P-713 - A DE NOVO MICRODELETION IN CHROMOSOME 8Q12.3Q13.2: ASSOCIATION WITH MILD INTELLECTUAL DISABILITY AND EPILEPSY?

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Introduction: Whole genome microarray techniques are a primary tool for the etiological assessment in patients with intellectual disabilities. As a result, several novel microdeletions have been demonstrated that could be causatively related to the disorder. Interpretation of array results is facilitated through the use of databases such as the European Cytogeneticists Association of Unbalanced Chromosome Abberations (ECARUCA; http://www.ecaruca.net).
Objectives: Clinical interpration of rare *de novo* microdeletions.
Aims: Detailed evaluation of a young female with unexplained intellectual disability.
Methods: Extensive neuropsychological, neurological, and genetic workup in a 9-years-old female patient with a history characterized by delay of psychomotor and speech development, mild to moderate intellectual disability and persistent sleep disturbances since the age of two.
Results: Several dysmorphic features were noticed including hypertelorism, downslanting palpebral fissures, a long, pear shaped nose, and low set, posteriorly rotated ears. Furthermore, she had a pectus excavatum, bilateral flat feet, and a sandal gap. Besides lowered intelligence, neuropsychological functioning disclosed impaired attentional capacities and executive control as well as weak motor skills. MRI of the brain disclosed no abnormalities. EEG demonstrated frequent epileptiform activity centroparietal bilaterally with marked increase during sleep, corresponding with continuous spike-waves during slow sleep (CSWS) syndrome. Array-CGH demonstrated a 3.57 Mb *de novo* microdeletion in chromosome 8q12.3.
Conclusions: This *de novo* 8q12.3q13.2 microdeletion syndrome is characterized by a specific combination of a rare form of juvenile epilepsy (CSWS), neuropsychological dysfunction, impaired language and motor skills.

https://doi.org/10.1016/S0924-9338(12)74880-0 Published online by Cambridge University Press