Myoclonus in Angelman Syndrome

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Angelman Syndrome is a neurogenetic imprinting disorder caused by loss of the maternally inherited *Ube3a* gene, characterized by epilepsy, limited expressive speech, sleep dysfunction, and movement disorders. Myoclonic seizures are often the first seizure type to appear, in rare cases in the first few years of life progressing to myoclonic status, associated with developmental regression. Additionally, there have been rare reports of prolonged episodes of myoclonus without electrographic correlate in adults with Angelman syndrome.

The medical records of 200 individuals seen at the Angelman Syndrome Clinic at the Massachusetts General Hospital and Lurie Center for Autism were retrospectively reviewed to identify and characterize myoclonic events as seizures or non-epileptic myoclonus. Prevalence of myoclonic seizures, myoclonic status and non-epileptic myoclonus were assessed and age of onset, duration of episodes, and electroencephalographic correlate were examined.

The aim of this study was to characterize various myoclonic episodes in Angelman syndrome. Myoclonuic seizures are identified as brief events, unless an individual has myoclonic status, and electroencephalographs reveal generalized spike and wave activity. Non-epileptic myoclonus occurred in nearly half of individuals over ten years of age, with age of onset 10-26 years, and the prevalence appears to increase with age. These events were captured on multiple video electroencephalographs and had no electrographic correlate. Episodes of non-epileptic myoclonus have a discrete beginning and end, no post-ictal period, and are not associated with significant alteration of consciousness or developmental regression. These episodes can be difficult to treat and are often refractory to medication; however, levetiracetam, clobazam, and clonazepam appear to be effective for some individuals. Myoclonic seizures are common in AS, typically occurring in young children and associated with epileptiform changes on electroencephalographs. Prolonged episodes are associated with developmental regression. While non-epileptic myoclonus does not cause regression, it does significantly impact quality of life.