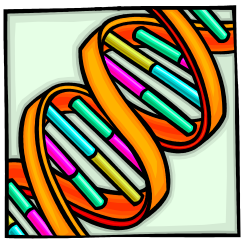

INFORMATION ON
PKU CARRIER TESTING
ADULT METABOLIC DISEASES CLINIC
VANCOUVER GENERAL HOSPITAL

WHAT IS PKU?

PKU (Phenylketonuria) is an inborn error of protein metabolism. Protein is made up of many different building blocks called amino acids. People with PKU cannot break down the amino acid phenylalanine. PKU is successfully treated with a modified diet. By following this special diet, the person with PKU can develop normally and live a completely full life.

WHAT IS A CARRIER?

PKU is a genetic disorder. To have PKU, a person has to be born with it. A baby has PKU from the moment it is conceived. The genes that cause PKU are passed to the baby from the mother and father. To have PKU, the baby has to get one copy of the gene from the mother and one copy of the gene from the father. The baby therefore gets a 'double dose' of the genes that cause PKU. This is referred to as **recessive inheritance**. The mother and father do not *have* to have PKU to pass it to their baby. They are called **carriers** of the gene that causes PKU.

I AM MARRIED TO A MAN WITH PKU. WILL I HAVE TO FOLLOW A SPECIAL DIET WHEN I AM PREGNANT?

No. If you do not have PKU, you do not have to be on a protein-restricted diet during pregnancy. Only women with PKU need to be on the special PKU diet during their pregnancies.

WHAT ARE THE CHANCES OF ME BEING A CARRIER?

About 1 in 50 people carry the gene that causes PKU.

IF I HAVE PKU, AND I DON'T KNOW IF MY PARTNER IS A CARRIER, WHAT ARE THE CHANCES OF US HAVING A CHILD WITH PKU?

There is a 1 in a 100 chance of a person with PKU meeting someone from the general population and together having a child with PKU.

1/50 (incidence of PKU gene in general population) x ½ (chance that if the partner was a carrier for the gene that the partner would pass it on) = 1/100

IS THERE A WAY TO FIND OUT IF I AM A CARRIER?

Yes, there are ways to detect carriers. However, before reading about the ways it can be done, ask yourself some questions that can help you decide if you even want to know about carrier testing.

Take a few moments and think about your answers to the following questions:

- Why do you want to find out if you carry the gene for PKU?
- If you were tested and found out you were a carrier, how would that influence your decision to have children?
- Would you choose NOT to have a child if you thought that there was a chance that it may have PKU?
- What if the test showed you were a carrier, you and your partner decided not to have children at all based on this information, and then it was later determined that you were not a carrier?
- What if the test showed you were not a carrier, and your baby was born with PKU (i.e. the test was wrong). How do you think you might feel about that?

Most people, when they think about these questions, realize that they would still want to have a baby even if there was a chance that the baby was going to have PKU. PKU is completely treatable and babies with PKU have normal lives. Therefore, it does not really make sense to think that you should not have a baby just because the baby might need a special diet, any more than it makes sense to think that you should not have a baby because it might be allergic to peanuts or because it has blue eyes!

There are 2 tests that may help determine whether or not you carry the gene for PKU but both tests have disadvantages.

1) **Phenylalanine Loading test**: This test involves having a blood test, eating a high protein meal, and having the blood test again.

Although the test itself is fairly straightforward, the results are not always accurate. This test is said to have **low predictive value**. That is, it has a significant risk of false results. For example, sometimes the test indicates that a person carries the gene for PKU when they really don't (**false positive**), and sometimes the test indicates that the person does not carry the gene for PKU when they really do (**false negative**).

We do not recommend this test, as you may make the important decision about if you want to have children based on inaccurate results.

2) **DNA Mutation Analysis**:

The only accurate way to determine if a person is a carrier is to have a DNA analysis done to identify the mutation in the gene. This is a very expensive test, and this test is not covered by B.C. Medical, thus you would have to pay for it yourself.

SUMMARY

1. **The PKU loading test is not 100% reliable**.
   There is a risk of incorrect results.

2. **The DNA Mutation Analysis is expensive and not covered by B.C. Medical.**

3. **PKU is successfully treatable with diet**
   If you did have a child with PKU, they would be healthy and of normal intelligence by following the special low protein diet and drinking the special formula.

HOW WILL I KNOW IF MY BABY HAS PKU?

All babies are tested shortly after birth for PKU. If your baby is found to have this disorder, you will be contacted immediately and referred to a metabolic team to help you with your baby's special diet.

Still unsure? Any other questions? Please contact us at the Adult Metabolic Diseases Clinic, Vancouver General Hospital
604-875-5965