

Genetics of PKU

Genetic information, which determines every person's characteristics like eye or hair color, is found on pairs of genes in every cell of the body. These genes serve as blueprints, or patterns in each person's genetic make-up. PKU is a genetic disorder inherited from both mother and father. It is passed to children in an autosomal recessive manner, meaning both parents must be carriers of the affected gene for it to be inherited by a child.

Most often, each parent of a child with PKU has one normal gene and one altered (PKU) gene. Every child of that couple receives one gene from each parent. A child that receives two normal genes (one from each parent) will be completely normal. A child that receives one normal gene and one PKU gene will be able to make enough Phe hydroxylase to use Phe normally and may not show any signs of PKU. He will be a *carrier* of the gene, however, and may pass this gene onto his children. A child who receives two altered genes, one from each parent, will have PKU.

If both parents are carriers of the PKU gene, they will have a one in four (25%) chance *with each pregnancy* of having a child with PKU. They have a two in four (50%) chance of having a child that is a carrier of the PKU gene, and a one in four (25%) chance of having a child with only normal genes. These probabilities are for each pregnancy, so this couple cannot assume that because they have one child with PKU, they will not have another.

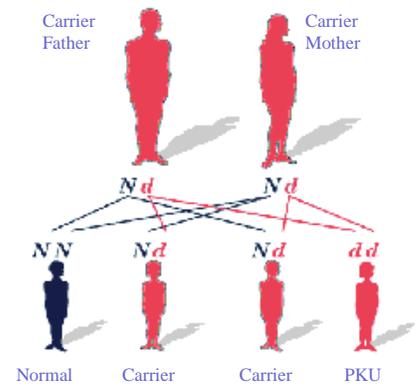
If one parent is normal and one parent is a carrier of PKU, the couple will have a one in four (25%) chance of having a child that is a carrier of PKU. If one parent is normal and one parent has PKU, all children will be carriers of PKU. If one parent is a carrier of PKU and one parent has PKU, all children will either have PKU or be a carrier.

Symptoms

If PKU has gone undetected or untreated, the infant may have some or all of these symptoms:

- ◆ Vomiting
- ◆ Irritability
- ◆ Hyperactivity
- ◆ An eczema-like rash
- ◆ Pungent, mousy odor to urine
- ◆ Convulsions or seizures
- ◆ Moderate to severe mental retardation
- ◆ Microcephaly (small head)

However, with early diagnosis and *life-long* nutrition support, a person with PKU can be normal if blood phe levels are always kept near normal. When PKU is diagnosed and treated, behavior problems and skin rash usually improve first. If diagnosis is delayed and mental retardation is already present, there may be some improvement as a result of the special diet, but brain damage that has already occurred will remain. For this reason, early diagnosis and treatment are essential to the quality of life and well-being of the child with PKU.



Diagnosis

PKU is diagnosed by means of a simple blood test performed on all newborns soon after birth. This test is required by all states before the baby leaves the hospital. It tests for excess levels of Phe in the blood. If the initial screening test result shows high levels of Phe, more tests are done to determine if the baby has PKU or another disorder. Some doctors hospitalize the baby because testing is more rapid and diagnosis and treatment can begin sooner.

