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| **ID** | **GENIE-P-825** | **HPO terms** |
| **Current age** | 26 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| Preserved cognition |  |
| Drug-resistant seizures |  |
| “Lennox-Gastaut” syndrome |  |
| Sleep-activated EEG |  |
| Ketogenic diet responder |  |
| **Epilepsy syndrome** | Epilepsy with myoclonic atonic seizures |  |
| **Onset of seizures** | 55 months |  |
| **First seizure** | Generalised tonic-clonic seizure | HP:0007334 |
| **Seizure types** | Convulsive status epilepticus  Generalised tonic-clonic  Myoclonic  Myoclonic-atonic  Tonic  Typical absence  Myoclonic-absence  Myoclonic status | HP:0002133  HP:0007334  HP:0002123  HP:0011170  HP:0010818  HP:0002121  HP:0011150  HP:0011151 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Normal |  |
| **Fine motor development** | Normal |  |
| **Vision** | Normal |  |
| **Speech and language development** | Normal |  |
| **Hearing** | Normal |  |
| **Cognition** | Marked deterioration in cognitive assessments following epilepsy onset, but still performed within the normal range |  |
| **Autistic features?** | No |  |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Myoclonic absence: 1-2 episodes per week |  |
| **Effective treatments** | Ketogenic diet |  |
| **Detrimental treatments** | Levetiracetam |  |
| **Other neurological features** | Essential tremor of the hands, left>right | HP:0007351 |
| **Non neurological features** | Developed shingles aged 2.5 years (exposed to chickenpox at birth) |  |
| **Height** | Unknown |  |
| **OFC** | 59..6 (+1.37) |  |
| **Family history** | Older brother: Single febrile seizure, aged 2 years  Mother’s father: epilepsy following cardiac arrest |  |
| **Neuroimaging** | 1996 – CT brain normal |  |
| **Ictal EEGs** | 1995 – Myoclonic jerks accompanied by generalised bursts of irregular spike and wave activity followed by rhythmic 2.5/second spike-wave bursts  1995 – Typical absences brought on by hyperventilation – two bisyhcnronous bursts of high voltage mixed spikes and slow waves at 2.5-3Hz lasting up to 10 seconds | HP:0001326  HP:0010848 |
| **Interictal EEGs** | 1995 – Polyspike and wave runs at 3Hz  2006 – Spikes occupying 30% of sleep. Frequent bursts of polyspike and wave during wakefulness. | HP:0002392 |