PARENT FACT SHEET:

Phenylketonuria (PKU) Prenatal/Pre-conception Testing of the Phenylalanine Hydroxylase (PAH) Gene

Carrier screening is a form of genetic testing which can determine if you carry a gene for certain genetic disorders. Many prospective parents or currently pregnant parents are offered carrier testing to detect if their children may be at risk for a genetic condition, particularly if the condition is treatable.

Phenylalanine Hydroxylase (PAH) is the gene which causes ***Phenyketonuria (PKU)***, and is included in many carrier tests offered to adults who would like to understand their genetic risks for a pregnancy. ***PKU*** causes difficulty breaking down the amino acid, phenylalanine (Phe). Phe can adversely impact the brain if it is severely elevated. PKU is a treatable disorder, and those identified by newborn screening and with ongoing adequate dietary treatment, with or without medication, are expected to have normal intelligence. For more information about PKU, please see https://www.newenglandconsortium.org/pku-open-page.

The PAH gene is responsible for the disorder ***Phenylketonuria (PKU)***, and the PAH gene also can also cause milder conditions than ***classical PKU***, including ***mild PKU*** and ***benign hyperphenylalaninemia*** (***hyperphe***). ***Hyperphe*** means there is higher than the typical amount of phenylalanine in the blood. Some people with elevated Phe need treatment with a special diet or medication whereas other do not require treatment. People with benign hyperphenylalaninemia do not require treatment and are expected to have normal growth and intellectual development.

If both parents are carriers of variants in the PAH gene, this could mean their baby is at risk for ***PKU*** but it could also mean their baby may be at risk for ***benign hyperphe***, with mild laboratory changes that do not cause symptoms or require treatment.

Your genetic counselor can help you understand your risk on the spectrum from benign hyperphe to PKU by looking up your genetic variants on BioPKU.
http://www.biopku.org/biopku/search-start.asp

If you and your partner are both carriers of variants in the PAH gene, we recommend consulting a metabolic physician who treats patients with PKU to help understand the genetic testing results for PAH deficiency.