

Neuropsychology and Metabolic Conditions: The Neurocognitive Profile of FOD/OAA and the benefits of neuropsychological assessment

Christopher Boys, PhD, LP
Pediatric Neuropsychologist
Associate Professor of Pediatrics

Clinical Neuropsychology

- Concerned with the behavioral expression of brain functions and brain dysfunction

Four different purposes of Neuropsychological Assessment

- Diagnosis
- Patient Care and Planning
- Rehabilitation and Treatment Evaluation
- Research

Diagnosis

- Can be useful in discriminating between psychiatric/behavioral and neurological symptoms
- Identifying a possible neurological disorder in a nonpsychiatric patient
- Helping to distinguish between different medical/neurological conditions (ADHD vs Seizure)
- Screening for identifying persons most likely at risk for some specified conditions (difficulties following prematurity; Fetal Alcohol Spectrum Disorders; Metabolic conditions)



Patient Care and Planning

- Patients referred for detailed information about their cognitive status and personality characteristics
- Precise descriptive information about cognitive and emotional status is essential for management
 - Effective planning usually depends on an understanding of a patient's capabilities and limitations;
 - the kinds of psychological changes they are undergoing;
 - and the impact of these changes on their experiences of themselves and on their behavior



Research

- Neuropsychological Assessment has been used to study the brain function and its translation into behavior,
- Also, investigating specific brain disorders and subsequent behavior/learning profiles
- Research in pediatric neuropsychology includes longitudinal assessment of subtle neurocognitive functions and their impact over time
 - PKU/metabolism
 - NICU
 - Case Conceptualization in specific disorders

Goals of Neuropsychological Assessment

- Conceptualization of Strengths and Weaknesses
- Assessment should drive interventions
- Serial assessment to insure interventions are effective and to monitor trajectory

Domains of Neuropsychological Assessment

- Cognitive/Intelligence
- Academic Achievement
- Attention
- Memory
- Language
- Executive Functions
- Emotional/Behavioral
- Adaptive Behavior

U of MN Metabolism Program

- 12 months and 24 months
 - Bayley Scales of Infant and toddler Development
- 4 years
 - Wechsler Preschool & Primary Scales of Intelligence (WPPSI) (cognitive)
 - CELF-Pre2 (language)
 - CBCL (emotional/behavior)
 - Vineland (adaptive behavior)

Metabolism Protocol (cont.)

- 6 years old
- 8-9 years (3rd-4th grade)
- 11-12 years (6-7th grade)
- 15 years
- 17-18 years
 - Focus on transition

Organic Acidemia

- Longitudinal Research regarding the neuropsychological functioning of children with OA has been quite limited.
 - Of the studies looking at OA, most have examined the functioning of children with CblC. Results of these studies varied with some reporting that children with CblC are intellectually disabled (e.g., Beauchamp et al 2009; Weisfeld-Adams et al 2013)
 - and others showing evidence of highly variable neuropsychological profiles (Bellerose et al 2016).
 - Only one study was identified that longitudinally examined the neuropsychological functioning of children with CblC. Beauchamp and colleagues (2009) presented one child who demonstrated persistently impaired intellectual abilities (IQ below 52) between the ages of five and twelve and a second child who demonstrated declining intellectual abilities between ages seven and twelve (IQ of 79 at time 1 and 59 at time 2).

Fatty Acid Oxidation Disorders

- With earlier identification and preventative treatments, mortality and morbidity rates have improved. However, in the absence of severe health and neurological effects from these disorders, subtle developmental delays or neuropsychological deficits have been noted. (Waisbren, Landau, Wilson, & Vockley, 2013)
 - Overall, 54% of these children identified through newborn screening experienced developmental challenges.
 - Speech delay or relative weakness in language was noted in 31% of children and motor delays were noted in 29%.
 - The majority of the children receiving psychological evaluations performed well within the average range with only 11% scoring <85 on developmental or intelligence tests.
 - These results highlight the importance of screening children with fatty acid oxidation disorders to identify those with language, motor, or cognitive delay.



Executive Functions

- Frontal Lobe Functioning
 - Not fully developed until 18-23 yrs
 - Have to be concerned about skills vs abilities
- Executive Functions
 - Allows an individual to deploy their skills
 - “Absent Minded Professor”
 - Trying Differently, Not Harder
- Often the reason for the 4th grade slump

Executive Functions

- Monitoring
- Inhibitory Control
- Shifting
- Working Memory
- Attention
- Planning/Organization

Executive Functions

- Working Memory
- Processing Speed
- Cognitive Flexibility/Shifting

Executive Functions

- Children with executive functions issues must be prepared to receive directions/instructions.
 - This allows for time to shift to new cognitive set or activity
 - Notable source of frustration for parents and teachers
 - This is where the child gets blindsided
 - Adults are on 3rd command, Child is on first
- Deficits in cognitive flexibility also will warrant specific accommodations. Child will need to be reminded to ‘stop and think’ before responding to task demands, and he will probably need cueing to keep him from continuing to respond in ways that are ineffective. Parents/teachers also should monitor child closely to insure that he understands directions for assignments.

Working Memory

- Working memory is essentially the ability to hold information in memory and perform a specific manipulation to the information.
- Individuals with working memory problems often have difficulty carrying out multistep activities, losing track of what they are doing as they work, or forgetting what they are supposed to retrieve when sent on an errand.
 - Restaurant Servers

Working Memory(cont.)

- Provide **simple templates** for routines that are repeated.
 - Each day must start fresh, regardless of how the previous day went.
- A template lays out the standard steps to complete a repetitive task and can be useful for a variety of home and school tasks. The template can be faded out when the procedure or task becomes automatic.
- However, this should be monitored closely so that the template can be brought back if it appears that it was faded too soon. The template can also be used to address problem areas such as homework completion, personal hygiene, time management (get a snack, math worksheet etc.).
- **IMPORTANT STEP:**
 - May need a reminder/behavior change component to *remember* to check the list

Summary

- Newborn Screening has helped to secure cognitive ability in a lot diagnosis;
- However, a subtle neurocognitive profile can remain that can be frustrating for kids, parents, and educators
- For more impacted children, assessment can provide good conceptualization and assist in planning interventions.