchanges when planning pregnancy and in the initial stages of pregnancy. It's a balance of providing adequate energy while restricting protein more than previously.

The following will help:

- Count exchanges accurately and keep a written record
- A sensible choice of the specially manufactured low protein foods
- Creative and imaginative use of fruits and vegetables in combination with specially manufactured low protein foods
- Use of energy supplements, if required.



Maintaining adequate nutrition

With the dietary restriction on protein and the increased nutritional demands of pregnancy it is important that you take the full prescription of synthetic protein. Bloods may be taken to check certain nutritional parameters when you come to clinic. Your nurse/dietitian will discuss this with you.

Folic acid 400 mcg should be taken as for all women planning pregnancy for 3 months prior to conception and for the duration of pregnancy. As pregnancy progresses there may be a requirement for additional supplements e.g. iron. These will be prescribed as appropriate, often by your local gynaecologist or general practitioner (GP).



Supplementation with Tyrosine

Tyrosine is an amino acid (protein) contained in your synthetic drink. Unless you have a milder form of PKU your Tyrosine levels tend to drop in the first trimester of pregnancy. At this stage you may be recommended to take a Tyrosine supplement which is available on prescription. Your dietitian will recommend the amount needed. In certain cases you may require



a medical support letter to ensure this product is provided to you as part of your LTI or medical card allowance.

DHA Supplementation

DHA is an essential fat that is important for you and your baby's brain and nervous system development. If your drink does not contain DHA you will be prescribed a supplement which contains 200-300mg daily. This supplement should be taken when planning pregnancy and throughout pregnancy, it should also be continued after the birth if you are breastfeeding. Your dietitian can arrange a prescription for a suitable supplement if required.



You Have Found Out You Are Pregnant- Congratulations!



You must **contact the metabolic unit** you are attending immediately and continue to pay very careful attention to your diet taking the prescribed number of exchanges and your synthetic protein drink. You will now be asked to send liquid **bloods twice weekly, normally on a Monday and a Thursday with results available on Tuesday and Friday.** It is really important that you have a voicemail set up on your phone for levels and advice as you might not always be available to take a call from the dietitian.



If the pregnancy is unplanned contact the metabolic service you attend **immediately.** More frequent bloods than twice a week may be required until the levels stabilise. A hospital admission can sometimes be required to achieve this.

Remember at any stage you have access to support from other members of the team e.g. Nurses and Doctors. Also, Psychologists and Social Workers where a service is available.

Post Delivery



Once the baby has been delivered, your diet will be adjusted to your usual intake prior to pre-pregnant management e.g. discontinue

Tyrosine supplement and reduce the number of exchanges to your usual pre-pregnancy intake. The team will continue to support you and follow you up in the outpatient clinic as usual.

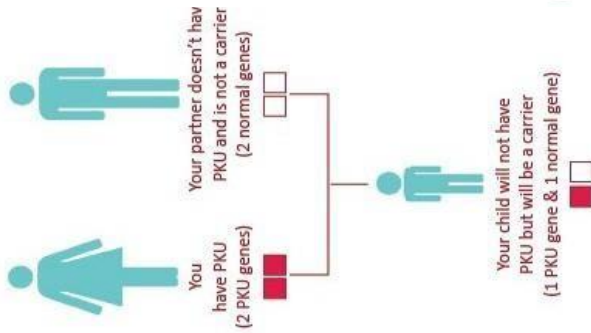
Breastfeed or bottle-feed the baby as you planned. If you plan to breastfeed we recommend you continue to send levels once a week until your diet and phe levels have stabilised. While breast feeding you are likely to remain on increased exchanges and synthetic protein compared to your pre pregnancy allowance.

The baby will have a blood spot test taken for Newborn Screening as is routine for all babies born in Ireland. A liquid sample is also taken on day 3 and day 10 to test the baby specifically for PKU. These samples are taken separately to the new born screening card as there is a short turn around for liquid samples. These will be sent to the Children's University Hospital, Temple Street for analysis. **Contact the maternity hospital you have been attending during your pregnancy for these results.**

Most mothers with PKU will not have children with PKU. A baby can only have PKU if both the mother and father carry a specific gene for PKU.

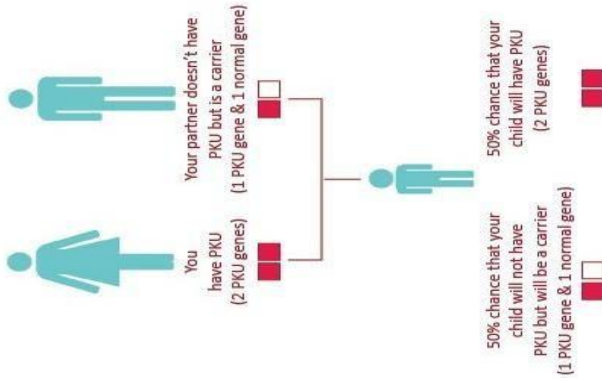
Your Child's Chances of Getting PKU

Most likely possibility:



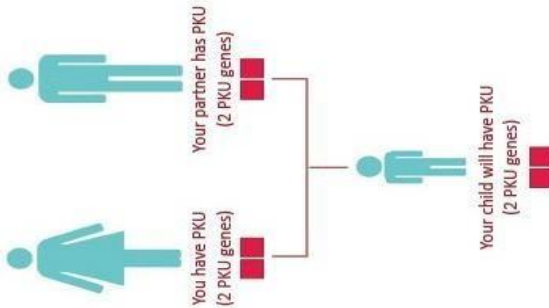
Your Child's Chances of Getting PKU

Rare possibility:



Your Child's Chances of Getting PKU

Very rare possibility:



Checklist for a successful Pregnancy



When Planning

- Arrange a clinic visit.
- Do you have a Long Term Illness (LTI) or medical card?
- Take full prescribed synthetic protein drink daily. Have a good supply of low protein foods.
- Make sure you have a prescription for your drink and low protein foods.
- Stick to the recommended exchanges.
- Do you need exchange lists, recipe ideas and scoops?
- **Send dried bloodspot cards weekly.**
- Ensure you and your partner are fully informed about pregnancy and PKU.
- Avoid pregnancy until your phe levels are within the desired range **(120 - 360mmol/l)**
- Start folic acid supplement (400mcg/d) 3 months prior to planning to conceive.
- Set up a voicemail on your phone if you don't have one.
- **If the pregnancy is unplanned you must contact the metabolic service you attend as soon as possible.**

During Pregnancy



- **Send bloods (liquid samples) twice weekly for the duration of the pregnancy**
- **Rice milk is not suitable during pregnancy**
- Continue to take synthetic protein drink daily – this may be increased during pregnancy
- Continue to take folic acid for the duration of your pregnancy
- Stick to prescribed exchanges – these will gradually increase as the pregnancy goes on
- Ensure adequate calories are taken daily - supplements may be given to ensure calorie requirements are met
- Take tyrosine as prescribed. Make sure you get a prescription and medical support letter if requested for reimbursement
- Ask your metabolic nurse for your medical letter for your obstetrician in your third trimester
- Ask your metabolic dietitian to send a letter to the dietitian in your maternity hospital if required in relation to your dietary management pre and post birth. This is sent approximately a month before you are due

- Filling your freezer with low protein meals can be helpful before your baby arrives. Ensure you have ordered low protein prescription foods again from your local pharmacy

After Delivery

- Post-delivery, gradually return to pre-pregnancy exchanges, stop tyrosine and continue to send regular bloods (dried bloodspots)
- If you are breastfeeding send once weekly levels and discuss on-going dietary changes with the dietitian
- If you are bottle-feeding send monthly levels and discuss on-going dietary changes with the dietitian

Contact Names & Numbers/Useful Notes:

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