Will my baby have PKU?

PKU is a genetic condition caused by a malfunctioning gene. Our genes are inherited in pairs, with one coming from each parent. A person with a mutation on one copy of the PKU gene is called a carrier. This person does not have PKU, but can pass the mutation on to his/her children. If you have PKU, you have a mutation on both of your PKU genes. You will pass on one of these mutations to every child you have.

When both partners are carriers (see Figure 1), there are four possible genetic outcomes in every pregnancy, each with a 25% chance of happening: (1) the child inherits one mutation from mom, or (2) one mutation from dad, and is a carrier, like mom and dad; (3) the child inherits a mutation from both parents and has PKU; or (4) the child inherits both copies of the normal or unaffected PKU gene, does not have PKU and is not a carrier. These possible outcomes are the same for each pregnancy.

If you have PKU, the chance that any of your children will have PKU depends on whether your partner is a carrier. If your partner is not a carrier (see Figure 2) he/she has only normal PKU genes to pass on. All of your children inherit a mutation from you and will be carriers. If your partner is a carrier (see Figure 3) your children will all inherit a mutation from you, and there is a 50% chance in each pregnancy that they will ALSO inherit a mutation from your partner and have PKU. If both you and your partner have PKU, all of your children will also have PKU (see Figure 4).

Key
- P = Normal PKU Gene
- p = Mutated PKU Gene