Nutricia Metabolics Webinar Series

NLC
NUTRICIA LEARNING CENTER

Did you know?

Nutricia now offers a

Diet-for-Life Care Coordinator

for your adult patients with PKU

Sharon Ernst, MPH, RDN, CSP, FAND, a metabolic dietitian with over 25 years of clinical experience is serving as our Diet-for-Life Care Coordinator.

To learn more, please email us at
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NUTRICIA
Metabolics
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Part Three:

Returning to Diet: Cognitive, affective, and motivational factors for people with PKU

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Returning to Diet: Cognitive, affective, and motivational factors for people with PKU

June 28, 2017

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Disclosures

- BioMarin NCT01889862. Receives salary support from Boston Children's Hospital for assisting in the clinical trial.
- Volunteer – Next Step, Inc. in Cambridge, MA
- Volunteer – HCU Network North America

Learning Objectives

- Understand the cognitive and affective (anxiety & depression) issues associated with adults with PKU when not following a low-PHE diet
- Discuss the importance of health behavior changes in line with individual's values
- Implement strategies to help patients overcome these challenges

PKU: A Very Brief Primer

- Phenylketonuria (PKU) is an inborn error of metabolism that results from mutations of the phenylalanine hydroxylase (PAH) gene.
- The incidence of PKU varies by country and ethnic group. In the United States the incidence is 1 in 13,500 to 19,000 births.
- Loss of PAH results in increased concentrations of phenylalanine in the blood and brain.

1. Scriver & Kaufman in Scriver, et al (eds). McGraw-Hill, 2001.
2. National Institutes of Health Consensus Development Panel. *Pediatrics* 2001, 972-82.

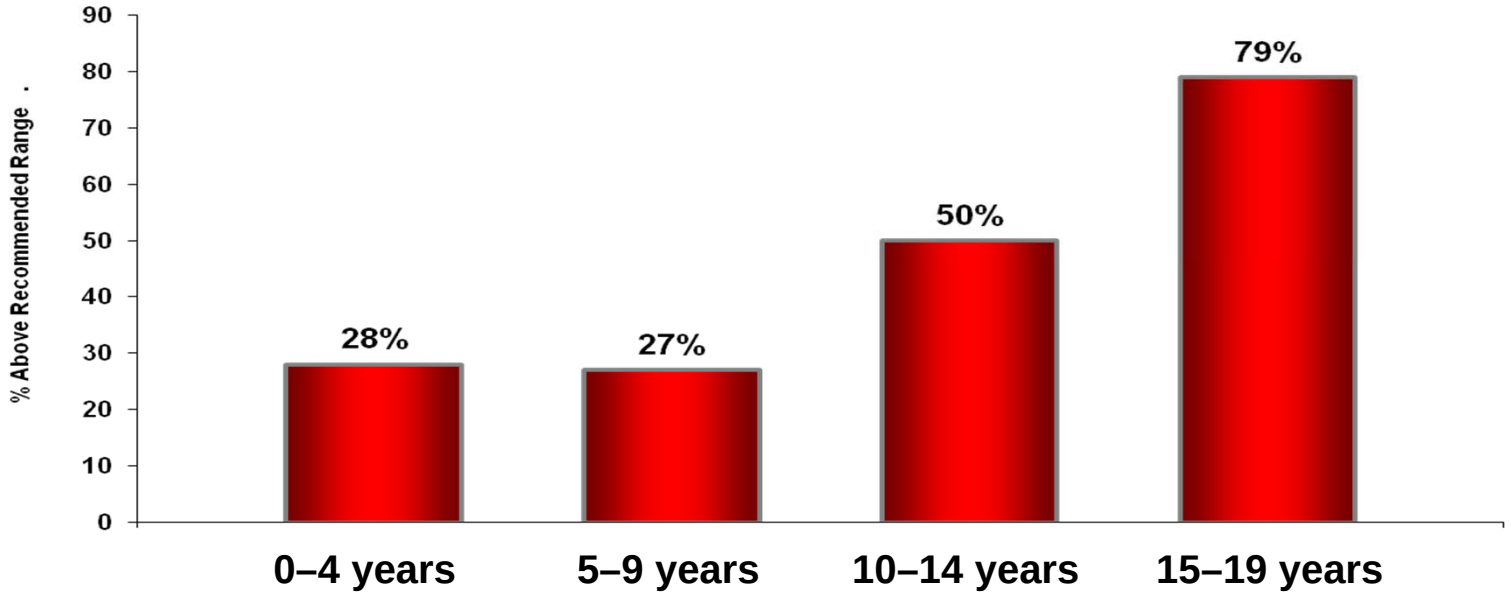
Mechanisms for PKU Pathogenesis

The mechanisms involved are still largely conjecture

- White matter disruption[1]
- Large neutral amino acid (LNAA) transport disruption[2]

1. **Mastrangelo et al. *Mol Genet Metab. 2015, 171-177***
2. **de Groot, et al. OJRD. 2013, 8:133**

Adherence to Diet is Difficult



Adapted from Walter JH, et al. *Lancet*. 2002;360:55-57.

PKU and Neurocognitive Functioning

1. High Phenylalanine (Phe) levels harm the brain

2. Traditional therapies do not completely protect individuals with PKU

3. Neurocognitive deficits make diet adherence more difficult

High PHE and Harm

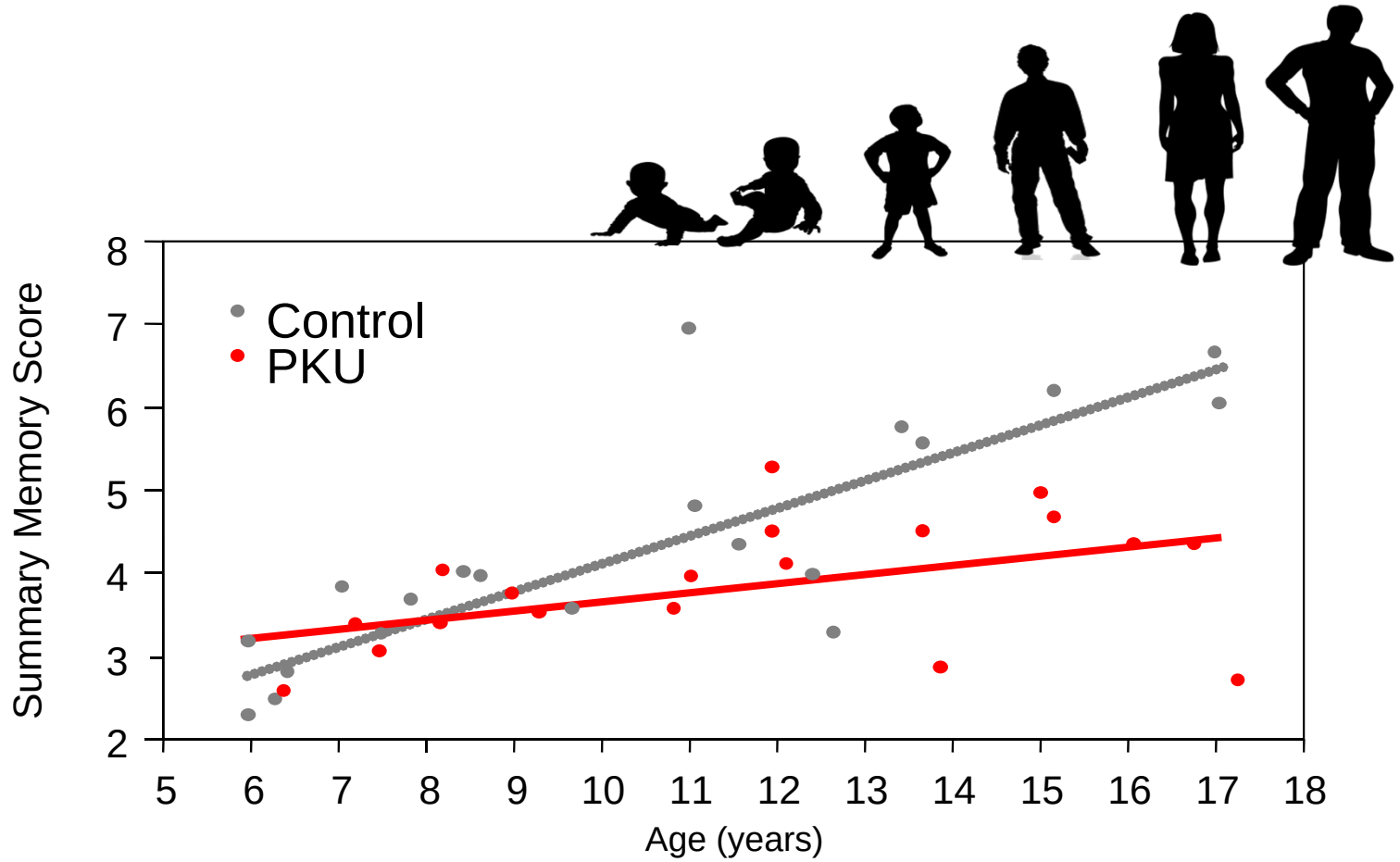
Blood Phe and IQ are Correlated in Individuals with PKU

Observation period	Correlation between blood Phe and IQ	Lifetime IQ loss for each 100 µmol/L (1.6 mg/dL) increase in blood Phe (IQ points)
Critical period (0–12 years old)	r = -0.35*	1.3–3.1
Lifetime (all ages)	r = -0.34*	1.9–4.1

*** *p* < 0.05**

Waisbren, et al. *Mol Genet Metab*. 2007;92:63-70.

Executive Deficits Across the Lifespan that May Worsen with Age



White DA, et al. *Neuropsychology*. 2001;15:221-229.

Areas of Executive Functioning

- Planning & Organization
- Response Inhibition
- Working Memory
- Problem Solving Strategies & Organizing information
- Processing Speed

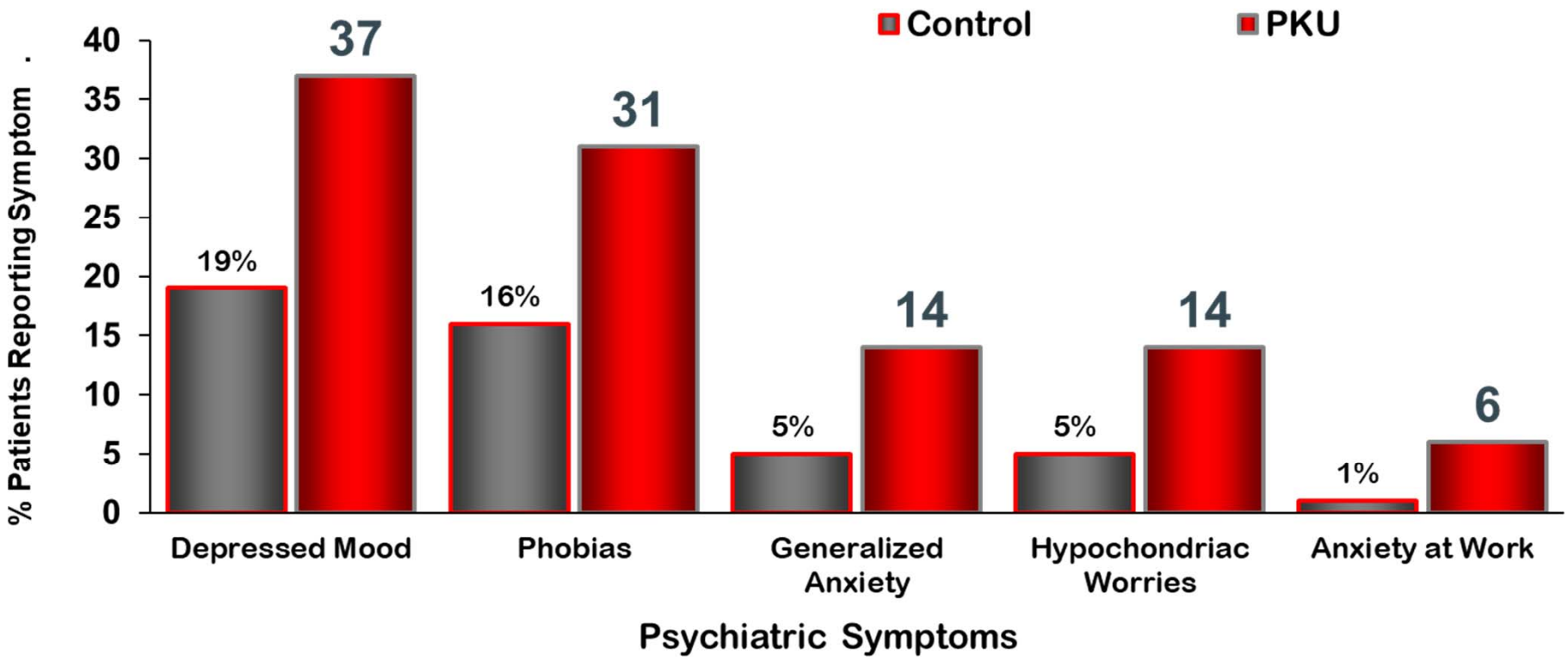
Jahja et al. *Mol Genet Metab.* 2013, S57-S61
Waisbren, et al. *Mol Genet Metab*. 2007;92:63-70.

PKU and Affective Symptoms

- Depression and anxiety are common and linked to non-adherence
- The earliest descriptions of PKU noted psychiatric problems.
- Anxiety, stress, and depression are heightened by lifetime and concurrent metabolic levels

DiMatteo, Lepper, & Croghan. *Arch Intern Med.* 2000, 2101-2107
Følling. *Physiol Chem.* 1934, 169-176
Clacy, Sharman, & McGill. *J Dev Behav Pediatr.* 2014, 388-391

Psychiatric Diagnosis



Adapted from Pietz J, et al. *Pediatrics*. 1997;99:345-350.

How to Help?
Build Identity Beyond Diagnosis

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How to Help?
Connect Diet to Identity and Values

Motivation frequently changes and is often formed in the context of relationships.

1. **R**esist the righting reflex
2. **U**nderstand your patient's motivations
3. **L**isten with empathy
4. **E**mpower the patient

Rollnick, Miller, & Butler. Motivational Interviewing in Health Care. Guilford Press

How to Help?
Specific Communication Examples

- Open questions – Closed questions
- Using a ruler
- Assess importance and confidence
- “What next?” Immediate concrete step.
- Summarize the patient’s words into themes

Rollnick, Miller, & Butler. Motivational Interviewing in Health Care. Guilford Press

How to Help?
Cognitive Functioning

- Assess ability/capability of adherence
  - Adaptive Functioning: Adaptive Behavior Assessment System (ABAS-3)
  - Executive Functioning: Behavior Rating Inventory of Executive Function (BRIEF)
- Address weak executive skills
  - Social support networks
  - Brief behavioral therapy
  - Stimulants used for ADHD?
  - Brain training websites?

How to Help?
Example of Building Executive Skills

1. Identify weak skills (e.g., emotion control)
2. Identify two to three recurring or repeating behaviors that indicate the deficit
3. Together, agree on a strategy and a cue to remind to use the strategy
4. Keep supports and supervision in place until mastery or success

Dawson & Guare. Smart but Scattered. Guilford Press

How to Help?
Affective Problems

- Manage metabolic levels
- Social support
- Psychotherapy
  - Behavioral Activation
  - Acceptance and Commitment Therapy
- Psychotropic medication

Additional information

New England Genetics Collaborative

https://www.gemssforschools.org/

New England Metabolic Consortium of Metabolic Programs

http://newenglandconsortium.org/

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THANK YOU!

Questions?

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