Results: Group means significantly diverged from age-expected performance by the second evaluation in all domains except semantic fluency. Weakness was identified on at least 1 verbal subtest for 79% of the sample at the first evaluation, and for 85% of the sample at the second evaluation. As a group, patients showed a significant increase in the number of weaknesses identified on performance-based measures from the first to second evaluation [t(47) = -3.60, p <.001]. Over half of the sample showed an increase in the rate of verbal weaknesses identified (56.3%). Those with more weaknesses over time had lower IQ at the initial evaluation [t(36) =-2.61, p=.013]. An increase in the number of weaknesses from first to second evaluation was not associated with tumor type/location, treatment modality, or demographic variables.
Conclusions: Brain tumor diagnosis in early childhood during rapid language development is associated with language impairments soon after diagnosis, and years after treatment completion. Causes for continued and increased impairment are multifactorial and risk cannot clearly be identified by demographic and treatment variables alone. Any early language weakness identification should signal need for intervention as the causes for difficulty are complex and these weaknesses are likely to persist and increase over time.

Categories: Cancer
Keyword 1: pediatric neuropsychology
Keyword 2: brain tumor
Keyword 3: language
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49 Case Study: Cognitive Deficits Associated with Norrie Disease

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Objective: Norrie disease is a rare, x-linked recessive genetic disorder associated with an NDP gene mutation. Males are predominantly affected. Typical symptoms include vision loss around the time of birth and progressive hearing loss. Cognitive and behavioral abnormalities also occur in 30-50% of individuals, including developmental delays, intellectual disability, cognitive regression, psychosis, and aggression. There is limited research, however, examining the neuropsychological deficits in adulthood resulting from Norrie disease, especially with neuropsychological data and in individuals without other neurological manifestations of the disease, such as seizures. Here, we present the neurocognitive profile of a patient with Norrie disease who presented for a cognitive evaluation in adulthood due to report of more recent memory decline.
Participants and Methods: Mr. Smith is a Caucasian male in his mid-40's who previously underwent genetic testing and was subsequently diagnosed with Norrie disease. As a result of his diagnosis, he experienced complete vision loss since birth and bilateral hearing loss that began in childhood and gradually worsened in adolescence. Medical history was otherwise unremarkable. Developmental milestones were met on time. Historical intelligence testing conducted in elementary school revealed borderline on one intelligence test to high average performance on other intelligence tests. However, he was retained grades several times due to factors such as behavioral disruptions and academic difficulties. He had been employed as an assembly line worker for many years, but had not worked for 10 years prior to the neuropsychological evaluation. Emotionally, he had a longstanding history of anxiety and endorsed mild anxiety and depression at the time of the evaluation. The patient first noticed memory difficulties in adolescence then noticed further decline four years prior to the neuropsychological evaluation (around when he received a left-ear cochlear implant), which had remained stable since onset.
Results: In the context of low average premorbid intellectual functioning, Mr. Smith's neurocognitive profile was notable for difficulties with alphanumeric set-shifting and abstract thinking, with otherwise preserved cognitive functioning. Weaknesses observed on testing may have represented longstanding weaknesses and did not rise to the level of a cognitive disorder. Affective distress was also suspected to have accounted for some of the cognitive lapses the patient reported experiencing with day-to-day functioning.
Conclusions: The current poster aims to contribute to the limited body of literature

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examining neuropsychological deficits in adulthood resulting from Norrie disease. This is especially critical given that the long-term cognitive dysfunction of this disorder is relatively unknown and could negatively impact patients' quality of life over time.

Categories: Genetics/Genetic Disorders
Keyword 1: genetic disorders
Keyword 2: neuropsychological assessment
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50 The Neuropsychological Profile of *SIN*3A-Related Disorder/Witteveen-Kolk Syndrome: A Case Study

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Objective: The *SIN*3A gene mutation is a rare genetic mutation with few reported cases (< 1 in 1,000,000) associated with autosomal dominant Witteveen-Kolk Syndrome, a rare neurodevelopmental syndrome only discussed in the past decade (Witteveen et al., 2016). This syndrome can be characterized by short stature, distinctive facial features, developmental delay, mild intellectual disability, autism spectrum disorder, hypotonia, and seizures (Balasubramanian et al., 2021), however a paucity of information regarding comprehensive neuropsychological functioning in these individuals is present in the literature, and even this recent review study noted that intellectual ability was rarely assessed through formal testing (6 of 28 cases). We present a case, "M", to help describe a potential pattern of neurocognitive strengths and weaknesses in this population.
Participants and Methods: The participant, "M", was diagnosed with a de novo mutation in the *SIN*3A gene at the age of 11 years with previous diagnoses of global developmental delay, hypotonia, autism spectrum disorder, specific learning disability in mathematics, developmental coordination disorder, and attention deficit hyperactivity disorder. M was seen for a comprehensive neuropsychological evaluation at 11 years of age at an academic medical center, which consisted of comprehensive review of medical and school records, parent and child interview, questionnaires, and performance-based testing.
Results: M's verbal and language skills emerged as a particular strength. Her verbal memory, verbal fluency, and verbal comprehension skills were all in the average range or above, as were reading, reading comprehension, and spelling skills. M demonstrated a pattern of notable weaknesses in visuospatial skills, including impaired visuospatial reasoning, visuomotor integration, visual scanning, visual perception, and visual memory. Additionally, M demonstrated a slight weakness in Low Average mathematics skills. M also demonstrated fine motor impairment with impaired speed, coordination, and accuracy. Although immediate auditory attention was noted to be average, performance on a test of sustained attention indicated a moderate persistence of attention concerns. Likewise, M's mother reported her to be very elevated on symptoms of both attention and hyperactivity/impulsivity. Finally, M's mother reported elevated concerns related to M's peer relations and atypical behaviors and below average adaptive skills.
Conclusions: Due to the rarity of M's de novo mutation in the *SIN*3A gene, M's pattern of weaknesses in visuospatial skills, fine motor skills, attention/executive functioning, and social skills, as well as her strengths in verbal skills can aid in further understanding the pattern of cognitive strengths and weaknesses in children with a mutation in the *SIN*3A gene. Additionally, given her mild weaknesses in math skills, it is possible that M's performance on mathematics assessments may be impacted by her visuospatial weakness and thus better conceptualized as a visuospatial issue rather than a learning disability. Overall, this case can aid in identifying specific cognitive risk factors, such as visuospatial skills, in this population and lead to more targeted assessment and intervention, and highlights the importance of more nuanced cognitive evaluation as reporting of a general cognitive ability score alone may obscure underlying patterns of cognitive strength and weakness.

Categories: Genetics/Genetic Disorders
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