
Background: Okur-Chung neurodevelopmental syndrome (OCNDS) is a rare genetic syndrome resulting from pathogenic variants in the CSNK2A1 gene. OCNDS is associated with language and motor delays, sleep challenges, intellectual disability, stereotypic movements, and autism spectrum disorder (Chung, 2022; Rushing & Chung, 2023). Sensory differences are widely reported in other neurodevelopmental genetic syndromes (e.g., Siper et al., 2017; Siper et al., 2021) and have yet to be explored in OCNDS. Characterizing sensory features may inform clinical care and improve understanding of syndrome-specific sensory phenotypes.

Objectives: To examine sensory reactivity in a cohort of individuals with OCNDS and to compare findings to a typically developing control group (TD) and an autism group (ASD).

Methods: Participants included 20 individuals with OCNDS (age 2-24 years, (Mage=12.4, SDage=7.3), and two comparison groups: 67 TD (age 2-12) and 174 ASD (age 2-12). Sensory reactivity was measured using the Sensory Assessment for Neurodevelopmental Disorders (SAND) (Siper & Tavassoli, 2021), which is a standardized clinician-administered observation and corresponding caregiver interview. The percentage of OCNDS participants who fell above clinical cutoffs on the SAND was calculated. A Wilcoxon Rank Sum was used to examine group differences. RStudio was used for analysis.

Results: OCNDS SAND Total Score showed no significant correlation with age and therefore the full sample was examined ($p=0.842$). Clinically significant Total SAND scores were present in 85% (17/20) of OCNDS participants, 45% in the Hyperreactivity Domain, 25% in the Hyporeactivity Domain, and 80% in the Seeking Domain. Within modalities, clinically significant scores were present in 55% of OCNDS participants in the Visual domain, 75% in the Tactile domain, and 45% in the Auditory domain. The OCNDS group showed significantly higher scores than the TD group for Total Score and across all domains (Hyperreactivity, Hyporeactivity, and Seeking, p 's <0.01). Participants with OCNDS showed significantly fewer overall sensory reactivity differences compared to the ASD group for Total Score, Hyporeactivity, and Seeking domains ($p=0.02$, $p=0.02$, $p=0.03$ respectively). Hyperreactivity scores were not significantly different between the OCNDS and ASD groups ($p=0.78$).

Conclusions: Results from the SAND suggest a unique sensory phenotype in individuals with OCNDS characterized by sensory differences in overall sensory reactivity and high levels of sensory seeking behavior in the majority of participants. While sensory differences were identified across all domains relative to controls, hyperreactivity was present in approximately half of participants and fell at similar levels to those with ASD, while hyporeactivity and seeking were less prevalent than in the ASD group. While OCNDS is a neurogenetic developmental syndrome, the presence of ASD in this OCNDS sample was not captured, which limits the interpretability of the comparisons. Importantly, this is the first known study to examine sensory reactivity in OCNDS and the utility of the SAND is evident as it was successfully collected on all participants. Larger samples are needed to further elucidate a syndrome-specific sensory phenotype that can be applied to OCNDS clinical and research interventions.

427.078 Sensory-Motor Features of Autistic Individuals with Intellectual Disability and Their Relation with Autism Severity, Adaptive Behavior and Repetitive Behavior

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Background: Despite several studies highlight the presence of sensory and motor impairments in a high proportion of individuals with autism, studies aimed at evaluating both sensory and motor features are scarce in the literature. Moreover, children and adolescents with ASD and intellectual disability, who represent a significant proportion of the ASD population, are underrepresented in autism research.

Objectives: to evaluate the sensory-motor features of autistic individuals with intellectual disability, and to verify their relationship with autism severity (AS), adaptive behaviour (AB), and restricted and repetitive behaviour (RRB).

Methods: Thirty-eight ASD subjects (age range: 5-14.06 years; 25 males) diagnosed according to DSM-5 criteria were recruited. Clinical characteristics such as non-verbal IQ, AS, AB and RRB were collected using the Raven Coloured Progressive Matrices (RCPM), Childhood Autism Rating Scale – Second Edition (CARS-2), Vineland-II Adaptive Behaviour Scale, and Repetitive Behaviour Scale Revised (RBS-R), respectively. The sensory-motor profile was investigated using the Sensory Profile - Second Edition (SP-2) and the Developmental Coordination Disorder Questionnaire (DCD-Q). The Spearman Correlation Test was used to analyse relationships between different outcome measures. A p-value <0.05 was considered significant.

Results: The CARS-2, RCPM, Vineland-II total, and RBS-R total scores were, on average, 40.32+/-6.99, 70.79+/-19.37, 27.92+/-11.83 and 25.92+/-12.53, respectively. The DCD-Q total mean score was 38.39+/-11.36; 30 of 38 individuals were identified as “suspected of DCD”.

Table 1 summarises the SP-2 sample results. The “Sensory quadrant” mean scores were in the “More than others” range in 3/4 quadrants (Seeking, Sensitivity and Registration). The “Sensory section” mean scores were in the “More than others” range in 2/6 sections (Touch and Oral). The “Behavioral section” mean scores were in the “More than others” range in 2/3 sections (Social/Emotional and Attentional).

The significant correlations between study variables are reported in Table 2. Correlation between SP-2 categories (“Much Less”, “Less”, “Typical”, “More”, “Much more”) and DCD-Q scores show a negative correlations between DCD-Q_”Total” score and SP-2_”Social/Emotional” categories, DCD-Q_”Control of Movement” subscore and both SP-2_”Social/Emotional” and SP-2_”Avoiding” categories, and between DCD-Q_”General _Coordination” subscore and SP-2_”Registration” categories. CARS-2 score negatively correlated with the DCD-Q_ Total and subtotal scores and with the SP-2_”Attentional” categories.

A partial correlation analysis, corrected for age, showed a correlation between Vineland-II_”Motor_Skills” domain and the DCD-Q_ Total and subtotal scores. Moreover, the Vineland-II_”Motor_Skills” domain negatively correlates with the SP-2_”Registration” categories. Moreover, Vineland-II_”Communication” domain correlated with the DCD-Q_ Total and subtotal scores, and negatively correlated with SP-2_”Conduct” categories.

Regarding RRB, RBS-R_”Stereotyped_Behavior” subscale score negatively correlated with DCD-Q total and subtotal scores and correlated with SP-2_”Conduct” categories. Moreover, RBS-R_”Self-injurious_Behavior” domain negatively correlates with DCD-Q total and “General coordination” scores and correlates with SP-2_”Registration” and SP2_”Tactile” categories. Finally, RBS-R_”Ritualistic_Behavior” subscale score correlates with DCD-Q total and “Control_Movement” scores and with SP-2_”Seeking” categories.

Conclusions: Our findings highlight that both sensory and motor impairment can have an influence on AS, AB and RRB in children with ASD and further support the relevance of sensory-motor early intervention in children with ASD and intellectual disability.



427.079 Sex-Specific Associations Among Sensory Features, Behavioral Inflexibility, and Emotional-Behavioral Challenges in Autistic Children

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