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Neurocognitive Functioning in a Young Female with Weaver Syndrome

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ABSTRACT

Weaver Syndrome is a low frequency syndrome caused by mutations in the EZH2 gene. Individuals with Weaver Syndrome have similar physical abnormalities as well as overall low cognitive functioning, motor deficits, and difficulties with independent living skills. This is a case study that presents the neurocognitive profile of n 11-year-old female diagnosed with Weaver Syndrome. This was the child's first neuropsychological evaluation following a long-standing history of concerns regarding her cognitive abilities, academic achievement, and independent living skills. Results of the evaluation revealed extremely low range cognitive functioning, as well as below age expectation performance across a majority of cognitive domains. While the child's neurocognitive profile was similar to prior research regarding cognitive outcomes in children with Weaver Syndrome, additional research on the neurocognitive profile of children with Weaver Syndrome is indicated.

Clinicians often assume ASD when a school-aged child presents with severe social difficulties. However, there are a host of alternative etiologies that should be entertained when social impairment is a primary referral concern. Methods: A 9-year-old girl was referred for a neuropsychological assessment given severe social impairment, immaturity, poor frustration tolerance, and significant attentional difficulties. She was described as desiring friends, but not knowing how to develop or maintain friendships. She was a term infant, although there was a strong suspicion of alcohol exposure during pregnancy. She was delayed with the acquisition of all milestones. Medical history was essentially unremarkable. She was attending 3rd grade and receiving support under Emotional Impairment. Results: She demonstrated Borderline intellectual abilities with a relative weakness in Working Memory. Abstract spatial processing was a weakness. Fine motor speed was impaired bilaterally. Multiple trial visual and verbal learning were impaired while recall for prose was a relative strength. Speech and language abilities were a relative strength, although articulation difficulties were evident. Deficits in reaction time, problem-solving, and mental flexibility were also apparent. Affect recognition was impaired. Caregiver report measures revealed a Social Communication Questionnaire score of 23, and peak T-score elevations on the Conner's Parent Rating Scale of 90 and 98 for Peer Relations and the ADHD Index, respectively. Conclusions: Although the referral suggested possible ASD and ADHD, suspected FASD was thought to be a more parsimonious diagnostic consideration. This case presentation will illuminate the differences in social and neuropsychological impairment in these three phenotypically similar disorders.

In the United States, the neuropsychological assessment of native-Greek speaking individuals commonly includes the use of translated

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instruments and interpretive services. This case presentation will consider the clinical implications of utilizing translated measures versus the clinical utility of culturally adapted instruments in the context of surgical language testing. Neuropsychological assessment is an essential component of the presurgical evaluations of craniotomy candidates. As such, the neuropsychological referral may include evaluating suitability for surgery, the localization and lateralization of pathology, determining surgical parameters and predicting surgical outcomes. Further, when surgical parameters threaten language functions, the neuropsychologist may be called to conduct intraoperative language testing. Due to the time limits of the surgical setting and increased risk of patient discomfort, brief and valid assessments are of critical importance. Yet, despite a growing body of literature illustrating the impacts of nonnative-speaker status and culture on neuropsychological instrument performance, intraoperative language assessments often rely on directly translated measures. Intraoperative language mapping was conducted to elucidate critical language areas in the vicinity of a left parietal neoplasm in a right-handed, native-Greek speaking, multilingual (Greek, English, Spanish) man. Baseline preoperative language assessments and intraoperative assessments were conducted in both English and Greek by a bilingual clinician. The salient issues explored will include a. the effective applicability of current language measures across cultural contexts, b. the psychometric properties and equivalency of translated assessments, and c. the use of the existing normative data.

***Keywords*:** Weaver syndrome; Neurocognitive function; Child; Genetics; Neuropsychology