

Commonly asked questions

1. What is PKU?

PKU, also known as phenylketonuria is a defect in the enzyme responsible for converting phenylalanine to tyrosine. The resulting build up of phenylalanine is neurotoxic.

2. How and when will we know if my child has PKU?

If your child's newborn screening result was greater than 6 mg/dl, your child probably has PKU or a disorder similar to PKU. If the result was lower your child either could still have PKU or it may have been a false positive result. The newborn screening test will be repeated and possibly blood amino acids will be measured to determine if your child has PKU or not and the degree of PKU, if he or she does have it. Typically the repeat newborn screening result will take up to 4 days to come back. Amino acids assayed at a metabolic lab may be available within 1-7 days.

3. How did my child get this?

PKU is an autosomal recessive disorder. This means that your child has 2 PKU genes, one from the mother and one from the father. Having only one PKU gene does not affect a person at all.

4. What does it mean for my child?

If your child has PKU, he or she will be on a special diet but, with this treatment, will be quite normal and indistinguishable from other children.

5. What is the treatment? Does it work? Is the diet difficult to do/expensive?

PKU is treatable by means of a special diet involving protein restriction and supplementation with a special formula. This treatment prevents the build up of toxic phenylalanine levels and means that your child can lead a healthy life. The diet is usually covered by insurance.

6. What about my other children/future children?

As PKU is an inherited condition it is important to have your other children checked out. If your other children are healthy and well they almost certainly do not have PKU but they may carry the gene for it like you. If you have any children who are retarded and have not been screened for PKU for any reason, it is possible that they may have the condition. It is important to identify this as sometimes treatment can help in previously undiagnosed children though it will not cure them of the brain damage.

It is therefore also important to let your obstetrician and pediatrician know about a child with PKU if you are planning future pregnancies so that they may discuss the options with you and prepare accordingly.