



# The Parent Perspective on Long-Term Follow Up

**Christine S. Brown, MS**  
**Executive Director, NPKUA**  
**[christine.brown@npkua.org](mailto:christine.brown@npkua.org)**

# Pictures



8/16/05



11/19/07



# I Never Asked When Our Oldest was Born without PKU...



- Will he look “normal”?
- Can he go to school?
- Will he need special accommodations?
- Can he play sports?
- Can he travel to foreign countries?
- Can he go to college?
- Can he get a good job?
- Can he get married and have kids of his own?



National  
**PKU**  
Alliance

# PKU is the Success Story of Newborn Screening .... Or is it?

- 15,500 Americans with PKU
  - 8,000 in clinic
  - 7,500 are lost to follow-up
- Why???
  - 1970s: believed that the brain was fully developed by age 7 or 8 so there was no detrimental effect on PKU children to discontinue the diet



# National Collaborative Study

- National Collaborative Study (1976-1984) and Effects of Maternal PKU on Pregnancy Outcome (1984-2002):
- Loss of IQ
- Decline in school performance
- Increase in behavior issues
- Psychosocial issues
- Depression
- Phobias
- Epilepsy
- Tremors
- Pareses
- Maternal PKU Syndrome

**LTFU: Treatment for Life!!!**



# Medical Community Culture

- PKU is solved
- Diet for Life
- These kids are fine
- Let's move on...
- Little LTFU



## Survey Background

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- June 2015
- Survey Monkey
- 625 respondents
- 53% parents/caregivers of PKU children
- 47% adults



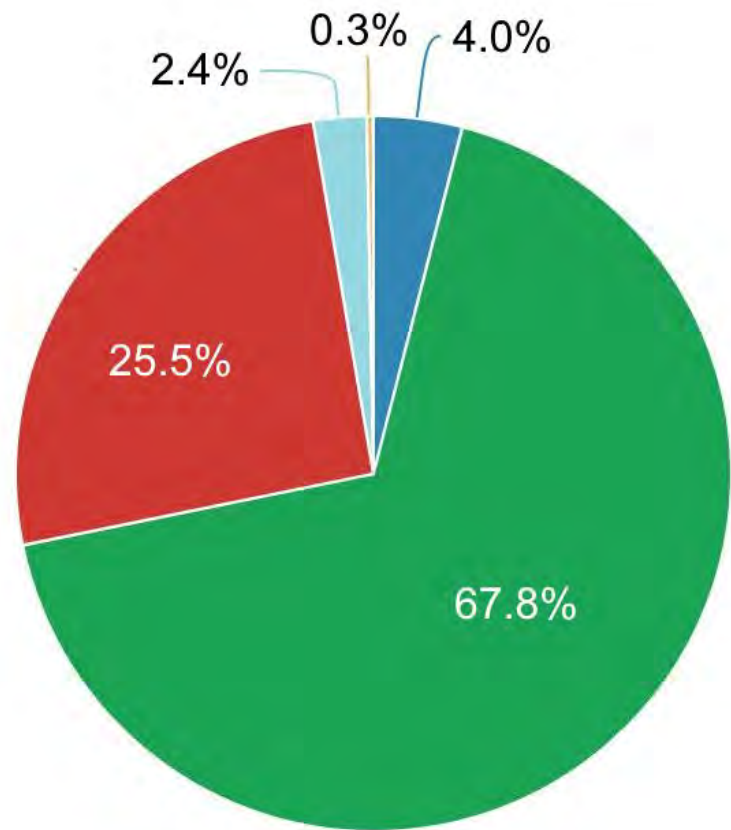
## Engaged Patients

### Engaged Patients






- Self selected
- Aware of the NPKUA
- Attend our meetings, involved in advocacy and in our educational programs
- 86% reported visiting a metabolic clinic in the last year for PKU care
- Only 8% said they hadn't visited a clinic in more than two years
- 61.8% said they had drawn their blood in the last month to monitor Phe levels

*Respondents overwhelmingly know what is needed to manage their PKU and have the access needed for optimal care in a clinic setting. However, challenges to dietary therapy were also evident.*



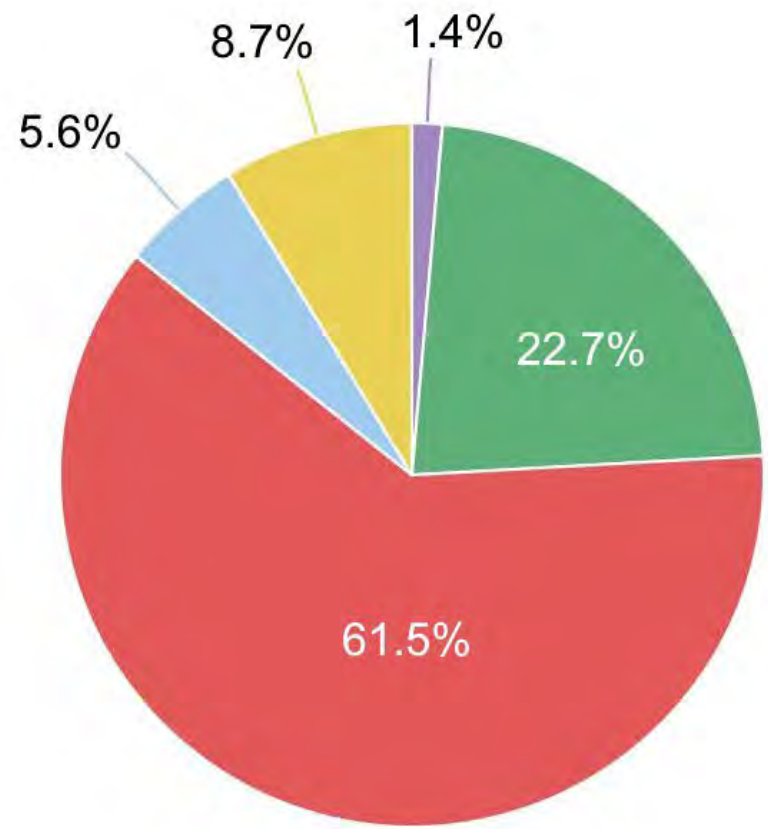







**Percentage of Blood Phe Values Within Range for Subjects with PKU less than 18 years of age (N=329)**

-  <2 mg/dL (120 μmol/L)
-  2-6 mg/dL (120-360 μmol/L)
-  >360 μmol/L
-  Not Known
-  Not Tested



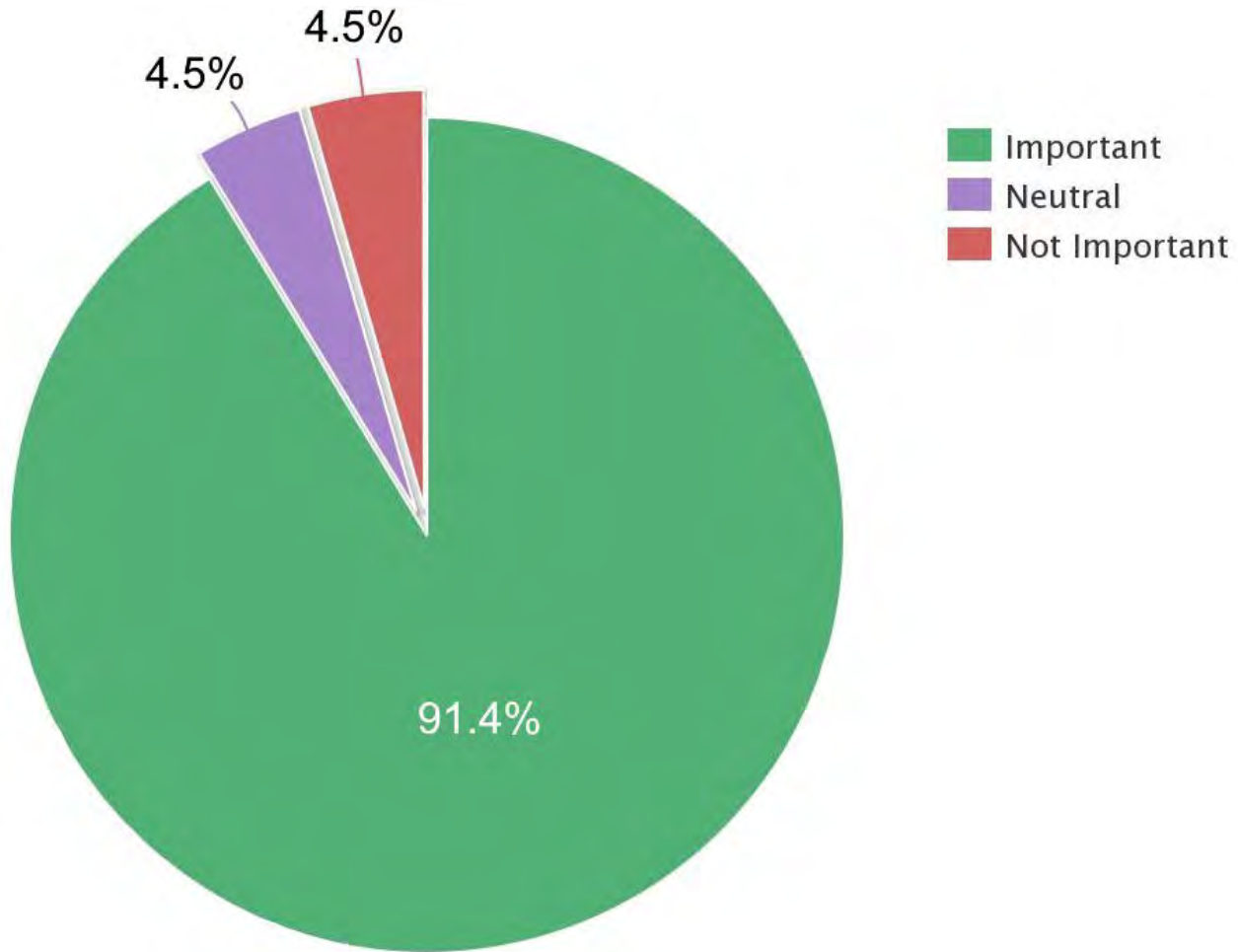
**Percentage of Blood Phe Values Within Range for Subjects with PKU more than 18 years of age (N=286)**



-  <2 mg/dL (120 μmol/L)
-  2-6 mg/dL (120-360 μmol/L)
-  >360 μmol/L
-  Not Known
-  Not Tested



### Percentage of Respondents Reporting on the Importance of the Development of New Treatments for PKU (N=474).



# Data

Table 1. Ranked Responses for Symptoms or Results were Most Desired when Considering New Treatments.

Preferences	Response Percent	Response Count
Drop in blood phe concentrations	87.5%	405
Attention span and ability to focus	65.7%	304
Executive function skills, such as the ability to plan, organize and prioritize	61.6%	285
Depression, anxiety and/or ups and downs in overall mood	55.1%	255
Processing speed - the ability to start and complete tasks	52.1%	241
Increase in energy	51.0%	236
Memory	49.5%	229
Lifting of "the fog"	43.0%	199
Bone loss	30.0%	139
Tremors	19.2%	89
Other damage such as muscle weakness, gait disorders, etc.	18.8%	87



## Dietary Therapy to Treat PKU is Effective, but not Optimal

- Dietary therapy doesn't control Phe levels within the recommended range for many
- Differences in white and gray matter in the brain
- Slight decrease in IQ, executive function, processing speed and emotional regulation when compared to siblings
- Anxiety, ADHD, depression rates higher



# Long Term Follow-Up is Needed!

*Saving babies lives does not end with the newborn screen... it is just the beginning*

