PKU Patient Registry

Survey Index

About the Participant Survey
Contact Information Survey
Demographics Survey
Family History Survey
Diagnosis Survey
Genetics Survey
Treatment/Diet Survey
Blood Phe Levels Survey
Medical History Survey
Education Survey
Female Reproductive Health Survey
Maternal PKU Survey
Measuring Mood Survey
Insurance Survey
Research Trials Survey
Three Introductory PKU Surveys

About the Participant

Contact Information

Demographics

Time Required

10 – 15 minutes for all 3 surveys. Surveys collect basic facts including contact information, weights and measures, and units used to measure phe levels. There are several mandatory questions on these surveys that will provide a basis for other questions in the registry. Most information will be known off hand.

Background work involved

Very little to none – questions about annual household income, nutritionist’s info, ethnic background may require a little research.

How this information may help

In addition to providing a way for the participant to be reached, this demographic information may shed light on difference in PKU care or outcomes based on where you live. Reviewing data on marital status or employment history may help shed light on how individuals with PKU fit into their community. Providing and updating contact information will allow us to contact you if you indicated that you would be interested in participating in other research projects.

Tip:

The About the Participant Survey will only be done once.

You will be asked to update the Demographics survey once per year.

The Contact Information survey can be updated at any time. Once submitted, it can be found under the “Surveys to Retake” tab.
Family History Survey

Time Required

5 minutes or less – This 4 question survey collects information about any family history of PKU and the educational status of parents of an individual with PKU.

Background work involved

Little to none

How this information may help

Individuals with a family history of PKU may provide researchers with an opportunity to look at PKU within a family unit and identify factors that may contribute to different outcomes. Knowing the educational background of parents may help shed light on the impact that has on the development or school performance of an individual with PKU.

This survey will only be done once. There will be no requests for updates. If family history changes (new sibling, family member with PKU is identified) the survey can be updated by contacting registry@pku.org.
Diagnosis Survey

Time Required
Less than 10 minutes. This 5 question survey asks general questions about the PKU diagnosis that should be known off hand.

Background work involved
Little to none. You will be asked the units the lab used (mg/dl or umol/l) to report diagnostic phe level.

How this information may help
Understanding the severity of PKU (mild to classical) can be used to look at how this may influence outcomes such as quality of life or growth. Information can help identify incidence based on region.

Tip
You will only be asked to complete this survey once.
Genetics Survey

Time required
Less than 5 minutes

Background work involved
Some - If Genetic testing was done – you will need to find and enter the results of PAH gene mutations. If possible you can upload a copy of the PAH mutation report using the Upload survey. *If no testing was done – this step is not necessary.

How this information may help
Genotype information in Registry could be used to develop genetic or genetically related therapies. Gene editing might require knowledge of specific mutations with research and treatment efforts directed in this area.

Tips
This survey will only be done once. You will not be asked to update again. If additional information becomes available you may update this survey by contacting registtry@pku.org.

*Please note – If PAH mutation testing has never been completed, as a registry participant you have an opportunity to access testing through the BabyGenes lab. NPKUA and BabyGenes have partnered to offer genetic testing at a reduced price. To learn more click here registtry@nplua.org

Version 1.0
17Apr2017

Hummingbird IRB Approved
04/18/2017

Back to Index
Treatment and Diet Survey

Time Required

20 minutes. One of the longer surveys containing ~ 18 questions about medical food, diet and medications. Questions cover specifics about formulas and low protein foods (including brands and doses). Questions also include current height and weight, frequency and type of blood testing (phe, tyrosine and other labs) and highest and lowest recorded blood phe level. About 1/3 of the questions are mandatory requiring an answer to submit the survey as final. If information is unknown at the time, the survey may be submitted as a draft and completed when information has been gathered.

Background work involved

Some – Looking up highest and lowest phe levels or names of annual laboratory tests may be necessary. Clinics may be helpful in providing this information.

How this information may help

Because diet therapy plays such an important role in disease management, understanding how different formulas or the low protein diet impact phe levels, growth and nutritional status is crucial. Having the ability to review what laboratory tests are used by clinics for monitoring and how this varies regionally could help standard monitoring practices.

Tips:

You will be reminded to update this important survey every 6 months. You will not be able to make any changes until that time.


**Blood Phenylalanine Survey**

**Time Required**

Varies depending on number of levels entered. Time required to enter one phe level is less than 1 minute. Phe and tyrosine levels will need to be added one at a time by using the + Add another record button (insert picture here). You will be asked for the phe or tyrosine result along with the date it was done. You may enter as many phe levels as you would like.

Personal Phe level charts, logs or graphs can be uploaded into the registry using the upload survey.

**Background work involved**

Some – accessing personal records or asking the clinic to provide with results.

**How this information may help**

One of the most important surveys in the registry that will provide the foundation from which many different activities can be planned. Outcomes can be measured and quality of life assessment can be compared with phe levels. A better understating of what the optimal range for blood phe control is could be determined. The role of tyrosine could be better understood as it relates to our current approaches to management.

**Tips:**

The Phenylalanine Levels survey can be updated at any time. Once submitted, it can be found under the “Surveys to Retake” tab. We encourage you to enter phe levels as you receive them to keep up to date.
Medical History Survey

Time Required

20 – 30 minutes. This is the longest survey in the registry with about 20 questions with many of these required. The questions ask about past hospitalizations and general physical and mental health in a variety of different areas. You will be asked for dates if they are known. Questions also cover exercise and stress management.

Background work involved

Some – Dates are requested (although not required) for diagnoses and hospitalizations. Clinics may be able to provide this information if not available.

How this information may help

Understanding or observing trends in illnesses in the PKU population could result in the development of treatments aimed at prevention. With a better understanding of risks – monitoring could be focused in these areas. A closer look at regional differences in outcomes could help develop more effective treatments.

Tips:

You will be asked to update the Medical History survey once per year.
Education Survey

Time Required
Less than 5 minutes. Contains 4 questions.

Background work involved
Little if any. If IQ test documentation is available it may be uploaded.

How this information may help
Identifying and documenting specific educational challenges for the PKU student could provide parents with the data needed to request and receive educational resources for their child.

Tips:
You will be asked to update the Education survey one per year.
2 Surveys for Females Only
Female Reproductive Health Maternal PKU

Time Required

Less than 5 minutes for Female Reproductive Health survey – 4 questions
10 – 15 minutes for Maternal PKU survey – if applicable to the participant.

The Maternal PKU survey collects information including phe levels for each trimester, mother’s symptoms and medical problems during pregnancy. Infant measurements following are also included.

Background work involved

Some – clinic may provide these if not available.

How this information may help

Just a few possibilities: What’s the safest range for a mom’s phe levels during a pregnancy? Does it make a difference if phe levels are high in the beginning? Are babies’ born to PKU moms bigger or smaller than babies born to mom’s who do not have PKU?

Tips:

This survey can be updated at any time.
Measuring Your Mood

Time Required
Less than 10 minutes – This survey has 14 questions that deal with feelings or moods. It is intended for children over 8 years old with a parent completing based on their impressions of their children’s behavior or adults 18 or older who would complete for themselves.

Background work involved
None – answers are based on feelings or observation of feelings.

How this information may help
Information from this survey could be used to help identify the cause of mood disturbances and potential treatments.

Tips:
This survey can be updated at any time.

Version 1.0
10APR2017
Hummingbird IRB Approved
04/11/2017

Back to Index
Insurance Survey

Time Required
Less than 10 minutes – 6 questions asking about insurance coverage of PKU treatment.

Background work involved
Minimal or none - One questions asks about annual out of pocket expenses

How this information may help
Barrier to insurance coverage of PKU treatment is a major problem across the United States. Data from this survey could help improve efforts currently underway to mandate better coverage of PKU treatments.

Tips:
You will be asked to update this survey once per year.

Version 1.0
10APR2017
Hummingbird IRB Approved
04/11/2017

Back to Index
Research Trials Survey

Time Required
Less than 5 minutes – 2 yes or no questions asking of the participant currently or in the past has participated in clinical trials.

Background work involved
None – no dates requested

How this information may help
Provides a picture of where individuals can access clinical trials and help identify underserved areas in the region.

Tips:
You will be asked to update this survey once per year.

Version 1.0
10APR2017

Hummingbird IRB Approved
04/11/2017