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| **ID** | **GENIE-P-412** | **HPO terms** |
| **Current age** | 10 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| Drug-resistant seizures |  |
| “Lennox-Gastaut” syndrome |  |
| Clusters of seizures |  |
| Electrical status epilepticus in slow wave sleep |  |
| Relative macrocephaly | HP:0004482 |
| **Epilepsy syndrome** | Epilepsy with myoclonic atonic seizures | N/A |
| **Onset of seizures** | 41 months |  |
| **First seizure** | Tonic seizure | HP:0010818 |
| **Seizure types** | Non-convulsive status epilepticus  Focal (plus impaired awareness)  Bilateral convulsive (GTCS)  Myoclonic  Myoclonic-atonic  Tonic  Typical absence | HP:0012847  HP:0002384  HP:0007334  HP:0002123  HP:0011170  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 10 months. Now slightly unsteady gait | HP:0002317 |
| **Fine motor development** | Normal until epilepsy onset – no poor coordination | HP:0002370 |
| **Vision** | Normal |  |
| **Speech and language development** | Poor articulation | HP:0009088 |
| **Hearing** | Normal |  |
| **Cognition** | Moderate learning disability | HP:0002342 |
| **Autistic features?** | No |  |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Bilateral convulsive: rarely  Tonic: Clusters every morning on waking  Absence: daily |  |
| **Effective treatments** | Clobazam  Levetiracetam  Ethosuximide |  |
| **Detrimental treatments** | Topiramate (drowsy) |  |
| **Other neurological features** | None |  |
| **Non neurological features** | Single right kidney  Left inguinal hernia repaired | HP:0000122  HP:0000023 |
| **Height** | 138.0cm (0.08) |  |
| **OFC** | 57.5cm (+1.92) | HP:0004482 |
| **Family history** | None |  |
| **Neuroimaging** | 2014 – MRI brain normal |  |
| **Ictal EEGs** | 2015 – bisynchronous slow spike wave discharges during tonic seizures | HP:0010847 |
| **Interictal EEGs** | 2015 – sleep activated frontocentrotemporal spikes, left> right  2015 – bisynchronous irregular spike wave complexes at 2-3 per second  2016 – frequent runs of high amplitude spike wave with a frontal empahsis | HP:0001326 |