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| **ID** | **GENIE-P-431** | **HPO terms** |
| **Current age** | 5 years |  |
| **Tags** | (Infantile-onset) epileptic encephalopathy | HP:0200134 |
| Focal epilepsy with language disorder |  |
| Clusters of seizures |  |
| Microcephaly | HP:0000252 |
| Short stature | HP:0004322 |
| **Epilepsy syndrome** | Unclassified | N/A |
| **Onset of seizures** | 8.5 months |  |
| **First seizure** | Focal seizure | HP:0012847 |
| **Seizure types** | Focal seizures  Status epilepticus | HP:0007359  HP:0002133 |
| **Developmental concerns prior to epilepsy onset?** | Yes |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Walks with a normal gait but falls easily | HP:0002527 |
| **Fine motor development** | Scribbles but cannot draw | HP:0010862 |
| **Vision** | Normal |  |
| **Speech and language development** | About 50 single words aged 5 years. Attends a specialist language unit | HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Moderate learning disability | HP:0002342 |
| **Autistic features?** | Yes, but no formal diagnosis (yet) | HP:0000729 |
| **Attention/concentration/ behaviour difficulties?** | Yes, but not diagnosed ADHD | HP:0000736 |
| **Current seizure frequency** | Seizure-free since starting Levetiracetam |  |
| **Effective treatments** | Levetiracetam |  |
| **Detrimental treatments** | None |  |
| **Other neurological features** | None |  |
| **Non neurological features** | Yellow discolouration of teeth | HP:0000705 |
| **Height** | 101.0cm (-2.1) | HP:0004322 |
| **OFC** | 49.5cm (-2.5) | HP:0000252 |
| **Family history** | None |  |
| **Neuroimaging** | 2011 – MRI Normal |  |
| **Ictal EEGs** | 2012 – focal seizures captured arising from one or other hemisphere. Bilateral suppression, followed by spike wave complexes (right temporal). Also fast discharges on left hemisphere with one seizure. | HP:0012013 |
| **Interictal EEGs** | 2011 - High amplitude slow background in keeping with cluster of epileptic seizures. Occasional independent right or left hemisphere sharp waves more prominent over right temporal region  2012 – slow background  2012 - Frequent sharp and slow wave complexes occur independently over both hemispheres. | HP:0010841  HP:0010845  HP:0011195 |