Objective: 22q11.2 Deletion Syndrome (22q11DS) is a multi-systemic disorder with great clinical heterogeneity. It is the most common microdeletion syndrome and one of the most common genetic causes of developmental delays (e.g., motor/speech). 22q11DS is estimated to occur between 1/2,000-4,000 live births. However, the diverse clinical presentation of 22q11DS and health inequities that impact ethnically, racially, linguistically, and economically marginalized groups, make early identification, diagnosis, and access to beneficial early interventions (e.g., speech/behavioral therapy) even more challenging. Therefore, 22q11DS' true prevalence may be larger than documented. Challenges associated with diagnosis, as well as neurocognitive, psychiatric, and medical co-morbidities associated with 22q11DS have been reported to affect the quality of life and well-being of people living with 22q11DS and their families. Yet, there is limited longitudinal data on lifelong functional outcomes of this population and the social factors that may shape them. This study aimed to 1) review the extant literature on adaptive functioning across the lifespan in 22q11DS and 2) report on relevant social and structural variables considered in the literature to contextualize adaptive functioning.
Participants and Methods: A scoping review was conducted between January-June 2022 across six electronic databases: PubMed, Scopus, PsycINFO, Ovid MEDLINE, EBSCO, and Embase. The 'building block' method was used to identify and design a comprehensive search strategy used to scan publications' titles, keywords, and abstracts. Citation mining strategy was utilized to identify additional relevant studies. The following inclusion criteria was met: 1) empirical studies conducted in humans, 2) participants with confirmed diagnosis of 22q11DS, 3) evaluation of adaptive functioning, 4) use of at least one standardized measure of adaptive functioning and 5) written or translated into English or Spanish.
Results: Eighty-four records were initially identified. After deduplication, abstract screening, and full record reviews, a total of twenty-two studies met inclusion criteria for this review. Only eight publications explored adaptive skills as one of their primary outcomes. Clinically significant symptoms of anxiety, withdrawal, anhedonia, and flat affect were associated with worse functional outcomes. Fifteen studies reported between one and three demographic variables (e.g., race/ethnicity, years of education), and only two studies documented mental health treatment status/history. Most studies reported lower adaptive abilities in participants with 22q11DS independent from their cognitive abilities, but the majority of participants scored between the below average range and exceptionally low range on measures of intellectual functioning. Nonetheless, information on contextual variables (e.g., educational/occupational opportunities) that may help to interpret these findings was lacking.
Conclusions: Methodological differences (e.g., definition and measurement of adaptive functioning), recruitment bias (small, clinic-based identified samples) and lack of information regarding contextual level factors, may be limiting our understanding of the neurocognitive and neuropsychiatric trajectories of people with 22q11DS. It is vital to increase representative samples in epidemiological/clinical studies, as well as research examining the social and structural factors (e.g., access to healthcare, socioeconomic position) that impact functional outcomes in this population to promote public health policies that can improve brain health across the lifespan.

Categories: Autism Spectrum Disorders/Developmental Disorders/Intellectual Disability
Keyword 1: multiculturalism
Keyword 2: diversity
Keyword 3: adaptive functioning
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23 Latent Profiles of Children Referred for Possible Autism

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Objective: Autism is a neurodevelopmental disorder characterized by impairments in social communication and the presence of restricted and repetitive behaviors (RRBs). Clinical diagnosis of autism is often complicated by heterogeneity in core autism traits and other individual characteristics (e.g., cognition).

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Previous literature suggests that degree of autism characteristics, cognitive ability, and age contribute to identifying homogenous subgroups of autism, which facilitates prognosis and treatment planning. The present study extends these findings by examining profiles of cognition, age, and autism characteristics (measured by the *Autism Diagnostic Observation Schedule, Second Edition* [ADOS-2]) in a clinical sample of school-aged children presenting with concern for possible autism. Profiles are also described according to whether children received an autism diagnosis and clinician ratings of emotional/behavioral problems, which have been shown to influence diagnostic clarity when assessing for autism.
Participants and Methods: We conducted a retrospective chart review of 188 children (68% male) ages 4-17 years (M=8.9) who were referred for an autism evaluation. Latent profile analysis was conducted using age, ADOS-2 Social Affect (SA) and RRB scores, and verbal and non-verbal intelligence quotients (VIQ/NVIQ). Model fit comparing 2, 3, 4, and 5-class models was assessed using log-likelihood, AIC, BIC, SABIC, entropy, and Lo, Mendell, and Rubin (LMR) and bootstrap likelihood ratio (BLRT) tests. The frequency of clinical autism diagnosis and ADOS-2 emotional/behavioral problems were calculated across profiles in the best-fitting model.
Results: The 5-class model demonstrated the best fit. The following characteristics were observed across five profiles: 1) mean age = 9.5 years, Low Average VIQ/NVIQ, and low SA (M=5.2) and RRB (M=0.7) scores; 2) mean age = 7.3 years, Average VIQ/NVIQ, and low SA (M=3.1) and RRB (M=0.8) scores; 3) mean age = 10.1 years, Low Average VIQ/NVIQ, and high SA (M=11.3) and RRB (M=4.2) scores; 4) mean age = 8.8 years, Average VIQ/NVIQ, and moderately high SA (M=9.6) and RRB (M=3.4) scores; and 5) Exceptionally High VIQ, Above Average NVIQ, and comparatively mid-level SA (M=6.6) and RRB (M=3.6) scores. Autism diagnosis and emotional/behavioral problems varied across profiles. Profiles 1 and 2 contained lower diagnosis rates (33% and 10%, respectively). Profiles 3 and 4 contained the highest diagnosis rates (97% in both), followed by profile 5 (75%). In terms of emotional/behavioral problems, Profile 2 exhibited the highest overactivity (56%). Profile 3 demonstrated the highest rate of tantrums/disruptive behaviors (20%).
Conclusions: Findings revealed distinct profiles of IQ and autism characteristics within a clinical sample of school-aged children referred for possible autism. Children with the highest scoring ADOS profile were older compared to other profiles. Higher and lower scoring ADOS profiles exhibited both lower and higher IQ scores. Descriptive analyses suggested that the frequency of autism diagnosis was notably higher in moderate and high-scoring ADOS profiles; however, emotional/behavioral problems were salient in only one low and one high-scoring ADOS profile. The findings suggest that higher-scoring ADOS profiles consistently demonstrated high autism diagnosis rates but varied across IQ and behavioral problems. These results have implications for interpreting these characteristics during clinical autism diagnosis.

Categories: Autism Spectrum Disorders/Developmental Disorders/Intellectual Disability
Keyword 1: autism spectrum disorder
Keyword 2: cognitive functioning
Keyword 3: assessment
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24 Individual Differences in CAMCOG-DS Performance in Children and Adults with Down's Syndrome and Relationship to Language and Reasoning

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Objective: The Cambridge Cognitive Examination for Down's Syndrome (CAMCOG-DS) was developed to assess cognitive functioning and dementia-related cognitive decline in people with Down's Syndrome (DS). It has been translated into different languages and is often used in international studies. Although adapted for people with intellectual disabilities

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