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| **ID** | **GENIE-P-506** | **HPO terms** |
| **Current age** | 16 years |  |
| **Tags** | (Infantile-onset) epileptic encephalopathy | HP:0200134 |
| “Lennox-Gastaut” syndrome |  |
| Drug-resistant seizures |  |
| Epileptic spasms | HP:0012469 |
| Clusters of seizures |  |
| Microcephaly | HP:0000252 |
| Short stature | HP:0004322 |
| **Epilepsy syndrome** | Lennox-Gastaut syndrome | N/A |
| **Onset of seizures** | 5 months |  |
| **First seizure** | Generalised tonic-clonic seizure | HP:0002069 |
| **Seizure types** | Generalised tonic-clonic seizures  Infantile spasms and epileptic spasms  Atypical absence seizures | HP:0002069  HP:0012469  HP:0007270 |
| **Developmental concerns prior to epilepsy onset?** | Yes |  |
| **Regression associated with epilepsy onset?** | Not clearly |  |
| **Gross motor development** | Has never walked or crawled. No head control at 5 months (before first seizure) | HP:0002194 |
| **Fine motor development** | Severe delay | HP:0010862 |
| **Vision** | Cerebral visual impairment | HP:0100704 |
| **Speech and language development** | Babbles smiles and laughs. Very little understanding | HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Profound learning disability | HP:0002187 |
| **Autistic features?** | Not clearly |  |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Clusters of GTCS every 3-4 months (7 seizures over a 24-72 hour period)  Atypical absences: 2-3 times per year  Spasms: very occasionally |  |
| **Effective treatments** | Levetiracetam  Sodium valproate  Nitrazepam effective for clusters |  |
| **Detrimental treatments** | None |  |
| **Other neurological features** | Progressive microcephaly  Lower limb hypertonia | HP:0000253  HP:0006895 |
| **Non neurological features** | Gastrostomy fed |  |
| **Height** | 141.0cm (-3.59) | HP:0004322 |
| **OFC** | 48.0cm (-5.3) | HP:0000252 |
| **Family history** | None |  |
| **Neuroimaging** | 2011 - Mild asymmetric posterior white matter volume loss. Myelination appropriate |  |
| **Ictal EEGs** | 2012 – No seizures captured |  |
| **Interictal EEGs** | 2012 - Focal frontotemporal sharp waves  2012 - Virtually continuous 3-4Hz theta activity, which at times had a sharper appearance, was seen to phase reverse over the right. This activity was occasionally reflected over the left. | HP:0011294  HP:0011296 |