Good Phenylalanine Control Is Essential

If a woman with PKU is not on a phenylalanine restricted diet then she will have a high concentration of phenylalanine circulating in her blood. This high concentration of phenylalanine will cross the placenta and can interfere with the normal development of the baby which can result in long-term or permanent damage.

The main abnormalities associated with high blood phenylalanine in the mother are:

• Congenital heart disease
• Damage to the brain
• Small head
• Low birth weight (under 2,500g)
• Non viable pregnancy

These are also known as PKU maternal syndrome. Since the development of the vital organs occur early on in pregnancy, in order to achieve the best possible outcome, phenylalanine levels should be controlled before conception. Therefore, it is strongly advised that pregnancy in a woman with PKU should be planned and good dietary control achieved before conceiving.

Monitoring Your Blood Phenylalanine Levels

Careful monitoring of your blood phenylalanine during the preconception period, and throughout pregnancy is recommended. The frequency and target levels may vary from one metabolic centre to another. Frequent monitoring allows for dietary adjustments by your metabolic dietitian. They will ensure that it is safe for you to conceive with such phenylalanine circulating in your blood, and that you and your baby receive adequate nutrients during your pregnancy. Your metabolic team may want to see you in the outpatient clinic at each trimester to re-assure your compliance and resolve any pregnancy related issues you may have.

The table shows a summary of monitoring that is required when planning a pregnancy:

<table>
<thead>
<tr>
<th>Bloodspot Monitoring Frequency</th>
<th>Target Phenylalanine</th>
</tr>
</thead>
<tbody>
<tr>
<td>Preconception</td>
<td>Twice per week</td>
</tr>
<tr>
<td>Pregnancy</td>
<td>Twice to three times per week</td>
</tr>
</tbody>
</table>

The questions on the following pages are most frequently asked of the medical teams looking after women with PKU who are considering a pregnancy. If you have a question which is not covered here, you can contact the NSPKU directly or your metabolic dietitian.

If you find out you are pregnant and you are not on the diet it is essential that you contact your PKU clinic immediately.
How long do I need to be on a strict phenylalanine restriction before I try to conceive?
Your blood phenylalanine level should be within the target range for 2-4 weeks but this may vary depending on your metabolic team. Once it is safe, you will be advised to discontinue your contraception and start trying to conceive. It is strongly recommended that women with PKU do not try to conceive until they have been on a well-established, controlled diet and their metabolic team are satisfied with their progress.

Does my partner need to be on the restricted diet too?
No, but it would be useful for him to learn about your diet so he understands what you have to do and support you. Men with PKU do not need to be on the dietary restriction to father children.

If I am on diet before conception, and my blood phenylalanine is well controlled throughout my pregnancy, will that mean I will have a healthy baby?
Your chances of having a healthy baby are greatly increased, but as with all pregnancies, there can be no guarantees about the outcome.

I have always been on the PKU diet, so how is this diet different to the preconception diet?
1. Your target levels will be stricter.
2. You may be on a lower amount of exchanges than usual, or require an additional phenylalanine free amino acid supplement.
3. You will be advised to weigh all your exchanges.
4. More frequent monitoring of your blood phenylalanine.

I have been off the PKU diet since my teens. Will that make it harder for me when I go on the preconception diet?
It may be harder for some women, however you can start from afresh. Your metabolic dietitian will guide and educate you on weighing out exchanges, menu planning, how to cook the low protein specialist foods, your low protein prescription, choosing a suitable phenylalanine free amino acid supplement and all the different aspects of following a restricted diet.

Is the diet strict throughout pregnancy?
The aim is to maintain the target phenylalanine levels throughout pregnancy to minimise the risks explained above. At the beginning, you may only be assigned a small number of exchanges. Your dietary regime will be adjusted by your metabolic dietitian depending on your phenylalanine levels. By the second trimester or from 16 weeks onwards, your exchanges may increase in line with the baby’s growth. By the third trimester, some women may be advised to take more than 30 exchanges to maintain the phenylalanine level in the ideal range.

Why can I have so many exchanges towards the end of my pregnancy and still keep my blood levels within target range?
This could be due to the following reasons:
• The foetus is growing rapidly and requires protein for growth
• The liver of the foetus starts to produce the enzyme used to convert phenylalanine to tyrosine

The preconception and pregnancy diet sounds like a lot of time and effort. How will I manage?
At first you may spend a lot of time thinking about your diet and food, but with practice and perseverance, the dietary restriction will gradually become easier. In time, it will become second nature. Planning is essential and you are required to be organised with your prescription, food shopping and meals. Ask relatives and friends to help. Spend some time cooking in batches and freeze your low protein food so that you do not have to cook every evening after work. There are many recipes available using the low protein prescription foods so you can add your exchanges separately. Have fun and don’t let the diet get in the way of life.

Apart from the diet, is anything else different about a PKU pregnancy?
Your antenatal and delivery care will be the same as for a non-PKU pregnancy. However, your obstetric team may take extra precautions if they have not managed a PKU pregnancy, and suggest additional scans. They may wish to monitor the baby’s growth and heart more closely.

Will I need to give birth in the hospital that manages my PKU?
No, most women can choose their antenatal care and birthing centre at their local hospital.

Can I breastfeed my baby?
Yes, even if you decide to discontinue the phenylalanine restricted diet after birth. The slightly higher level of phenylalanine in your breast milk will not be a problem for your baby. If the heel prick result is positive for PKU, a paediatric metabolic dietitian will advise you on feeding your baby.

When my baby is born, will it be tested for PKU immediately?
All babies in the UK have a blood spot (heel prick) test on day five of life and this will screen for PKU and eight other rare but serious health conditions. Some hospitals may take the blood spot earlier as they may not be familiar with mothers with PKU. They will use the same blood card which you have been using to monitor your phenylalanine.
FREQUENTLY ASKED QUESTIONS CONTINUED

Will I be informed about the results of this heel prick?
In some regions, the midwife or newborn screening co-ordinator will notify you of the result. Your metabolic dietitian or nurse can help to chase up the result for you if you provide them with the baby’s details.

Will my child inherit PKU?
Your child will only inherit PKU if the father is a carrier. The carrier incidence for PKU in the UK is 1 in 50 making the chance of your child inheriting PKU at approximately 1 in 100.

Can I find out if my partner is a carrier?
Since there are over 400 different mutations of PKU identified, tests for carriers are time consuming and can be inconclusive. Your partner may carry a different mutation that has not yet been identified by geneticists, and each person carries mutations for different traits. Obtaining the information regarding your partner’s mutation does not often alter the decision to have children therefore this is not available in the UK at current.

Can I talk to other women who have been through a PKU pregnancy?
Yes and this can be arranged with their agreement. You should remember that PKU pregnancies are managed individually and no pregnancy is the same. You should be obtaining the relevant information, motivation and guidance from your metabolic team, and getting the support from your partner, or other women with PKU.

For further information on planned, and unplanned pregnancy please refer to www.nspku.org or ask your metabolic dietitian.

This document has been produced by the National Society for Phenylketonuria (UK) Ltd (NSPKU) and its Medical Advisory Panel (MAP).