The Neuropsychological Evaluation and What It Can Tell Parents

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Children’s Healthcare of Atlanta
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Overview

- Introduction
- What is Neuropsychology?
- What is a Neuropsychological Evaluation?
- What can the Neuropsychological Evaluation tell parents?
- Specific implications of a neuropsychological evaluation for individuals with X and Y chromosome variations
Children’s Healthcare of Atlanta (CHOA)
Children’s Healthcare of Atlanta (CHOA)

• One of the largest pediatric clinical care providers in the U.S.
• 3 Hospitals & the Center for Advanced Pediatrics
• Marcus Autism Center
• 20 neighborhood locations
• 43 telemedicine locations around Georgia
Neuropsychology

A child with cognitive or behavioral problems often needs special attention and help. Our team is specially trained to work with children and teens that have unique challenges and needs. We collaborate with specialists from across our system to deliver care.

Meet our neuropsychology team
CHILDREN'S HEALTHCARE OF ATLANTA

CHOA Department of Neuropsychology

- 12 Pediatric Neuropsychologists
- Collaborate with many departments
  - Genetics, Neurology, Neurosurgery, Rehabilitation, etc.
- Birth to 21 years
- Wide variety of neurological & medical conditions
  - Developmental (Genetic, prematurity)
  - Acquired (Brain injury, stroke)
- Clinical care, research, and training
eXtraordinarY Clinic - Atlanta

• Collaboration with the eXtraordinarY Clinic at Emory
• Multidisciplinary focus includes:
  – Genetics, Nursing, Urology, Endocrinology, Neuropsychology
  – Serves families locally, regionally, and nationally

• Team members:
  – Amy Talboy, M.D.
  – Sharron Close, PhD, MS, CPNP-PC, FAAN
  – Leonidas Panagiotakopoulos, MD
  – Akanksha Mehta, MD, MS
What is Neuropsychology?
What is neuropsychology?

- Specialty area of psychology
- Cognition and behavior in relation to brain functioning
- What are the psychological implications of neurological conditions?
  - Effects on thinking, learning, behavior, and emotions
- Clinical neuropsychology
  - Use psychological tests to assess cognitive functioning, make connections with underlying brain structure and functioning
What is a Clinical Neuropsychologist?

• Doctoral training in clinical psychology
  – PhD not MD
• Specialized coursework (e.g., Neuroanatomy)
• Clinical training with patients with medical conditions
  – Work in medical settings
  – Collaborate with MDs, nurses, therapists
• Licensed as a psychologist/specialty in Neuropsychology
• Assess patients throughout the lifespan
  – Adult
  – Geriatric
  – Pediatric
Pediatric Neuropsychology

• Infants, children, and adolescents
• Knowledge of development
  – How the brain and nervous system develop
  – Children’s cognitive and emotional development
  – Typical and atypical patterns
• Additional considerations:
  – Role of the family and environment
  – Child’s development to this point
  – Educational implications
    • How neurological conditions will affect learning
    • What are the academic expectations for the child
Pediatric neuropsychologist role:
What is a Neuropsychological Evaluation?
Goals of Neuropsychological Evaluation

• Use psychological tests to assess cognitive functions
  – Strengths and weaknesses
  – Establish baseline level of functioning
  – Assess changes over time
• Interpret data in relation to neurological/medical condition
• Make diagnoses of psychological conditions
• Make recommendations to help family and child

• Disability qualifications
• Transition to adulthood
• Guardianship determinations
Similar/Related Terms

• Psychological Evaluation
• Psycho-educational Evaluation
• School Psychology Evaluation
• Educational Assessment
• Developmental Evaluation

Comparison:

• May utilize same tests, make same diagnoses
• Different types of training
• Goal may differ:
  – Neuropsychological: Understand/interpret findings in relation to brain functioning and/or brain development.
Pediatric Neuropsychological Assessment: Multiple sources of data

• Interview with patient and family
• Teacher reports
• Patient’s history
  – Birth/Developmental
  – Medical
  – Educational
  – Social/emotional
• Behavioral observations
• Standardized rating scales
• Objective test data:
  – Information gathered in a structured way
  – Compares individual with typical expectation for age
Normal Curve:

“Normal” IQ scores range from 85 to 115—the average score of 100, plus or minus 15 points.
Areas often assessed:

- Intellectual functioning
- Visual
- Language
- Sensory/Motor
- Memory
- Attention
- “Executive functioning” skills:
  - Problem solving
  - Behavioral regulation
- Academic achievement
- Adaptive Functioning
- Behavioral, social, and emotional functioning
Assessment process

• What specific tests are used based on:
  – Individual concerns of the patient/family
  – Age of child
  – Language comprehension or language output
  – Motor abilities
  – Sensory deficits
    • Vision
    • Hearing
• Tests may need to be adapted if there are sensory or motor limitations
Interpretation of results

• A lot of data is collected as part of the assessment...
• Identify patterns of strengths and weaknesses from data
• Integrate test data with:
  – Background information
  – Observations
  – Previous testing
• Provide interpretation, diagnoses
• Identify risks for patient
  – Predict likely outcomes
  – Guide family expectations
• Make recommendations based on risks
How can results from the Neuropsychological Evaluation inform parents?
Recommendations

- Medical Team
- School Team
- Family
- Therapists/Other Professionals working with child
- Resources
Medical Team Recommendations

• Information about child’s level of cognitive functioning
• Child’s ability to understand information about condition
• Adaptive functioning
  – Independence in Daily Living skills
  – Adaptive skills assist transition to adulthood
• How to address behavioral challenges
• Emotional functioning
  – Depression/Anxiety
• Is there a need for additional interventions/therapies
  – Pharmacological intervention
  – Psychological Therapy
Academic Recommendations

• Establish Special Education services
  – Individual Educational Program (IEP)
    • Eligibility designation – Other Health Impaired, LD
    – Behavior Intervention Plan
  • Accommodations: Section 504 Plan
  • Modified Instructional setting
    – School
    – Classroom within school
      • Self contained
      • Co-taught/inclusion
      • Pull-out/resources services
Academic Recommendations

• Examples of educational accommodations
  – Testing – Extra time, testing in small setting
  – Breaks – Fatigue, assist attention
  – Keyboarding/computer use instead of writing
  – Use of calculator
  – Meetings with school counselor
Academic Recommendations

• Educational Strategies common in X and Y variations:
  – Learning Disabilities
    • Reading (Dyslexia)
    • Math (Dyscalculia)
    • Writing (Dysgraphia)
      – Remediation strategies
      – Resources
  – Attention Disorders (ADHD)
    • Environment supports in classroom
      – Location of seat, distractions
    • Need for breaks, varying routine
    • Gaining child’s attention in class
Academic Recommendations

• Executive Functioning skills:
  – Impulsivity, emotional regulation
  – Flexibility/rigidity
  – Working memory
  – Planning/organization
  – Techniques to improve planning, organization
  – Study skills

• Language Development
  – Modifications to how information is presented
  – Eliciting verbal output in class
  – Speech-therapy
Academic Recommendations

• Motor Skills
  – Poor handwriting
  – Impacts note-taking, longer written output
  – Test-taking accommodations

• Behavioral management
  – Classroom strategies
  – Positive reward system
  – Functional Behavior assessment
Recommendations for Family

• Behavioral strategies at home
  – Positive reward system
• Providing emotional support
  – Therapist
  – Psychiatrist
• Organization at home
  – Study skills
  – Tutoring
• Promoting adaptive skills
  – Expectations for daily living skills
• Sleep hygiene
Empower family

- Provide education about child’s skills, medical condition
- Explanation of findings
- Give family information and resources:
  - To work with their school
  - To work with medical team
- Resources:
  - Parent-to-Parent of Georgia (p2pgai.org)
  - Wrightslaw.com
    * Special education law and advocacy
  - www.ldonline.org
Specific implications for individuals with X and Y chromosome variations
What are X and Y Chromosome variations?

• In **1 in 400** births the chromosome combination differs from 46, XY (male) or 46,XX (female)
• Characteristic physical/medical and cognitive findings
• May be less severe and more variable than other chromosome abnormalities (e.g., Down syndrome)
• Still have widespread risks:
  – Developmental delays
    • Language
    • Motor difficulties
  – Learning and attention disorders
  – Emotional and adaptive functioning problems
  – Social development
# Prevalence of X and Y chromosome variations

<table>
<thead>
<tr>
<th>Karyotype</th>
<th>Also Known as</th>
<th>Est. Prevalence</th>
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<tbody>
<tr>
<td><strong>Males</strong></td>
<td></td>
<td></td>
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<tr>
<td>47, XXY</td>
<td>Klinefelter Syndrome</td>
<td>1:650</td>
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<tr>
<td>47, XYY</td>
<td></td>
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<tr>
<td>48, XXXY</td>
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<tr>
<td>48, XYYY</td>
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<td>?</td>
</tr>
<tr>
<td>49, XXXXY</td>
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<td>1:85,000</td>
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<tr>
<td>49, XXXYY</td>
<td></td>
<td>?</td>
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<tr>
<td>49, XYYYY</td>
<td></td>
<td>?</td>
</tr>
<tr>
<td><strong>Females</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>45, X0</td>
<td>Turner Syndrome</td>
<td>1:2,500</td>
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<tr>
<td>47, XXX</td>
<td>Triple X, Trisomy X</td>
<td>1:1,000</td>
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<tr>
<td>48, XXXXX</td>
<td>Tetrasomy X</td>
<td>1:50,000</td>
</tr>
<tr>
<td>49, XXXXXX</td>
<td>Pentasomy X</td>
<td>1:250,000</td>
</tr>
</tbody>
</table>
47, XXY (Klinefelter Syndrome)

• Physical features
  – Can be subtle early in life
  – Language/motor delays
  – Hypotonia, hypertelorism, clinodactyly, hyperextensible joints, flat feet
  – Tall stature emerges most during adolescence
• Micro-orchidism, gynecomastia occurs later in adolescence or early adulthood
• Testosterone replacement common
• Osteoporosis, intention tremor, thyroid problems, autoimmune diseases, Type II diabetes
47, XXY (Klinefelter Syndrome)

- IQ often in the average range
- Nonverbal and visual skills often a relative strength
- Often have language and verbal deficits
  - Delays in speech development
  - Abstract verbal reasoning
  - Language-based learning disability in 50-80%
- Can have attention and “executive” difficulties
  - ADHD
  - Low frustration tolerance
- Motor skill weaknesses (Joint laxity and hypotonia)
47, XXY (Klinefelter Syndrome)

- Behavioral and Emotional features
  - Elevated anxiety and depressive symptoms
    - May increase in adolescence
    - Social cognition, immaturity
  - Poor self-expression, shyness, and frustration may also be related to language deficits
  - Increased rates of autism or psychosis when patients are sampled from clinical settings or support groups
47, XXY Recommendations

- Often in mainstream school setting
- Learning Disability remediation
- Language difficulties
  - Speech therapy
- Supports for attention, executive functioning
- Motor skill weakness
  - May fatigue easier in school – adjust schedule
  - Help for writing, notetaking
- Emotional support – psychologist
  - Anxiety may increase in adolescence
- Social skills training
- Psychopharmacology for ADHD, Anxiety, Depressed mood
47 XYY (Jacob’s Syndrome)

• Only 15-20% of males with XYY are diagnosed in lifetime
• Tall stature
• Developmental delays, hypotonia
• Motor tics, tremors, possible seizures
• Often have normal fertility
47, XYY

- Generally average IQ
- Speech delays common
- Stronger nonverbal, visual-motor skills
- 50-70% can have learning disabilities
- Attention problems in up to 80%
  - Often more hyperactive, impulsive than for KS
- Increased social skill deficits and ASD
- Increased risk for anxiety, depression, and other psychiatric disorders
- Many similar recommendations as with 47, XXY
48, XXYY / 48, XXXY / 49,XXXXY syndromes

• Rare, more cognitive and behavioral impairments
• Similar physically as in KS but can have more severe medical problems
  – Seizures, cardiac, genitourinary, musculoskeletal abnormalities
• Usually much lower verbal than nonverbal scores
• Emotional, adaptive, behavioral problems are common
• Recommendations:
  – Often will require self-contained classroom
  – Functional academic skills
  – Adaptive functioning skills important
  – Therapeutic interventions
47, XXX / Trisomy X / Triple X syndrome

- 1959, 47,XXX Karyotype was identified
- Only 10% are diagnosed in lifetime
- Physical and medical features
  - Tall stature, flat feet, joint hyperexensibility
  - Kidney and genitourinary problems
  - Risk of premature ovarian failure
  - Hypotonia, tremor, motor skill deficits, seizures
Neuropsychological findings in Trisomy X

- Mean IQ score 15-20 points lower than controls
- Stronger Nonverbal than Verbal skills
- Expressive language delays
- Language based learning disabilities
  - Math LDs can occur as well
- Delayed fine and gross motor skills
- Attention problems
- Shyness, anxiety, mood problems, can be worse in adolescence
Trisomy X Interventions

• Increased height, poor physical endurance
  – School accommodations
• Language interventions
• Learning disability remediations
  – Reading Disabilities
  – ADHD
  – Executive functioning
• Promote adaptive skills
• Emotional assessment
• Psychopharmacology if needed
Tetrasomy X and Pentasomy X

• Less common

• Dysemorphic features: Hypertelorism, epicanthal folds, clinodactyly, plagiocephaly
  – Cardiac malformations, cleft palate, hip dysplasia
  – Hypotonia, Developmental delays, tremor, tics, seizures
  – Pubertal delays, ovarian failure

• Significant neuropsychological impairments
  – IQ range typically impaired
  – Verbal skills usually in similar range
  – ADHD, autism spectrum disorders higher
  – Behavioral and emotional problems
  – Dysregulated mood and behaviors
Tetrasomy X and Pentasomy X

• Recommendations
  – Special education services
    • Functional academic skills
    • Foundational skills
  – Language therapy
  – Motor skill supports
  – Focus on adaptive functioning, life skills
Acknowledgements

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