The Parent Perspective on Long-Term Follow Up

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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?
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

  - 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

• June 2015
• Survey Monkey
• 625 respondents
• 53% parents/caregivers of PKU children
• 47% adults
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)
Percentage of Blood Phe Values Within Range for Subjects with PKU more than 18 years of age (N=286)
Table 1. Ranked Responses for Symptoms or Results were Most Desired when Considering New Treatments.

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