

Assessing Cognitive and Social-Emotional Functioning in Individuals with PKU: Tools for Use in the Metabolic Clinic by Psychologists and Non-Psychologists

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INTRODUCTION

Most metabolic clinics world-wide have adopted the “treatment for life” philosophy for PKU presented in the 2000 NIH Consensus Development Conference Statement.¹ This recommendation was based on evidence that elevated Phe levels adversely affect cognitive function and that individuals with PKU who adhere to recommended dietary restrictions have fewer medical and mental disorders. Since this time, researchers have provided further evidence for cognitive deficits and social-emotional problems in individuals with PKU that were treated early and continuously with diet alone, with elevated Phe, reconfirming the “treatment for life” philosophy.

NIH now suggests that “...This statement is more than five years old and is provided solely for historical purposes. Due to the cumulative nature of medical research, new knowledge has inevitably accumulated in this subject area in the time since the statement was initially prepared. Thus some of the material is likely to be out of date, and at worst simply wrong.”¹.

There is increasing evidence that cognitive and social-emotional problems are present in early and continuously diet-only treated individuals whose Phe levels are considered well-controlled (based on the NIH consensus statement). This evidence has led to a call for updated guidelines to address concerns that the current standard of care for PKU is inadequate.^{2,3} Van Spronsen and Bugard also expressed the opinion that new guidelines should include “a neuropsychological test battery that can be performed relatively easily in various centers throughout different countries with the aim of signaling sub-optimal outcomes”.

In December of 2008, a group of 10 psychologists and a psychiatrist with expertise in neuropsychological assessment and PKU convened to: (1) discuss evidence of cognitive deficits and social-emotional problems associated with PKU and (2) recommend a uniform neuropsychological and behavioral test battery that may be routinely administered in metabolic clinics with or without a staff psychologist.

ASSESSMENT TOOLS SELECTION PROCESS

Upon reviewing evidence of cognitive and social-emotional problems in PKU, the group deliberated to determine what best constitutes a uniform assessment battery for PKU. It was determined that an ideal assessment for use in all metabolic clinics would be:

- ▶ Applicable across the life-span
- ▶ Inclusive of psychological domains in which deficits most commonly occur in PKU (i.e., adaptive behavior, executive function and social-emotional function) due to well-supported literature of these dysfunctions in PKU
- ▶ Reliable and valid
- ▶ Easily administered by non-psychologists, because psychologists are rarely staff members in metabolic clinics
- ▶ Quick to administer
- ▶ Cost-efficient
- ▶ Able to establish and detect changes in baseline levels of function, indicate when additional testing and expertise are required, provide data to advance research

THE ASSESSMENT METHOD FOR PKU

Upon review of assessment tools using the above selection criteria, recommendations were made to develop and implement the Uniform Assessment Method for PKU. According to the Uniform Assessment Method, the following questionnaires are administered:

- ▶ For ages 0–2 years: Adaptive Behavior Assessment System-Second Edition (ABAS-II)⁴. The ABAS-II provides 9 measurements in 3 categories (conceptual, social, and practical) that combine to create an overall General Adaptive Composite (GAC).
- ▶ For ages 2–17 years: Behavior Assessment System for Children-Second Edition (BASC-II)⁶ and Behavior Rating Inventory of Executive Function (BRIEF)⁵. BASC-II includes emotional and behavioral scores related to childhood attention deficit disorders, anxiety, and depression. The BRIEF provides an overall score of executive function and subscale scores related to memory, attention, organization, and planning.
- ▶ For adults: BRIEF, Beck Anxiety Inventory (BAI)⁷, and Beck Depression Inventory - Second Edition (BDI-II)⁸ for adults. The BAI and BDI-II provide scores related to anxiety and depression in adults.

TABLE 1: UNIFORM ASSESSMENT METHOD FOR PKU SUMMARY

PSYCHOLOGICAL DOMAIN	INFANTS AND TODDLERS (0-2 years)	CHILDREN (>2 to <18 years)	ADULTS (18 + years)
Adaptive Behavior	ABAS-II	--	--
Executive Functioning	--	BRIEF-P or BRIEF	BRIEF-A
Social/Emotional Functioning	--	BASC-II	BAI & BDI-II

ABAS-II: Adaptive Behavior Assessment System-Second Edition, BRIEF: Behavior Rating Inventory of Executive Function, BRIEF-P: Behavior Rating Inventory of Executive Function Preschool Version, BRIEF-A: Behavior Rating Inventory of Executive Function Adult Version, BASC-II: Behavior Assessment System for Children-Second Edition, BAI: Beck Anxiety Inventory, BDI-II: Beck Depression Inventory-Second Edition

INTEGRATING THE UNIFORM ASSESSMENT METHOD INTO METABOLIC CLINICS

The recommended questionnaires may be administered each time the child or adult with PKU attends clinic in order to assess changes in functioning related to adjustments in treatment strategies, patient compliance, or elevated blood Phe levels. There are no practice effects and the age is taken into consideration because standard scores based on normative data are used. If an individual is determined to have problems, further evaluation by a psychologist or psychiatrist should occur. A physician or psychologist must be consulted immediately if responses indicate that a patient may be at risk for harming him/herself or others.

THE BENEFITS: IMPROVED PATIENT OUTCOMES AND A STANDARD RESEARCH TOOL

The Uniform Assessment Method for PKU addresses an unmet need in the care of individuals with PKU; namely diagnosing and monitoring deficits in cognitive and social-emotional functions. The Uniform Assessment Method will function as a longitudinal surveillance program to identify at-risk patients to be referred to health-care professionals who can better assess their deficits and serve their ongoing treatment needs. The selected screening measures may be administered at each clinic visit to assess changes in function related to metabolic status, age or treatment (e.g., Phe-restricted diet, food supplements or sapropterin dihydrochloride [BH4]).

Although there is an abundance of published literature on cognitive deficits and social-emotional problems in PKU, sample sizes within individual studies are small. Additionally, the assessment tools and control groups utilized are often different, making results difficult to compare across studies. The implementation of the recommended Uniform Assessment Method for PKU in metabolic clinics will provide a wealth of comparable data with a large sample size, bringing greater clarity to research questions such as the effects of treatment and the frequency of deficits in the PKU population. The requirement for demographically-matched control groups would also be of less concern, as each recommended assessment tool is validated and provides normative data based on large control groups.

CONCLUSION

Published evidence strongly suggests that an unmet medical need exists in the PKU population under current PKU treatment strategies, and that individuals with PKU are living with cognitive and social-emotional problems that are treatable. A paradigm shift in clinical management to focus on quality of clinical outcomes is required to address the cognitive and social-emotional dysfunction commonly seen in individuals with PKU and to enhance treatment. By implementing the Uniform Assessment Method for PKU, metabolic clinics can monitor the neurological and cognitive functioning of these patients and identify at-risk individuals with PKU who may benefit from psychological evaluations and possible interventions. In addition, this uniform assessment approach facilitates PKU research, as data may be pooled across multiple clinics using a consistent battery of reliable and valid assessment measures.

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