WHAT THE STUDY INVOLVES

5 research visits to the Emory Genetics Clinic or Emory Hospital CIS over a 12 month time period.

Visits will be for

- Eligibility screening and baseline assessment
- Research follow-ups at 4 weeks, 4 months, 8 months, and 12 months after starting Kuvan™

At each visit, we will collect a small blood sample, overnight urine sample, height and weight, vital signs, and provide you with questionnaires to fill out. The baseline visit and the 12 month follow-up visit will also involve a DEXA scan to determine body composition and bone density, and Indirect Calorimetry which measures the energy your body uses while at rest.

Additional non-research standard care visits may be included so that we can best monitor your health and nutrition status while you are taking Kuvan™.

Contact Information

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www.genetics.emory.edu/NUTRITION/BH4andPKU
### Eligibility

**You or your child with PKU may be eligible to participate in this clinical research opportunity if...**

- You or your child are at least 5 years of age
- You or your child have been diagnosed with Phenylketonuria (PKU)
- You intend to try the prescription Kuvan™ for yourself or for your child
- You or your child have not taken BH4 or Kuvan™ for at least 8 weeks.
- You or your child of reproductive age are not pregnant or planning to become pregnant
- You can provide informed consent on your own behalf or on behalf of your child

### About This Research Opportunity

#### What is BH4?

BH4 (Tetrahydrobiopterin) is a molecule that humans naturally make. BH4 helps the PAH (Phenylalanine Hydroxylase) enzyme convert Phenylalanine (Phe) into Tyrosine. Kuvan™ is a pharmaceutical (prescription drug) form of the helper BH4 molecule. In people with PKU, also known as Phenylketonuria, the PAH enzyme does not function well causing high levels of Phe to build up in the body. Too much Phe in the body can be toxic. Medical food (formula) and a low-Phe diet help keep people with PKU healthy. For about 50% of people with PKU, taking extra BH4 helps to convert more Phe into Tyrosine. In these people, Kuvan™ can help control blood Phe levels and increase dietary Phe tolerance.

#### What is the purpose of this study?

This clinical trial will help us to better understand and improve treatment options for those diagnosed with PKU by providing the opportunity to monitor health and nutrition for PKU patients who are interested in trying the prescription drug Kuvan™. Over a 12 month time period, we will look at the following specific things in our study volunteers: diet, growth, body muscle and body fat, bone density, blood markers of nutritional health, blood Phe and Tyrosine levels, levels of neurotransmitters (chemicals produced by the nervous system) in blood and urine, quality of life, as well as feelings and attitudes about Kuvan™ itself.

#### Who can volunteer?

We are seeking individuals with PKU who are interested in volunteering for this clinical research opportunity. Please look at the eligibility requirements listed in this brochure to determine if you would be able to enroll in this study.

#### Study Plan

Individuals with PKU who enroll will have their Phe response to Kuvan™ determined within 4 weeks. PKU patients who exhibit at least a 15% reduction in blood Phe will be considered “responders”. Both responders and non-responders to Kuvan™ will be allowed to stay in the trial for the whole 12 months. A clinical dietitian will be available to help PKU subjects with their diet and to make any proper adjustments to protein allowance or medical food (formula) prescription.

#### Who is managing the study?

- **Rani H. Singh, PhD, RD:** Lead Investigator
- **MaryJane Kennedy, RN:** Research Coordinator
- **Teresa Douglas, MS:** Research Assistant