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 an 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.

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