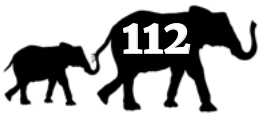

112: Cortical myoclonus as a distinct motor phenotype of 22q11.2 deletion syndrome

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Background: Beyond Parkinson's disease, hyperkinetic movement disorders, including myoclonus, are increasingly recognized as neurologic manifestations associated with 22q11.2 microdeletion, yet neurophysiological and clinical characterization remains limited. **Methods:** We describe findings in adults with a typical 22q11.2 microdeletion who underwent expert neurologic evaluations for myoclonus between 1996 and 2025. Data were obtained through retrospective review, collecting relevant information regarding demographics, genetics, and available electro-clinical data (phenomenology, electromyography, EMG and/ or electroencephalography, EEG). **Results:** Among 290 adults, 15 (5.2%; 53.3% female) were identified with myoclonus based on clinical and neurophysiologic evaluation. Median age at neurological assessment was 29.2 (IQR: 21.7) years, and median age at onset was 24.0 (IQR: 11.4) years. Epilepsy was present in only 4/15 (28.6%) and clozapine-requiring psychotic disorder in 5/15 (33.3%), in addition to the myoclonus diagnosis. Clinically, myoclonus was most often observed at rest (13/15, 86.7%), and predominantly affected the distal upper extremities (12/15, 80.0%), followed by the face (6/15, 40.0%), and proximal upper extremities and trunk (4/15, 26.7% each). Neurophysiologic evidence supported cortical localization and physiology in all cases: short EMG burst duration (15/15, 100%; median 40, IQR: 5.0 ms), and presence of giant somatosensory evoked potentials (6/15, 40.0%), short-latency EEG discharge preceding myoclonic jerks (5/15, 33.3%), and negative myoclonus (2/15, 13.3%). **Conclusions:** This study extends prior findings of cortical myoclonus as a distinct movement disorder entity associated with 22q11.2DS, which may occur independently of epilepsy or clozapine exposure. These findings may support 22q11.2 microdeletion as a genetic etiology of cortical myoclonus. Neurologic assessment is essential for accurate diagnosis, localization, and clinical decision-making, and should include neurophysiologic evaluation.