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| **ID** | **GENIE-P-597** | **HPO terms** |
| **Current age** | 10 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| Drug-resistant seizures |  |
| Clusters of seizures |  |
| Electrical status epilepticus in slow wave sleep |  |
| Nonconvulsive status epilepticus |  |
| “Lennox-Gastaut” syndrome |  |
| Ketogenic diet responder |  |
| **Epilepsy syndrome** | Epilepsy with myoclonic atonic seizures |  |
| **Onset of seizures** | 48 months |  |
| **First seizure** | Generalised tonic-clonic seizure | HP:0007334 |
| **Seizure types** | Convulsive status epilepticus and clusters  Focal (plus impaired awareness)  Bilateral convulsive (GTCS)  Myoclonic  Atonic  Tonic  Typical absence | HP:0012847  HP:0002384  HP:0007334  HP:0002123  HP:0010819  HP:0010818  HP:0002121 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Normal until epilepsy onset – walked at < 1 year. Now delayed (learning to ride bike aged 10) | HP:0002317 |
| **Fine motor development** | Normal |  |
| **Vision** | Normal |  |
| **Speech and language development** | Slow at reading | P:0010522 |
| **Hearing** | Normal |  |
| **Cognition** | Moderate learning disability | HP:0002342 |
| **Autistic features?** | No |  |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Tonic: Most nights  Absence: Occasional |  |
| **Effective treatments** | Rufinamide – effective for drops  Ketogenic diet – all seizure types |  |
| **Detrimental treatments** | None |  |
| **Other neurological features** | None |  |
| **Non neurological features** | Recurrent ear infections as an infant |  |
| **Height** | 131.0cm (-0.87) |  |
| **OFC** | 52.0cm (-1.48) |  |
| **Family history** | Younger half brother (to father) has speech delay |  |
| **Neuroimaging** | 2011 – MRI brain: Persistent abnormal area of increased signal in the left occipital lobe in keeping with cortical dysplasia (compared with scans from 01/06/2011) | HP:0002539 |
| **Ictal EEGs** | No captured events have ever had an EEG correlate |  |
| **Interictal EEGs** | 2011 - Abnormal EEG background showing high amplitude moderately slow activity. In addition infrequent bursts of ill-defined irregular bilateral spikes seen with frontal emphasis and sometimes followed by run of slow waves  2011- Episodes of high amplitude, bisynchronous, spike and slow wave activity are seen, more frequently in the morning  2011- Runs of generalised spike/polyspike and wave are seen during events and when he is less alert | HP:0010845  HP:0011193  HP:0010850  HP:0002392 |