Neurodevelopmental Disorder associated with 8p23.1 Microdeletion Syndrome: A Pediatric Case Study

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Objective

- 8p23.1 microdeletion syndrome is a rare genetic disorder involving a partial deletion of chromosome 8.
- Children with this microdeletion are at increased risk for prenatal growth deficiency, congenital heart defects, craniofacial abnormalities, seizures, intellectual deficits, language impairments, motor deficits, and behavioral disorders.

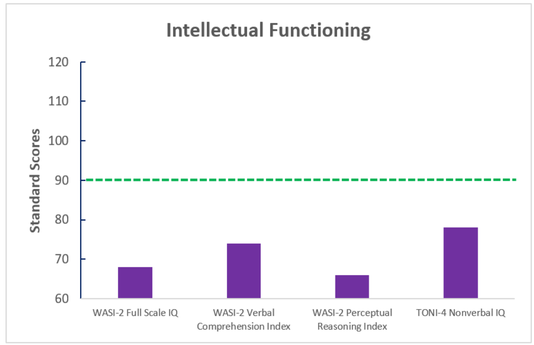
Method

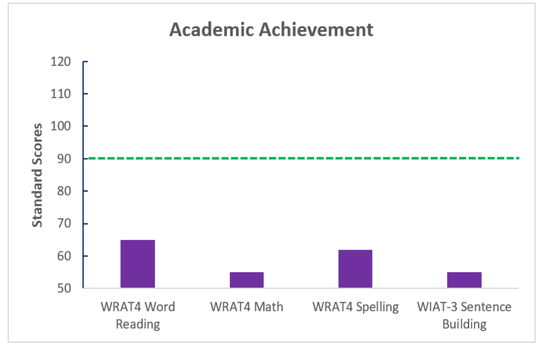
- This case study presents on the neuropsychological profile of a 9-year-old female with 8p23.1 microdeletion syndrome who had a history of absence seizures, heart valve disease, microcephaly, and developmental delays.
- Patient previously underwent psychological testing at age 8 and received diagnoses of ADHD and Intellectual Disability; however, her parents now had concerns for ASD.
- Current testing battery included: *WASI-2, TONI-4, WRAT4, WIAT-3, WRAML2, CMS, EVT-3, PPVT-5, NEPSY-2, Beery VMI, CPT-3, ADOS-2, SCQ, BRIEF-2, BASC-3, ADHD Rating Scale-5, and ABAS-3.*

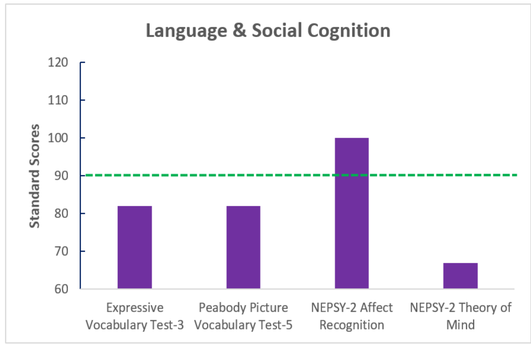
Results

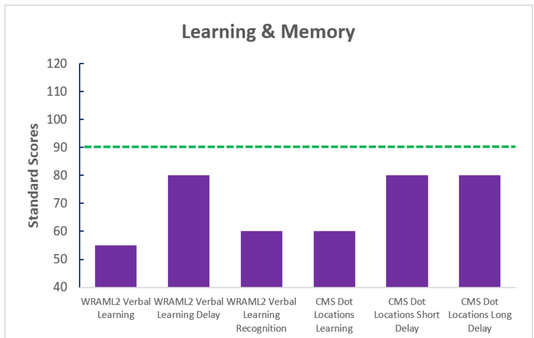
- Neuropsychological testing revealed impairments in intellectual functioning, adaptive functioning, language, academic achievement, attention, executive functioning, memory, visuomotor skills, and social cognition.
- Concerns for social-emotional functioning were endorsed on multiple measures (e.g., SCQ, ABAS-3, and BASC-3) and were consistent with patient's classification of "Autism (High)" on the ADOS-2.
- Patient was diagnosed with: **Neurodevelopmental Disorder associated with 8p23.1 Microdeletion Syndrome, ASD with Intellectual and Language Impairment, and ADHD - Combined Presentation.**
- Of note, discrepancies were observed between parent and teacher report on questionnaires. This was likely due in part to the teacher having limited observations of patient because of the school year recently starting.

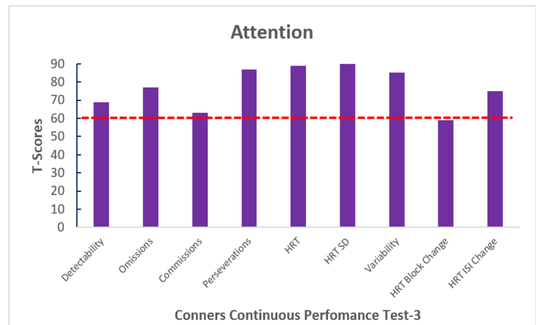
Select Measures from Neuropsychological Battery

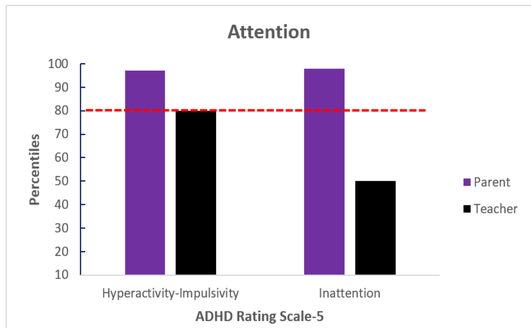


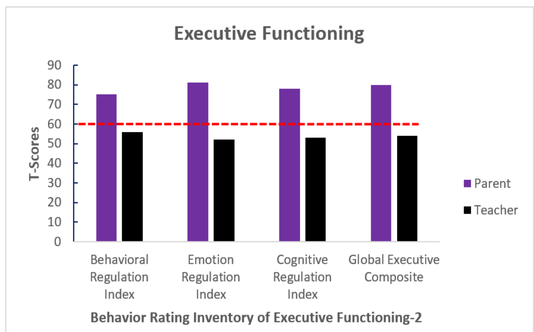


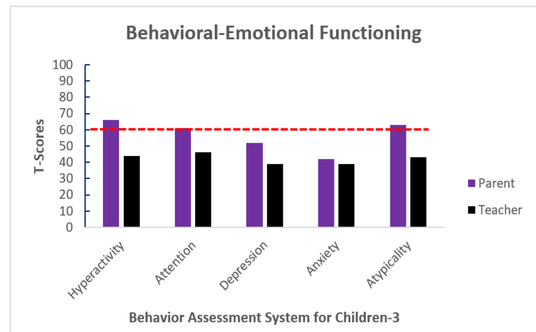


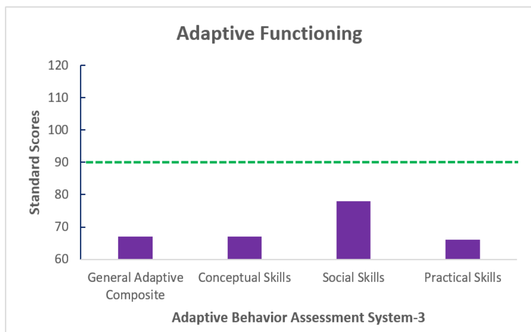












Conclusions

- Recommendations were made to modify the patient's special education academic goals, initiate various interventional services at school (e.g., speech therapy, occupational therapy, and social work), and start private ABA therapy.
- Continued monitoring and medication management with neurology and psychiatry were recommended as well given the patient's history of absence seizures and problems with attentional-behavioral regulation.
- This case demonstrates the importance of comprehensive neuropsychological evaluation and treatment of children with certain rare genetic disorders such as 8p23.1 microdeletion syndrome in order to help improve neurodevelopmental outcomes in these patients.

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