INTRODUCTION

Most metabolic clinics worldwide 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 Thus some of the material is likely to be out of date, and at worst simply wrong.”1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 Bugaj 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)5 and Behavior Rating Inventory of Executive Function (BRIEF)6. 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.

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