

Abstract
Poster Session A

Wednesday, November 13, 2019 5:30 pm – 7:00 pm

NEUROPSYCHOLOGICAL DOMAINS: OTHER

A-58

Rare Case of Klinefelter Syndrome with 13/14 Balanced Translocation and Absence Epilepsy: Impact of Combined Genotypes on Cognitive Neuropsychological Phenotype

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Objective: Klinefelter syndrome (KS) and Robertsonian translocation of 13/14 [rob t(13;14)] are the most common sex-chromosome disorder and chromosome rearrangement, respectively (Engels et al., 2008; Skakkebaek, Wallentin, & Gravholt, 2015). Both are associated with increased risk of cognitive/intellectual disability (ID). A case of KS and de novo (i.e., unbalanced) rob t(13;14) was previously reported (Gül & Şayli, 1994). A case of KS with balanced rob t(13;14) and well-controlled generalized absence epilepsy will be presented with consideration for pediatric neuropsychological practice. **Method:** Neuropsychological evaluation of a 12-year-old, right-handed boy diagnosed with comorbid KS, rob t(13;14), and generalized absence epilepsy. Particular attention was given to language given his KS diagnosis. The patient is in a 12:1:1 self-contained classroom with speech-language therapy and social skills groups in place. The patient's mother is confirmed to have rob t(13;14), whereas paternal contribution is unknown. **Results:** Adolescent with a history of language difficulties, especially comprehension. Recent school-based WISC-V FSIQ was in the extremely low range ($SS = 53$), with weaker verbal comprehension and working memory. Academic achievement was globally very low. Expressive and receptive language, visual perception and motor coordination were extremely low to low average. Verbal list learning and visual attention were near average to average. **Conclusions:** This case contributes to the very limited body of pediatric neuropsychological data on the combined genotype of KS with rob t(13;14) and absence epilepsy. Both the KS and rob t(13;14) cognitive phenotypes have been characterized as highly variable, with the comorbidity a likely increased risk for ID.