

Leaflet on Pregnancy in Women with PKU

The NSPKU produces various publications which provide information about Phenylketonuria for those with PKU and their carers.

Below is an extract of our leaflet for women with PKU who are considering starting a family.

During pregnancy the blood of mother and baby come into close contact, making sure that the developing baby receives enough oxygen and food. When the phenylalanine concentration in the mother's blood is high, too much phenylalanine is passed to her baby. The high concentration of phenylalanine may harm the baby before birth.

WHAT IS THE RISK?

There is a high risk that women who have severe PKU will have babies with some abnormality unless preventative treatment is given. What is more in pregnancy dietary control needs to be even tighter than in childhood.

WHAT TYPE OF DAMAGE CAN OCCUR?

The greatest risk is in the early weeks after conception and during the first half of pregnancy. Within a few weeks of the first missed period irreversible damage may occur. The baby, including its brain, may not grow properly so that birth weight is reduced and the baby may be mentally retarded. In addition, organs such as the heart, bowel or eyes may be malformed. Occasionally these abnormalities cause an early miscarriage or death of the baby around the time of birth.

WHAT CAN BE DONE TO PREVENT THESE EFFECTS?

There is now good evidence that the risk of damage to the baby is greatly reduced if the mother re-starts a strict low phenylalanine diet before conception has occurred. The control during pregnancy needs to be stricter than in childhood in order to prevent damage occurring. A woman with Phenylketonuria who wishes to have children should consult a specialist with experience in the management of Phenylketonuria **WELL BEFORE BECOMING PREGNANT, AND WHILE STILL PRACTISING EFFECTIVE CONTRACEPTION.**

A woman who has conceived whilst on a normal diet should contact her medical advisers as a matter of urgency.

WILL THE BABY HAVE PKU?

It is unlikely that the children of a woman with PKU will have PKU themselves, although all of them will carry the condition. For the children to have PKU the father **MUST** be a carrier (only 1

in 60 of the population of the UK are carriers). If a woman with Phenylketonuria does marry a carrier then, at each pregnancy, there is a fifty-fifty chance her child will have Phenylketonuria. All her children will, in any case, be tested in the first week after birth and, if the child has Phenylketonuria, early treatment will be given.

CAN THE BABY BE BREAST FED?

This should be possible, your paediatrician or dietitian will advise you about this.