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| **ID** | **GENIE-P-14** | **HPO terms** |
| **Current age** | 22 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| “Lennox-Gastaut” syndrome |  |
| Drug-resistant seizures |  |
| Ketogenic diet responder |  |
| Episodes of nonconvulsive status |  |
| Microcephaly | HP:0000252 |
| Short stature | HP:0004322 |
| **Epilepsy syndrome** | Unclassified polymorphic epilepsy |  |
| **Onset of seizures** | 13 months |  |
| **First seizure** | Febrile convulsion | HP: 0011171 |
| **Seizure types** | Febrile convulsions  Status epilepticus  Generalised tonic-clonic seizures  Myoclonic seizures  Atonic seizures  Absence seizures  Epilepsy partialis continua | HP:0011171  HP:0002133  HP:0002069  HP:0002123  HP:0010819  HP:0002121  HP:0012847 |
| **Developmental concerns prior to epilepsy onset?** | Yes but mainly regarding weight gain |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Independently mobile. Rides horses. Kicks a ball. | HP:0002194 |
| **Fine motor development** | Scribbles but cannot draw | HP:0010862 |
| **Vision** | Good with glasses |  |
| **Speech and language development** | 30-40 words before seizure-onset. Now only makes noises. Understands a lot. | HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Probably moderate intellectual disability | HP:0002342 |
| **Autistic features?** | No |  |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | 4-5 convulsive seizures per day and multiple absences |  |
| **Effective treatments** | Phenytoin – dose dependant improved seizure control  Ketogenic diet – improved cognition and EEG  Rufinamide – effective for drop seizures  Clobazam – effective for myoclonic seizures  Steroids – reversed epilepsy partialis continua |  |
| **Detrimental treatments** | Hyoscine |  |
| **Other neurological features** | Lower limb spasticity and contractures | HP:0006895 |
| **Non neurological features** | Failure to thrive in infancy  Short stature  Obstructive sleep apnoea | HP:0001508  HP:0004322  HP:0002870 |
| **Height** | 140cm (-5.36) | HP:0004322 |
| **OFC** | 53.2cm (-2.37) | HP:0000252 |
| **Family history** | Brother stillborn at 27 weeks |  |
| **Neuroimaging** | Cranial USS 15/01/1997 - normal |  |
| **Ictal EEGs** | 1999: Bursts of high voltage spike-wave and polyspike wave over all areas  2002: High voltage right hemisphere partial status (continuous irregular spike and slow wave complexes) – resolved by high dose steroids  2002: Virtually continuous bisynchronous high amplitude slow (1-1.5/sec) spike and wave | HP:0002392  HP:0011197  HP:0010847 |
| **Interictal EEGs** | 1999: Photic stimualtion induced 2 short bursts of 4-5/second spike/polyspike and wave | HP:0010849  HP:0002392 |