

Given the limited number of patients available for characterization, the full extent of symptoms is yet to be described. The most common features observed include absence seizures, myoclonic-ataxic epilepsy (onset from 7 months to 6 years, mean 3.7 years) and mild-to-moderate intellectual disability. Speech difficulties and behavioral problems have been described. The most common EEG pattern observed comprises irregular, high ample, and generalized spike-and-waves. To date, the most extensive patient collection was published by Johannessen et al⁵ and includes 34 patients. In this cohort, cognitive development was impaired in 33/34 (97%) subjects; 28/34 (82%) had mild to moderate intellectual disability, with language impairment being the most common feature. Epilepsy was diagnosed in 31/34 patients with a mean onset at 3.7 years. Cognitive assessment before epilepsy onset was available in 24/31 subjects and was normal in 25% (6/24). Two patients had speech delay only, and 1 had severe intellectual disability. After epilepsy onset, cognition declined in 46% (11 out of 24) of patients. The most common seizure types were absences, myoclonic, and atonic seizures. Sixteen patients (47%) fulfilled the diagnostic criteria for myoclonic-ataxic epilepsy. Seven additional patients had different forms of generalized epilepsy, and two had focal epilepsy. Electroencephalography (EEG) findings were available in 27/31 patients showing irregular bursts of diffuse 2.5-3.5 Hz spikes/polyspikes-and-slow waves in 25/31. Two patients developed an EEG pattern resembling electrical status epilepticus during sleep. Ataxia was observed in 7 out of 34 patients of Epileptic Encephalopathy (21%).