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Abstract

Poster Session C

Friday, November 15, 2019 8:00 am – 9:30 am

DEVELOPMENTAL AND PEDIATRIC: OTHER

C-70

Neuropsychological Profiles of Patients With SCN8A-Related Epilepsy

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Objective: Two cases of SCN8A-related epilepsy (a sodium-channelopathy) are presented. SCN8A-related epilepsy with encephalopathy (SCN8A-REE), the most common form, is typically characterized by refractory seizures, developmental delays, and intellectual disability (ID) but recently discovered variants have shown broadly normal cognition. Current cases highlight the heterogeneity seen with differing de-novo pathogenic variants. Method: Case 1, a 6-year-old right-handed girl, presented with SCN8A-REE and a missense pathogenic variant (c.802A>C), not previously documented. History includes speech and motor delay, with focal motor seizures starting at 4-months. Early EEG showed bilateral centroparietal epileptiform discharges. Case 2, an 8-year-old right-handed girl, presented with SCN8A-related epilepsy with c.5630A>G pathogenic variant with seizure onset at 5-months. Initial EEG showed right occipital spikes. Results: Case 1 currently shows motor and language delays and prominent motor tics. Testing documented ID with fairly global neuropsychological deficits (i.e., academics, attention/executive functions, memory, visual-spatial skills, fine motor, language). In contrast, Case 2 shows low average intellect and average academics, but evaluation documented attention deficits, fine motor delays, and behavioral issues in addition to tics; she was diagnosed with Attention-Deficit/Hyperactivity Disorder, Oppositional Defiant Disorder, Obsessive Compulsive Disorder, and Tourette's. Conclusion: These cases expand limited knowledge regarding neuropsychological functioning of children with SCN8A-related epilepsy with unique de-novo pathogenic variants. While SCN8A-REE is clearly associated with ID, other pathogenic variants may show better preserved intellect, despite other neuropsychological and behavioral concerns.

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