Cognition and metabolic parameters in early-treated adult patients with phenylketonuria

Raphaela Muri\textsuperscript{1,2,3}, Stephanie Abgottspon\textsuperscript{1,3}, Michel Hochuli\textsuperscript{1}, Regula Everts\textsuperscript{1,4}, Roman Trepp\textsuperscript{1}

\textsuperscript{1}Department of Diabetes, Endocrinology, Nutritional Medicine and Metabolism, Inselspital, Bern University Hospital and University of Bern, Switzerland
\textsuperscript{2}Support Center for Advanced Neuroimaging (SCAN), University Institute for Diagnostic and Interventional Neuroradiology, Inselspital, Bern University Hospital, Switzerland
\textsuperscript{3}Graduate School for Health Sciences, University of Bern, Switzerland
\textsuperscript{4}Neuropediatrics, Development and Rehabilitation, University Children’s Hospital, Inselspital Bern

BACKGROUND

Phenylketonuria (PKU)

Consequences if untreated

- Rare inherited disease
- Deficient liver enzyme
- High Phenylalanine (Phe)
- Severe intellectual disabilities

Treatment

- DIET
- Avoid protein
- Mostly vegetables and fruits

ISSUE

Despite early-initiated and continuous treatment, adults with PKU still show mild cognitive abnormalities, especially in:

- IQ
- Executive functions
- Attention

HYPOTHESES

- Patients vs. controls:
  - Cognitive performance in patients

- In patients only:
  - Cognitive performance
  - Phenylalanine, Tyrosine (Tyr), and Phe:Tyr ratio

METHODS

Cognitive assessments

Amino acid profile (only patients)

- Phenylalanine
- Tyrosine
- Phe:Tyr ratio

Sample

<table>
<thead>
<tr>
<th>PKU</th>
<th>Controls</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age (Mean ± SD)</td>
<td>31.9 ± 9.0</td>
</tr>
<tr>
<td>Gender</td>
<td>11 female 12 male</td>
</tr>
<tr>
<td>Phe (Mean ± SD)</td>
<td>726.7 ± 206.0</td>
</tr>
<tr>
<td>Tyr (Mean ± SD)</td>
<td>42.4 ± 10.2</td>
</tr>
</tbody>
</table>
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4Neuropediatrics, Development and Rehabilitation, University Children’s Hospital, Inselspital Bern

RESULTS

PKU group
Control group

IQ

Executive functions

Attention

Metabolic parameters & cognitive performance

RESULT

1 Mean = 100, SD = ± 15 (see gray area)
2 Mean = 10, SD = ± 3 (see gray area)

* p < .05
** p < .01
*** p < .001
NS not significant

Phe & working memory
Phe & flexibility
Phe & sustained attention
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DISCUSSION

As hypothesized, patients show:

- intelligence score
- performance in attention
- performance in executive functions

No relationship between cognitive performance and concurrent Phe, Tyr, or Phe:Tyr ratio

Maybe other parameters such as lifetime Phe-levels are better predictors for cognitive performance

STUDY-TEAM

Prof. Dr. med. Regula Everts
Dr. med. Roman Trepp
PD Dr. med. Michel Hochuli
MSc. Stephanie Abgottspon
MSc. Raphaëla Muri
BSc. Gianni Ruschetti
BSc. Nathalie Schwab
BSc. Anna Wyss

COLLABORATORS

Prof. Dr. Bernadette Jansma
Prof. Dr. med. Roland Wiist
PD Dr. rer. nat. Christian Rummel
MSc. Michael Rebsamen
Prof. Dr. Roland Kreis
PD Dr. med. Yosuke Morishima
Prof. Dr. Shawn Christ
Prof. Dr. Nicolas Langer

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