PREGNANCY IN WOMEN WITH PHENYLKETONURIA (PKU)

Whilst all young women with phenylketonuria will be aware that good dietary phenylalanine control is especially important in pregnancy, not all know exactly why that is so, or what is required.
The following questions and answers are those most frequently asked of the medical teams looking after women with PKU who are considering pregnancy. If you have a question which is not covered here you can direct it to the Society at our address or email at info@nspku.org or telephone 0845 603 9136.

WHY IS GOOD PHENYLALANINE CONTROL ESSENTIAL?

If a woman with PKU is not on a strict low phenylalanine diet she will have a high concentration of phenylalanine circulating in her blood. This high concentration of phenylalanine will be carried across the placenta and can interfere with the normal development of her un-born child with permanent, long- term and sometimes fatal results.
The main foetal abnormalities resulting from high maternal blood phenylalanine levels are associated with the development of the heart and brain. The formation of the face may also be affected, and a small head and low birth weight are not uncommon.
Since the heart of the unborn child starts to develop at around 5 weeks, before a woman may even know she is pregnant, we feel strongly that **PKU pregnancies must be planned** and good dietary control achieved **before** conception.

WHAT IS REQUIRED

Blood phenylalanine values must be lowered to within the range of 100 – 250 µmol/L before conception and maintained there throughout pregnancy. When this is done the risk of heart defects is virtually eliminated and the incidence of all the other abnormalities associated with maternal phenylketonuria is very greatly reduced. Frequent blood phenylalanine monitoring and dietary adjustments are necessary in order to ensure the best possible phenylalanine control. Patients are asked to send in a blood test twice each week during the preconception period and three times each week throughout pregnancy and the dietitian will be in regular contact by telephone. It is often necessary to make changes in diet at very short notice to keep the blood phenylalanine within the ideal range.
The frequency of visits to the PKU clinic will also be increased – usually every 1-2 months during pregnancy.

QUESTIONS OFTEN ASKED...

Will my child inherit PKU?

Your child cannot inherit PKU unless the child's father is a carrier. The incidence of carriers for PKU in the UK is about 1 in 50 making the chance of your child inheriting PKU approximately 1 in 100.

Can I find out if my partner is a carrier?

Since there are over 400 different mutations of PKU identified so far, tests for carrier status are both time consuming and expensive, and can be inconclusive. They may be inconclusive in the sense that your partner may not carry one of the so far identified mutations but could possibly be a carrier for another mutation not yet recognised. While it might be interesting to try to find out if the partner is a carrier, obtaining that information does not often alter the decision to have children since PKU is not a life threatening disorder.

It sounds as though I shall have to think much harder about what I eat, and spend a lot of time planning menus. I have a full time job, how will I find the time?
At first you will have to think hard about what you shall eat. If you do not find this is so, you are probably not doing it properly! However, with practice and perseverance, and a refusal to slip back into the old casual way of doing things, gradually the diet becomes easier. Patients have said that, in time, it becomes second nature.
Planning is essential and being terribly organised helps, though does not come naturally to all! Enlist the help of relatives or friends who understand the importance of your diet and have a cooking /freezing session at weekends. Plan ahead so that you have all the recipes ready for them to follow and all the ingredients to hand. The recipes for freezing are best to be exchange-free then you can add exchanges separately. Have fun and don't forget to offer your friends some ordinary food!

Does the diet remain very strict all the way through the pregnancy?
The aim of the diet is to keep the blood phenylalanine between 100-250 µmol/L throughout the pregnancy. At the beginning, a diet of just 3 exchanges may be required to achieve this and during the first sixteen weeks small adjustments in the number of exchanges will be made to prevent the blood phenylalanine from going either too low or too high. From sixteen or twenty weeks onwards we see an increase in phenylalanine tolerance and the number of exchanges must be increased, sometimes quite rapidly, to prevent the blood phenylalanine from going too low. By thirty weeks the diet of some patients will have increased to 30 exchanges and at the end of the pregnancy most patients will require 32-36 exchanges to maintain their phenylalanine in the ideal range.
This dramatic increase in exchanges does not mean that the diet is any less strict. It still has to be done with the same degree of accuracy and care but it means that more "normal" food may be used and even exchanges of high protein foods like meat can be included.

Why is it that I can have so many exchanges towards the end of my pregnancy and still keep my blood phenylalanine within the accepted range of 100 – 250 µmol/L ?
We think the main reasons for this are:-
1. The foetus starts to grow more rapidly and uses more protein in growth.
2. The liver of the foetus starts to produce the enzyme responsible for converting phenylalanine to tyrosine.

One patient described the second point very well when she said "Oh yes, that's when the baby starts to do what I can't do." This is right, the mother, because of her PKU, cannot produce enough of the enzyme known as phenylalanine hydroxylase, but when her unborn child starts to produce it we see a fall in the mother's blood phenylalanine and as a result the exchanges must be increased.

When my baby is born will it be tested for PKU straight away?
No.This would only be misleading. Your baby will be born with a slightly raised blood phenylalanine because of your PKU. This will fall quite rapidly and, provided your baby does not have PKU, the phenylalanine will remain within normal values even after milk feeds have been fully established. If however, your baby has inherited PKU the blood phenylalanine level will be slightly raised at birth but will continue to rise as milk feeds are introduced.
All babies in the UK are tested for PKU between 6-10 days of life and this is appropriate for your baby too.

Will I be informed about the result of the test for PKU?
Normally parents are not told the result of the test unless their child has inherited PKU but dietitians try to obtain the result and report it to the parents as soon as possible.

Can I breast feed my baby?
"Yes, and even if you choose to come off your low phenylalanine diet when the baby is born you can still breast feed your baby because the slightly higher phenylalanine level in your breast milk will not be a problem for your baby."

If we knew that my partner was a carrier would he need to be on diet too?
No, but he should learn all about your diet so that he understands what you have to do and can be of help and support to you.
Men with PKU do not need to be on diet to father children.

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

How long do I need to be on a strict low phenylalanine diet before I may try to conceive?
Your blood phenylalanine will come down to the ideal range within 1-2 weeks of commencing pre-conception diet. We recommend that patients do not try to conceive until they have been on a well controlled diet for at least a month.

Apart from my diet is any thing else different about a PKU pregnancy?
Obstetrically there is no difference. Your ante-natal and delivery care will be the same as for a non-PKU pregnancy except that your obstetrician may request additional scans to monitor the baby's growth more closely.

Will I have to have my baby in the hospital I attend for my PKU?
No. Most women choose to have their ante-natal care and give birth to their babies in their local hospital. This is fine because having PKU does not make the process of giving birth any different.

I have been off my low phenylalanine diet since I was 12years old. Will that make it harder for me when I want to go on a strict low phenylalanine diet for a planned pregnancy?
Using the experience of many PKU pregnancies the rather surprising answer to this question is quite definitely "no"! It is often harder for someone who is already on some degree of phenylalanine restriction to tighten up their diet to the required standard than it is for the person with PKU to start afresh and go from an entirely normal, unrestricted diet to a strict low phenylalanine diet.
We believe the reason for this is that the diet young adults with PKU are on is very different to the diet for a planned pregnancy and they do not realise just how different until they come to do it and are actually shown in very practical terms how it must be done.

I have never come off my PKU diet. In what ways is the diet for a planned PKU pregnancy so different?
1. You will be aiming for phenylalanine levels of 100 – 250 µmol/l. Blood phenylalanine values of 400-700 µmol/L are too high.
2. You will need to weigh all your exchanges – no estimating or guessing.
3. It is essential that you take the number of exchanges prescribed every day. If you are on 5 exchanges it must be 5 each day, not 4 one day and 6 the next day.
4. Some foods may no longer be considered suitable for use as exchange items. For example foods of uneven composition such as muesli or soup containing chunks of various vegetables.
5. There can never be a day when you say "I shall relax my diet a little." The diet continues whether it is Christmas, or your birthday or you are away on holiday. This does not mean that you cannot have special food for example at Christmas; you can as long as it conforms to your dietary prescription. There are lots of really good recipes available and we have a wider choice of prescribable low protein foods here than in any other country making it possible to create an appetising, and totally exchange-free meal, should that be necessary.

What if my baby has PKU, could I still breast feed it, and would it still be alright for me to come off diet?

This is an area about which there is still not very much experience but the advice we give is yes you can still breast feed your baby if he/she has PKU. The baby would be put on a low phenylalanine diet which would mean taking the special phenylalanine-free formulas well as breast milk. The baby's blood phenylalanine would of course need to be monitored regularly and the amount of special phenylalanine-free formula feed would be adjusted as necessary to keep the blood level in the range of 120 – 360umol/L.

It is unlikely that your baby will have PKU but in this happened you would need to discuss breastfeeding and your own diet with your doctor.

Can I talk with other women who have been through a PKU pregnancy?

Yes, this can be arranged with their agreement but you should remember that PKU pregnancies are managed individually and no pregnancy is the same. While getting in touch with other PKU women may make you feel less isolated, of much greater importance is getting the right information, motivation and guidance from an experienced metabolic team.

Produced by the National Society for Phenylketonuria
and its Medical Advisory Panel.

N S P K U

The National Society for Phenylketonuria (United Kingdom) Ltd.

The Society is a registered charity.
It offers support to people with PKU and their families by producing various publications including a quarterly newsletter, organising formal and informal meetings and conferences.

Further information and details can be obtained by contacting:-

The NSPKU Helpline on: 0845 603 9136

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