Hyperphenylalaninemia (HPA)
Information for Parents

What is the heel prick test for?
Every baby in the UK is offered a heel prick test (Newborn Blood Spot Screening) to identify several rare but treatable medical conditions at a very early age, before they become apparent or cause harm. Your child’s heel prick test was positive as it showed a raised level of phenylalanine. Phenylalanine (phe) is an essential building block of protein in the body.

What is Hyperphenylalaninemia (HPA)?
HPA is a much milder form of a rare genetic condition called phenylketonuria (PKU). Babies with PKU require early and lifelong dietary treatment to maintain the phe levels in the body within an acceptable range. Sustained, high blood levels of phe can harm the brain and without early and effective treatment children with PKU can develop learning difficulties and developmental delay.

HPA means that phe is present in higher amounts than usual in the blood, however babies with HPA do not usually require dietary treatment as the blood phe level is usually within an acceptable range even without treatment. Children with HPA are healthy and will grow and develop as well as unaffected children and do not come to any harm.

What causes HPA?
When the body is unable to break down and get rid of the excess phe, it accumulates in the body causing raised levels of phe in the blood. This is how we identified HPA in your baby. Phe is required by the body to produce protein, which is essential for several vital functions, including growth and development in children. We obtain phe from protein in the food that we eat. The body uses the phe that it requires and gets rid of any excess that it does not need. The body does this through chemical reactions that break down phe to either produce energy or other chemicals.

Is monitoring required?
Blood phe levels will be monitored as required to ensure that they are maintained within the acceptable range. No dietary treatment is usually required. Your baby will be expected to grow and develop as normal.

Why has my baby got HPA and could I have prevented HPA in my baby?
HPA is an inherited genetic condition and not a result of anything that you may or may not have done either during or before this pregnancy. It is not possible to test parents or the growing baby during the pregnancy to identify this condition before birth. Therefore it is not possible to know if a baby is going to be affected with HPA or not.

Will the phe levels remain the same throughout life?
HPA is a lifelong condition that does not ‘go away’ but may change at various stages in your baby’s life. Blood phe levels vary during the day and from day to day. Your baby’s phe levels will also vary at different times. However, it is unlikely to require treatment. If phe levels remain higher than the acceptable range, dietary treatment may be required. However this is unusual.

Do phe levels have to be monitored in my baby?
Initially frequent blood samples (once a week) may be required until it is certain that the phe level remains generally within the acceptable range. After this further blood tests may no longer be necessary. If required parents/carers may learn how to take blood samples.

The only time later in life that frequent blood tests will be required is during pregnancy in women with HPA.

So does this mean when my baby is grown up we can forget about this condition?
When women with HPA wish to have children, it is very important to monitor blood phe levels before and during pregnancy. The phe levels should be very strictly controlled before and during pregnancy in order to prevent harm to the unborn baby while it is developing and growing. Before and during the pregnancy, special dietary treatment may be required for women with HPA in order to adequately control the blood phe level.

Pregnancy should always be planned, so that the blood phe levels in women with HPA can be controlled at the low levels required to achieve a good outcome in the unborn baby.
What happens when my daughter leaves home?
It is important for your daughter to be aware from an early age of the precaution and special care required around the time of pregnancy. It would be advisable for her to maintain contact with a specialist metabolic centre or attend the metabolic clinic at least once a year. Her GP will also be able to help her access the nearest specialist metabolic service at the appropriate time. *

It is essential to reinforce the importance of contraception and the risk of unplanned pregnancy in girls and women with HPA.

Where should my daughter, who has HPA, seek help before pregnancy?
Your daughter should be managed jointly by a metabolic dietitian in a specialist metabolic centre or specialist team. [www.nspku.org/contact/emergency](http://www.nspku.org/contact/emergency)

What about boys with HPA?
Boys may also want to maintain contact with a specialist metabolic centre. However, boys are not currently known to have any special issues relating to HPA in later life and so no specific treatment is usually required.

Can HPA occur again in my family?
HPA is inherited from both parents who are themselves unaffected ("carriers"). Any future pregnancy has a 1 in 4 chance of your baby being affected. An affected baby with HPA should be identified during newborn screening. However, it would be helpful if the midwife/newborn screening team were informed that a sibling has HPA.

You will be provided with further information by the Newborn Screening/metabolic/genetic team during your first consultation.

For more information about Newborn Blood Spot Screening please visit: [www.nhs.uk/Conditions/pregnancy-and-baby/Pages/newborn-blood-spot-test.aspx](http://www.nhs.uk/Conditions/pregnancy-and-baby/Pages/newborn-blood-spot-test.aspx)

*If you as a family move home and/or when your daughter leaves home it is important that the metabolic clinic is informed of any change of address and contact details.

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