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| **ID** | **GENIE-P-25** | **HPO terms** |
| **Current age** | 7 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| “Lennox-Gastaut” syndrome |  |
| Drug-resistant seizures |  |
| Ketogenic diet responder |  |
| Episodes of nonconvulsive status |  |
| Sleep-activated EEG |  |
| **Epilepsy syndrome** | Epilepsy with myoclonic atonic seizures |  |
| **Onset of seizures** | 30 months |  |
| **First seizure** | Atonic seizure | HP:0010819 |
| **Seizure types** | Non-convulsive status  Convulsive  Myoclonic  Atonic  Tonic  Typical absence | HP:0012847  HP:0002069  HP:0002123  HP:0010819  HP:0010818  HP:0011147 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Walked at 12 months. Now unsteady on feet | HP:0002317 |
| **Fine motor development** | Poor fine motor skills | HP:0010862  HP:0002370 |
| **Vision** | Normal |  |
| **Speech and language development** | Normal |  |
| **Hearing** | Normal |  |
| **Cognition** | Moderate intellectual disability | HP:0002342 |
| **Autistic features?** | No |  |
| **Attention/concentration/**  **behaviour difficulties?** | Major difficulties with attention, concentration, impulsivity, and hyperactivity | HP:0100710, HP:0000736, HP:0000752 |
| **Current seizure frequency** | 1-3 convulsive seizure per night  1-3 tonic seizures per night  Daily clusters of typical absence seizures |  |
| **Effective treatments** | Ketogenic diet  Ketamine  Zonisamide  Prednisolone |  |
| **Detrimental treatments** | Lamotrigine |  |
| **Other neurological features** | None |  |
| **Non neurological features** | None |  |
| **Height** | 118.6cm (-0.92) |  |
| **OFC** |  |  |
| **Family history** | Mother’s half-sister’s daughter has well-controlled epilepsy  Father’s brother’s son has had a single febrile convulsion |  |
| **Neuroimaging** | 2012 – MRI brain normal |  |
| **Ictal EEGs** | 2012 - high voltage mixed polyspikes and slow waves are seen bilaterally | HP:0012001 |
| **Interictal EEGs** | 2011 - There is a slight excess of slow activity in the EEG over all areas but no features to aid with syndromic diagnosis.  2012 - In light sleep fairly frequent generalised polyspike wave discharges are seen  2013 – NCSE: frequent multifocal spikes and spike wave complexes | HP:0002392  HP:0011189 |