The Neuropsychology of Neurofibromatosis

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Neuropsychology is a clinical specialty concerned with the **cognitive**, **behavioral**, and **emotional** problems that may arise from known or suspected brain dysfunction.
What is a Neuropsychological Evaluation?

A neuropsychological evaluation is a comprehensive assessment of cognitive and behavioral functions using a set of standardized tests and procedures.
What is assessed?

The pediatric neuropsychologist may look at a broad range of abilities, evaluating skills not usually tested by the clinical or school psychologist.

- General intellect
- Achievement skills, such as reading, writing and math
- Executive skills, such as organization, planning, inhibition, and flexibility
- Attention (simple, complex, and sustained)
- Learning and memory
- Language
- Visual-spatial skills
- Motor coordination
- Behavioral and emotional functioning
- Social skills
What have we learned?

NF can impact a wide range of cognitive, academic, and emotional functions including:

• Academic Achievement (Learning Disabilities)
• Attention (Attention Deficit Hyperactivity Disorder, ADHD)
• Executive Functioning (Flexibility, Cognitive Control)
• Learning and memory
• Visual-spatial skills
• Motor coordination
• Behavioral and emotional functioning
• Social skills

But...There is NO clear cognitive pattern.
Academic difficulties present in 50%-60% of NF1 patients

Recent Danish study (2008) found 75% of NF1 patients performed at least 1 standard deviation below grade level peers in at least one academic domain:

- Reading (technical & comprehension)
- Spelling
- Mathematics

- While only 10% did not have problems in any aspect of school functioning, these children still displayed impairment in nonverbal long term memory, executive functions, and attention.

Dual Route model of reading:

- The lexical reading procedure involves visual whole-word recognition

- The sublexical reading procedure involves rule-based grapheme-phoneme correspondences, allowing the skilled reader to sound out unfamiliar words and nonwords (or pseudowords)

Study of 30 NF1 children (Ages 7-12) found:

- 67% demonstrated deficits in one or more reading sub-skills
- 75% met criteria for phonological (sublexical) dyslexia
- none met criteria for lexical dyslexia
- 20% classified as mixed dyslexia

Suggests a deficit at the level of the phonological module impairs the ability to segment spoken words into their underlying phonologic constituents and successfully link letters with their corresponding sound(s)

Importantly

- Only 17% of the same cohort (5/30) were identified as having a difficulty in reading when a traditional IQ/AA discrepancy was employed, even though 63.3% of participants were rated as below grade level in aspects of literacy by teacher report.

- Traditional discrepancy-based methods as employed by schools may not be sufficiently sensitive to identify children who experience reading difficulties.

What cognitive domains can impact math performance?

- Executive functioning/attention (failure to monitor output / changing task demands)
- Poor working memory (story problems)
- Visual spatial impairments (difficulty in learning to recognize shapes and hence, operation symbols and numerals)
- Simple motor coordination and visual motor abilities (affect writing and drawing skills)

Math Disabilities appear to result from several factors

The Neuropsychology of Neurofibromatosis (NF) Attention

Approximately 50% of NF1 patients satisfy criteria for ADHD

- Objective measures reflect impairments in sustained attention and response inhibition, as well as cognitive control

- ADHD like symptoms may contribute to academic and social problems

- Low doses of methylphenidate significantly improve performance on measures of persistence

- At one year follow up, it was demonstrated that stimulant medication can lead to improvement in cognitive, academic, and social problems of children with NF1 and ADHD
Impairment is one of the most commonly reported problems.

Visual-spatial deficits are so common that some clinical investigators have suggested that their presence might help in the diagnosis of marginal cases of NF-1. In one recent study, using discriminant analysis, 90% of individuals with NF-1 were correctly identified based on their performance on a combination of visual-spatial tests (Schrisher et al. 2003).
Social and emotional problems in children with NF1

They were perceived by peers and teachers as being more sensitive and isolated and as less likely to be leaders. They were also less liked and had fewer best friends and reciprocated friendships.
Neurofibromatosis (NF) Emotions

Mean Z-score

Social Problems
Attention Problems
Thought Problems
Anxious/Depressed
Withdrawn
Somatic Complaints
Aggressive Behavior
Delinquent Behavior
Total
Internalizing
Externalizing

NF1
Unaffected
21% of Swedish adult sample displayed Dysthymia in a 12 year follow up study

<table>
<thead>
<tr>
<th>CPRS item number and description</th>
<th>NF1 patients without psychiatric diagnosis (n = 25)</th>
<th>NF1 patients with psychiatric diagnosis (n = 12)</th>
<th>All NF1 patients (n = 37)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>mean</td>
<td>S.D.</td>
<td>mean</td>
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<tr>
<td>Reported variables</td>
<td></td>
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<tr>
<td>(1) Sadness</td>
<td>0.00</td>
<td>0.00</td>
<td>0.71**</td>
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<tr>
<td>(3) Inner tension</td>
<td>0.35</td>
<td>0.51</td>
<td>1.17***</td>
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<tr>
<td>(4) Hostile feelings</td>
<td>0.19</td>
<td>0.40</td>
<td>0.96**</td>
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<tr>
<td>(6) Pessimistic thoughts</td>
<td>0.08</td>
<td>0.27</td>
<td>0.63***</td>
</tr>
<tr>
<td>(7) Suicidal thoughts</td>
<td>0.02</td>
<td>1.00</td>
<td>0.63**</td>
</tr>
<tr>
<td>(8) Hypochondriasis</td>
<td>0.13</td>
<td>0.39</td>
<td>0.38*</td>
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<tr>
<td>(9) Worrying over trifles</td>
<td><strong>0.21</strong></td>
<td>0.40</td>
<td>0.67***</td>
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<tr>
<td>(11) Phobias</td>
<td><strong>0.40</strong></td>
<td>0.77</td>
<td>0.67**</td>
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<tr>
<td>(15) Fatiguability</td>
<td>0.08</td>
<td>0.39</td>
<td>0.58*</td>
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<tr>
<td>(16) Concentration difficulties</td>
<td>0.08</td>
<td>0.27</td>
<td>0.50*</td>
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<tr>
<td>(17) Failing memory</td>
<td>0.23</td>
<td>0.51</td>
<td>0.75*</td>
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<tr>
<td>(19) Reduced sleep</td>
<td><strong>0.40</strong></td>
<td>0.74</td>
<td><strong>1.00</strong>*</td>
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<tr>
<td>(23) Autonomic disturbances</td>
<td>0.35</td>
<td>0.56</td>
<td>0.83**</td>
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<tr>
<td>(24) Aches and pains</td>
<td>0.38</td>
<td>0.65</td>
<td><strong>1.08</strong>**</td>
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</tbody>
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Zoller & Rembeck (1999)
The Neuropsychology of Neurofibromatosis (NF)

Value of Neuropsychological Evaluations in NF:
- Identify an individual's unique neuropsychological profile
- Determine eligibility for special services
- Tailor remedial teaching strategies for the specific cognitive functions that are impaired
- Monitor appropriate dose/efficacy of stimulant medications
- Identify appropriate academic accommodations
- Direct individuals to appropriate services (i.e., counseling) when appropriate