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| **ID** | **GENIE-P-31** | **HPO terms** |
| **Current age** | 9 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| Focal epilepsy with language disorder |  |
| Electrical status epilepticus in slow wave sleep |  |
| Drug-resistant seizures |  |
| ADHD | HP:0007018 |
| **Epilepsy syndrome** | Epileptic encephalopathy with status epilepticus in slow wave sleep | N/A |
| **Onset of seizures** | 69 months |  |
| **First seizure** | Prolonged focal seizure with impaired awareness | HP:0012847 |
| **Seizure types** | Focal status epilepticus  Convulsive status epilepticus  Focal (plus impaired awareness)  Myoclonic | HP:0012847  HP:0002133  HP:0002384  HP:0002123 |
| **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** | Moderate learning disability | HP:0002342 |
| **Autistic features?** | No |  |
| **Attention/concentration/ behaviour difficulties?** | Yes. ADHD diagnosis | HP:0007018 |
| **Current seizure frequency** | Focal seizures: 2 per day |  |
| **Effective treatments** | Steroids normalise EEG but no clinical improvement  Sodium valproate helps both EEG and seizures |  |
| **Detrimental treatments** | None |  |
| **Other neurological features** | None |  |
| **Non neurological features** | Mild asthma |  |
| **Height** | 137.0cm (+0.41) |  |
| **OFC** | 52.5cm (-1.1) |  |
| **Family history** | Mother migraine (without aura) |  |
| **Neuroimaging** | 2013 – MRI brain mild cerebellar tonsil ectopia, more on right than left |  |
| **Ictal EEGs** | No EEG correlate to events captured |  |
| **Interictal EEGs** | 2013 – bifrontal spike-wave discharges  2014 – multifocal sharp and spike wave activity which becomes more prominent in sleep  2014 – continuous spike wave activity in slow wave sleep | HP:0010850  HP:0011196 |