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| **ID** | **GENIE-P-195** | **HPO terms** |
| **Current age** | 16 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| “Lennox-Gastaut” syndrome |  |
| Drug-resistant seizures |  |
| Ketogenic diet responder |  |
| Episodes of nonconvulsive status |  |
| Seizure clusters |  |
| Short stature | HP:0004322 |
| Relative macrocephaly | HP:0004482 |
| **Epilepsy syndrome** | Unclassified polymorphic epilepsy |  |
| **Onset of seizures** | 22 months |  |
| **First seizure** | Cluster of GTCS | HP:0002069 |
| **Seizure types** | Non-convulsive status epilepticus  Generalised tonic-clonic seizures  Myoclonic seizures  Atonic seizures  Absence seizures  Epileptic spasms | HP:0002133  HP:0002069  HP:0002123  HP:0010819  HP:0002121  HP:0011097 |
| **Developmental concerns prior to epilepsy onset?** | Was not walking at 22 months  Had a few single words at 22 months |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Walked at 3.5 years | HP:0002194 |
| **Fine motor development** | Cannot write. Can colour in | HP:0010862 |
| **Vision** | Normal |  |
| **Speech and language development** | Had a few single words by aged 2 years, but no sentences. Clear regression of language after epilepsy onset. Communicates with single words, pointing, and gestures. Can follow adult conversation | HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Severe intellectual disability | HP:0010864 |
| **Autistic features?** | Yes, but no formal diagnosis |  |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | > 30 epileptic spasms per day. Clusters of early morning GTCS approx. every 6 months. |  |
| **Effective treatments** | Ketogenic diet – dramatic reduction in absence seizures  Lorazepam – effective for clusters of seizures |  |
| **Detrimental treatments** |  |  |
| **Other neurological features** | Macrocephaly | HP:0000256 |
| **Non neurological features** |  |  |
| **Height** | 139cm (-2.07) | HP:0004322 |
| **OFC** | 55cm (+0.35) | HP:0000252 |
| **Family history** | Brother has myoclonic astatic epilepsy and relative macrocephaly. Father had a single febrile convulsion and has macrocephaly |  |
| **Neuroimaging** | 08/11/2005 – normal MRI  19/02/2010 – normal MRI and MRS |  |
| **Ictal EEGs** | 2005 - Abnormal background with frequent generalised polyspikes | HP:0012001 |
| **Interictal EEGs** | 2005 - Non-convulsive status. Irregular slow spike (1-3 per second) and wave associated with twitching and cessation of activity. | HP:0001326  HP:0010847 |