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
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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.

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51 Cognitive Functioning in Neurofibromatosis Type 1: The Role of Community Socioeconomic Status

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Objective: Neurofibromatosis type 1 (NF1) is a neurogenetic disorder associated with increased risk of neuropsychological challenges. While research has evidenced associations between environmental factors and neurocognitive development, few studies have examined the role that socioeconomic status (SES) plays on neuropsychological development in NF1. The aim of this study is to examine the relationship between community SES and cognitive/psychosocial outcomes in a neuropsychology clinic sample of pediatric NF1 patients.
Participants and Methods: The sample consisted of 47 youth with NF1 (M age=11.91, SD=3.69). The sample was 51.1% female, 72.3% White, 19.1% Black/African American, and 4.3% Hispanic. All participants had completed neuropsychological assessments for clinical purposes at an outpatient clinic in an urban, midwestern medical center.
Data from neuropsychological measures and demographic information were pulled from records and entered into a de-identified dataset. The Wechsler Intelligence Scales, California Verbal Learning Test (CVLT), Woodcock Johnson Test of Achievement, and parent- and teacher-report versions of the Behavior Assessment System for Children (BASC) and the Behavior Rating Inventory of Executive Function (BRIEF) were used to examine broad neuropsychological functioning.
The Area Deprivation Index (ADI) measures SES at the community level, as opposed to the individual level. It is composed of 17 factors related to education, poverty, employment, and housing. This information is used to assign index scores by zip code, with scores on a scale of 1-10 and 10 indicating the highest level of socioeconomic disadvantage. Mean ADI for this sample was 4.02 (SD=1.93).
Results: Mean neurocognitive scores were consistently in the low average to average range. Parent and teacher scores on the BASC were in the average range. Mean scores on the BRIEF indicated Global Executive Composite scores in the mildly and moderately elevated range for parents and teachers, respectively. Correlational analyses revealed significant associations between ADI scores and immediate recall performance on the CVLT (Trials 1-5; r=.37; p=.03) and the BRIEF Planning and Organization subscale (r=.35; p=.02). Both remained significant after controlling for FSIQ (CVLT: rFSIQ=.49, p=.003; BRIEF: rFSIQ=.38; p=.02).
Conclusions: Mean cognitive scores for the sample are consistent with existing literature demonstrating that individuals with NF1 are at risk of reduced functioning in several domains. Sample mean ADI of 4 indicates a relatively low level of disadvantage in this sample., ADI was significantly associated with two variables, and greater deprivation was associated with better list learning performance. This suggests that the role of community SES is likely nuanced in how it impacts neurocognitive development. Results provide mild evidence of an association between ADI and learning and planning/organization. However, limitations to the current study, including a small sample size, and the retrospective nature, likely limits a more detailed understanding of the true relationship between community resources and cognitive and psychosocial outcomes among children with NF1. Future research comparing larger low and high ADI samples is necessary to fully examine the relationship between these factors. With better understanding of how community SES may limit or support neurocognitive and psychological growth in this population, more effective interventions can be designed for this group whose members are at notable risk for cognitive and psychological challenges.

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