The Neuropsychological Evaluation in the Assessment of Autism Spectrum Disorders

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Disclosures

• Danielle N. Ager, PsyD – Nothing to disclose
Following this session:

• Participants will be able to identify core components of a neuropsychological evaluation.
• Participants will understand the benefits of using the neuropsychological evaluation with autism spectrum disorder.
• Participants will better understand etiology and brain regions implicated in autism spectrum disorders.
• Participants will identify the expected profiles for autism spectrum disorder on the neuropsychological evaluation.
What is a Neuropsychological Evaluation?

▶ “Clinical neuropsychology is an applied science concerned with the behavioral expression of brain dysfunction.” ~ Lezak, Howieson, & Loring

▶ Links brain functioning with behavior

▶ Can be useful in distinguishing between psychiatric and neurological conditions

▶ Descriptive / qualitative evaluation that can provide useful information to tailor an individual’s treatment based on strengths and weaknesses
Components of a Neuropsychological Evaluation

- Intellectual Functioning
- Language
- Visual Perception
- Attention
- Executive Functioning
- Processing Speed
- Memory
- Motor Skills
- Behavior / Emotional Functioning
DSM–5 Diagnostic Criteria for Autism

- Persistent deficits in social communication and social interaction across multiple contexts
  - Deficits in social–emotional reciprocity
  - Deficits in nonverbal communicative behaviors used for social interaction
  - Deficits in developing, maintaining, and understanding relationships

- Restricted, repetitive patterns of behavior
  - Stereotyped movements
  - Insistence on sameness / inflexible adherence to routines
  - Restricted interests that are abnormal in intensity/focus
  - Hyper or hyposensitivity to sensory input
Etiology of Autism

- 85% → Unknown etiology
  - 30% → Dysmorphic features and/or brain abnormalities
  - 70% → No physical abnormalities
- 15% → Known genetic abnormality

(www.genome.gov)
Genetics and Autism

- First evidence – twin study in 1970s
- 60% concordance rate in monozygotic twins
- 3% concordance rate in dizygotic twins
- Higher rate of autism among siblings (compared to general population)
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- 60% concordance rate in monozygotic twins
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- Higher rate of autism among siblings (compared to general population)
- HOWEVER → no single gene or group of genes has been linked to autism
  - There may be upwards of 15 interacting genes linked to the autism phenotype
  - 900 or more genes are abnormal in ASD
Genetics and Autism

- 5 percent of children diagnosed with autism have a known chromosomal abnormality
  - Type and extent of chromosome abnormality varies greatly
  - X chromosome involvement (majority of those w/ autism are male)
  - Most consistent association is 15q11–q13, 17q11–21, 2q24–31
  - Also linked w/ 7q, 16p, 2q, 17q
Genetics and Autism

- Other syndromes with comorbid ASD
  - Retts syndrome
  - Tuberous sclerosis
  - Angelman syndrome
  - 22q deletion syndrome
  - Joubert syndrome
  - Potocki–Lupski syndrome
  - Smith–Lemili–Opitz syndrome
  - Timothy syndrome
  - Fragile X syndrome
  - Epilepsy
Environmental Factors and Autism

- Teratogens may be related to autism
  - Maternal rubella infection
  - Maternal ethanol use
  - Exposure to valproic acid in utero
Brain Regions Implicated in Autism

- Primary cortical features
  - Small neuronal cell size
  - Increased neuronal density
  - Megalencephaly
  - Altered organization of “minicolumn” structure
Brain Regions Implicated in Autism

Many children with autism have anomalies in some of the brain structures shown below. Malformations in these areas can lead to symptoms of autism.

- **Frontal lobes**: Involved in planning functions.
- **Hippocampus**: Memory and learning area.
- **Cerebellum**: Involved in motor skills and shifting attention from one task to another.
- **Amygdala**: Involved in emotion.
- **Temporal lobe**: Auditory, speech and memory centers.
Neurobiological Findings

- Dramatic increase in brain volume (1\textsuperscript{st} 2 years of life)
Amygdala Theory of Autism

- Fewer neurons in amygdala
- Fewer neurons in the lateral nucleus of amygdala
- Small clusters of tightly packed neurons in the medial nuclei of the amygdaloid complex
- Reduced activation of the amygdala amidst increased activity and greater reliance on other brain regions
Hippocampus

- Critical in formation of memories
- Enlarged in ASD compared to neurotypical peers
- Smaller than expected neurons and increased cell-packing density
Cerebellum

- Involved in motor coordination/control, integration of sensory perception, but also in many other brain processes (language processing)
- Reduced size and number of Purkinje neurons in the vermis
- Atypical volume in cerebellum
Mirror Neurons

- Neural substrates important in perspective taking, imitation, and empathy in ASDs
- Rizzolatti’s group found that the same nerve fired when a monkey reached to pick up an object or when the monkey simply viewed a human picking up the object
- In humans also involves understanding the intentions of others, experience the emotions of others
- fMRI → children with ASDs showed a lack of mirror neuron activity in the pars opercularis of the inferior frontal gyrus, which was negatively correlated with social impairment
Why do a neuropsychological evaluation with this population?

▶ Identify common comorbidities
▶ Differential diagnosis
▶ Determine strengths and weaknesses
▶ Inform the treatment plan
▶ Establish a baseline and document changes in performance
Cognitive Functioning

- 50–70% of individuals with ASD have comorbid intellectual disability (IQ below 70)
- Cognitive ability is a good predictor of social behavior / social outcomes
Cognitive Functioning

- Children with low IQ tend to have lack of interest in peers
- Children with average IQ may show social awkwardness, immaturity, rigidity, which are impediments to making friendships, even when they desire friendships
- Children with low IQ may show multiple stereotypical repetitive behaviors, unusual visual behaviors
- Children with average IQ may be resistant to changes in routines, may be preoccupied with unusual topics (restricted interests)
Adaptive Functioning

- Skills and behaviors necessary for age appropriate day to day functioning
  - Such as communication, self care, community use
- To have diagnosis of ID, must also have adaptive deficits
- In children with ASD, adaptive functioning is often significantly impaired, especially on measures of social skills and social communication
Attention Skills

- Different types → sustained, shifting, selective
- In ASD, there are both strengths and weaknesses in the attention domain
- Individuals with ASD:
  - Tend to orient to a new stimulus much more slowly
  - Show impairment in shifting attention
- Orienting to social stimuli (e.g., someone calling a child’s name) is particularly impaired in ASD
Language Skills

- Impairments in communication are common in ASD
- Unusual aspects of language are present, such as echolalia
- Prosody and pragmatics are impaired in ASD with relatively spared phonology and syntax
- Delayed development of language (e.g., first words, speaking in phrases)
Motor Skills

- Children with ASD exhibit notable deficits in fine and gross motor functioning
- Can have significant motor developmental delays
- Hypotonia is commonly comorbid with ASD as well as unusual postures
Executive Functioning

- Complex mental processes such as response inhibition, cognitive flexibility, planning, and problem-solving skills
- Individuals with ASD:
  - Deficits in set shifting and planning, switching and strategy
  - Typically demonstrate broader deficits and more impaired EF skills compared to other neurodevelopmental conditions
  - Deficits in executive skills are associated with deficits in social behavior, communication, and adaptive skills in ASD
Memory

- Includes: visual memory, verbal memory, short term, episodic, rote, and facial memory

- Individuals with ASD:
  - Verbal memory impaired; visual–spatial memory is intact
  - Facial memory and memory for social situations is impaired
Academic Functioning

- Individuals with ASD:
  - May perform below expectations on measures with a comprehension component
  - May show strengths in decoding or basic word identification but have problems with reading comprehension
  - Writing tends to be a weakness
Visual Spatial Skills

- Typically considered an area of strength in ASD
  - Enhanced discrimination, greater aptitude in feature detection, and memory for simple visual stimuli may explain the superior performance in VS skills
Social / Emotional Functioning

- Social skills deficits is one of the core diagnostic criteria for ASD
- Impaired processing of emotions also common in ASD
- Deficits in theory of mind
Social / Emotional Functioning

- ADOS–2
  - Diagnostic measure of current behaviors that are indicative of autism
  - Contains specific tasks eliciting social and play interactions that are then coded on a behaviorally anchored scale
  - Allows for the standardization and systematic observation of behaviors and symptoms common in ASD
Case Example
Meet Timothy

- 6 year old boy who was previously diagnosed with ASD
- Referred by his pediatrician to help with treatment planning due to concerns about ADHD

**Presenting symptoms**
- Unclear speech
- Problems understanding language, mixes up the order of words when speaking, speech regression
- Problems with eating and dressing
- Poor eye contact
- Tends to overreact to neutral social interactions, problems interacting with peers
- “Aggressive” at home, particularly in response to disciplinary measures
- Anxiety triggered by loud noises/crowds
- Restless, trouble sitting still
Meet Timothy

• **Developmental history**
  • Born full term, pregnancy complicated by 2-vessel umbilical cord
  • Mother first noticed problems at 10 months of age, temper tantrums lasting up to 60 minutes.
  • Spoke first words at 14 months, short phrases at 2 years, walking at 10 months of age, toilet trained by 3 years.
  • Diagnosed with autism at 3 years of age following comprehensive evaluation, presented with lack of interest in peers at that time.

• **Medical history**
  • RSV at 3 weeks old, hospitalized, no complications

• **Educational history**
  • Self-contained classroom with an IEP for ASD

• **Behavioral Observations**
  • Did not speak to examiner during testing, no eye contact
  • Flat affect, did not smile once
  • Very active during testing, moving about the room, needed many breaks
Measures Administered

• **Intellectual Functioning**
  - Ireton Child Development Inventory (CDI)
  - Wechsler Intelligence Scale for Children, Fifth Edition (WISC-V, selected subtests)

• **Language**
  - Peabody Picture Vocabulary Test, Fourth Edition (PPVT-4)

• **Visual Perception**
  - Beery-Buktenica Developmental Test of Visual-Motor Integration, Sixth Edition (Beery-VMI)

• **Attention / Executive Functioning**
  - Behavior Rating Inventory of Executive Functions, Second Edition (BRIEF-2)
  - Children’s Category Test, Level 1 (CCT)

• **Memory**
  - Wide Range Assessment of Memory and Learning, Second Edition (WRAML-2, selected subtests)

• **Motor Skills**
  - Grooved Pegboard
  - Hand Dynamometer

• **Academic Skills**
  - Bracken School Readiness Assessment, Third Edition (Bracken)
  - Wechsler Individual Achievement Test, Third Edition (WIAT-III, selected subtests)

• **Adaptive Skills**
  - Adaptive Behavior Assessment System, Second Edition (ABAS-II; Parent Form)

• **Behavior / Emotional Functioning**
  - Gilliam Autism Rating Scale, Third Edition (GARS-3)
  - Behavior Assessment System for Children, Second Edition (BASC-2; Parent report)
Results – Intellectual Functioning

On an overall measure of his nonverbal intellectual skills, he scored in the Average range (WISC-V, Nonverbal Index = 109, 73rd percentile). Among the index scores that were able to be calculated, that comprised the overall scores, there was variability in TTT’s skills. Specifically, Timothy’s understanding of visual-spatial relationships showed High Average skills [Visual Spatial Index (VSI), 77th percentile]. His fluid knowledge was also in the High Average range [Fluid Reasoning Index (FRI), 88th percentile]. In contrast, TTT scored in the Borderline range on tasks assessing information processing speed [Processing Speed Index (PSI), 2nd percentile].

In addition to formal cognitive test measures, Timothy’s mother completed an inventory evaluating his overall level of development compared to same-aged peers. On this measure, Mrs. Mom endorsed a developmental delay with a 3 year, 7 month age equivalent (CDI, General Development, >30 percent below age level).
Results - Language

It was difficult to assess Timothy’s expressive language skills due to his limited verbal responsiveness during the assessment. His receptive vocabulary skills were Low Average (PPVT-4, 18th percentile). On an inventory assessing Timothy’s expressive and receptive language skills (CDI), Mrs. Mom endorsed developmental delays in both domains. Specifically, her ratings of Timothy’s Language Comprehension skills were consistent with a 2 year, 9 month age equivalent (>30 percent below age level). With regard to Expressive Language, Mrs. Mom’s ratings fell at a 3 year, 2 month old age equivalent (>30 percent below age level).
Results – Visual Perception

On a timed measure, his block construction skills fell within the Average range (WISC–V, Block Design, 50th percentile). He scored within the Average range on an untimed visual motor task in which he was required to copy various designs that increased in complexity (Beery–VMI, 34th percentile). Upon further analysis of the component skills required for effective drawing, Timothy’s visual perceptual skills were Average (Beery–VMI Visual Perception, 70th percentile). However, his motor coordination skills fell in the Low Average range (Beery–VMI Motor Coordination, 13th percentile). On a timed paper and pencil measure assessing Timothy’s visual motor coordination and speed, he performed within the Average range (WISC–V, Coding, 25th percentile) when required to transcribe abstract symbols. Of note, Timothy attempted to hold the response booklet in the air and respond during this task.
Results – Attention and Executive Skills

On measures of visual attention and working memory, Timothy’s performance fell within expectations for his age. On a task that involved more complex executive skills, he scored in the Average range (Children’s Category Test, Level 1, 50th percentile). This task incorporates deductive reasoning, cognitive flexibility, memory, novel visual/verbal problem solving, and requires Timothy to detect concepts underlying visual arrays presented to his and incorporate examiner feedback about the accuracy of his response choices to guide his decision making. On a measure of visual working memory skills in which Timothy viewed pictures and then selected the sequence in which the pictures were presented to him, he scored in the Average range (WISC-V, Picture Span, 37th percentile). However, on a measure of visual attention and speed, Timothy performed within the Extremely Low range on a measure of visual scanning skills in which he scanned rows to find matching symbols (WISC-V, Symbol Search, <1st percentile).

In addition to formal measures of attention and executive functions, Timothy’s mother completed a rating scale relating his skills in this domain (BRIEF-2). The parent profile was deemed valid, consistent, and not overly negative in perceptions of his behavior. Mrs. Mom endorsed Clinically Elevated ratings on scales assessing his ability to regulate his own emotions, and to shift his attention between activities. In addition, there were Mildly Elevated ratings on scales assessing Timothy’s ability to inhibit impulses; to initiate his own involvement in activities; to use working memory skills; and to plan and organize himself. Remaining scales were within expectations for his age and gender, including his ability to organize his belongings; to evaluate his own work; and his capacity to observe and evaluate his own behavior.
In the visual memory domain, Timothy showed Low Average overall skills (WRAML-2, Visual Memory Index, 12th percentile). His ability to replicate small geometrical designs immediately after very brief presentation was Average (WRAML-2, Design Memory, 25th percentile). After a delay, his ability to recognize the designs among distracters was Low Average (WRAML-2, Design Recognition, 9th percentile). He also scored in the Low Average range when he studied pictures of everyday scenes and identified changes when presented with slightly altered versions (WRAML-2, Picture Memory, 9th percentile). Following a delay, he was able to recognize elements of the pictures with Low Average skills (WRAML-2, Picture Memory Recognition, 9th percentile).
Results – Academic Skills

On a measure of academic readiness, his acquisition of basic concepts, such as identifying colors, letters, numbers, sizes and shapes, was assessed. Overall, Timothy’s mastery of basic concepts indicating school readiness was in the Average range (Bracken, School Readiness Composite, 66th percentile). Qualitatively, he correctly identified 10 of 10 colors, 15 of 15 letters, 18 of 18 numbers, 15 of 22 sizes/comparisons, and 20 of 20 shapes.

In the area of mathematics, Timothy’s ability to compute various types of math problems fell in the Average range (WIAT-III, Numerical Operations, 70th percentile). With regard to written expression skills, his ability to quickly write letters of the alphabet was also Average (WIAT-III, Alphabet Writing Fluency, 47th percentile).

On an inventory assessing Timothy’s knowledge of basic concepts, Mrs. Mom’s ratings of his mastery of Numbers fell at a 5 year old age equivalent (CDI, at age level), and her ratings of his mastery of Letters also fell at a 5 year old age equivalent (CDI, at age level).
Results – Motor Skills

Timothy’s grip strength was Average in his dominant right hand and High Average in his non-dominant left hand grip strength was Low Average (Hand Dynamometer, Dominant Hand, 47th percentile; Non-Dominant Hand, 84th percentile).

Timothy’s fine motor coordination when placing pegs into a pegboard under the pressure of time was in the Extremely Low range on the right and left hand unimanual trials (Purdue Pegboard, Dominant Hand, <1st percentile; Non-Dominant Hand, <1st percentile). On the bimanual trial in which he was required to place the pegs into the board simultaneously with his right and left hands, Timothy scored in the Superior range (Purdue Pegboard, Bimanual Trial, 95th percentile).

On a developmental inventory, Mrs. Mom endorsed Fine Motor skills that fell at a 3 year, 1 month age equivalent, showing a developmental delay (CDI, Fine Motor, >30 percent below age level), and her ratings of TTT’s Gross Motor skills were also developmentally delayed, falling at a 2 year, 6 month old age equivalent (CDI, Gross Motor, >30 percent below age level).
Results – Social / Emotional Functioning

Mrs. Mom completed a broadband rating scale that provided valid and reliable descriptions of TTT’s current emotional and behavioral functioning (BASC-2). Timothy’s mother endorsed a single Clinically Significant elevation on the Atypicality domain, indicating the presence of strange or odd behavior and that he may seem disconnected from his surroundings. There were also At-Risk elevations on the following clinical scales: Hyperactivity, Aggression, Depression, Withdrawal, and Attention Problems.

On an inventory that gauges Timothy’s Social Skills, Mrs. Mom endorsed a developmental delay, with a 2 year, 5 month old age equivalent (CDI, >30% below age level).

On the GARS-3, Mrs. Mom rated Timothy’s behaviors as having a Very Likely probability of autism (GARS-3, Autism Index, 25th percentile). Mrs. Mom’s ratings on subscales geared toward restricted behavior patterns; poor social communication and social interaction skills; unusual speech patterns; a rigid, intense cognitive style; and excessive emotional sensitivity were not consistent with this diagnosis (GARS-3, Restricted/Repetitive Behaviors, 16th percentile; Social Communication, 25th percentile; Social Interaction, 37th percentile; Maladaptive Speech, 16th percentile; Cognitive Style, 63rd percentile; Emotional Responses, 63rd percentile).

A clinician rating scale geared toward identifying features of an autism spectrum diagnosis showed Mild-to-Moderate Symptoms of Autism Spectrum Disorder (CARS-2, 35th percentile).
Results – Adaptive Skills

Overall, Mrs. Mom’s ratings fell within the Average range for Timothy’s age (ABAS-III Parent Form, General Adaptive Composite, 55th percentile). In particular, there were impairments (in the Borderline range) on scales assessing: Timothy’s social skills; and his personal care skills, such as dressing and maintaining hygiene.

On BASC-2 adaptive scales, Mrs. Mom’s ratings revealed Clinically Significant elevations on the Adaptability, Activities of Daily Living, and Functional Communication scales. These ratings indicate that Timothy has extreme difficulty adapting to changing situations, that he has difficulty performing simple daily tasks safely, and that he has difficulty communicating to get his needs met. There were also At-Risk elevations in the parent ratings on the Social Skills and Leadership scales, highlighting problems with social communication as well as difficulty making decisions and being creative in group settings.

On another developmental skills inventory, Mrs. Mom rated Timothy’s overall adaptive skills as developmentally delayed, falling at a 3 year, 1 month old age equivalent (CDI, >30 percent below level).
Diagnoses

- Autism Spectrum Disorder, Level 1, Requiring Support
- Attention Deficit Hyperactivity Disorder, Combined Type
- Selective Mutism
Recommendations

- Medication for hyperactivity/inattention
- Home based therapy for behavioral management
  - Applied Behavior Analysis with goals including:
    - Development of more functional communication
    - Social skills
    - Problems adjusting to changes
    - Daily living skills
    - Improving self-regulation skills
    - Decreasing undesirable behaviors
  - Parent training on ABA techniques to help w/behavior
Recommendations

- Occupational Therapy
  - Sensory integration therapy

- Speech therapy
  - Address problems with functional communication, selective mutism, and problems with social communication skills.

- Extracurricular activities

- Hippotherapy

- Social Skills Training in a small group setting
Recommendations

School based services
- Alter the IEP to include 2 exceptionality classifications: ASD and Other Health Impairment (OHI)
- Inclusion classroom but co-teaching setting
- One to one aide, if has behavior difficulties in school
- Minimize transitions
- Preferential seating
- Sensory breaks, frequent breaks
- Multi-modal teaching techniques
Take Home Messages

- There are several brain related abnormalities implicated in autism spectrum disorders.
- A neuropsychological evaluation is designed to measure different areas of brain functions.
- There is no one single neuropsychological profile for an individual with autism.
- Autism falls on a spectrum, with a wide array of behavioral presentations, and therefore the assessment of autism should be more comprehensive.
- Conceptualizing ASD in terms of many facets of functioning using a neuropsychological evaluation allows different and potentially more meaningful interpretations for these disorders.
- When specific deficits and areas of strengths are identified, the treatment plan can be tailored for an individual.
References