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| **ID** | **GENIE-P-913** | **HPO terms** |
| **Current age** | 7 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| (Infantile-onset) epileptic encephalopathy | HP:0200134 |
| Clusters of seizures |  |
| Sleep-activated EEG |  |
| Microcephaly | HP:0000252 |
| **Epilepsy syndrome** | Unclassified myoclonic epilepsy | N/A |
| **Onset of seizures** | Day 1 of life |  |
| **First seizure** | Myoclonic seizures | HP:0002123 |
| **Seizure types** | Myoclonic  Eyelid myoclonia | HP:0002123  HP:0011168 |
| **Developmental concerns prior to epilepsy onset?** | N/A |  |
| **Regression associated with epilepsy onset?** | No (seizures first noticed on day 1 of life) |  |
| **Gross motor development** | Normal |  |
| **Fine motor development** | Struggled a little with drawing at nursery |  |
| **Vision** | Normal |  |
| **Speech and language development** | Normal |  |
| **Hearing** | Normal |  |
| **Cognition** | Difficulties with memory and special perception | HP:0002354  HP: 0010794 |
| **Autistic features?** | Yes, but no formal diagnosis | HP:0000723 |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Myoclonic seizures: now rare, clusters when has a viral infection |  |
| **Effective treatments** | Ethosuximide |  |
| **Detrimental treatments** | Topiramate (increased seizures and caused hallucincations) |  |
| **Other neurological features** | Nocturnal vomiting episodes of uncertain aetiology |  |
| **Non neurological features** | Asthma  Neonatal hypoglycaemia | HP:0002099  HP:0001998 |
| **Height** | 115.5cm (-1.11) |  |
| **OFC** | 49.0cm (-2.99) | HP:0000252 |
| **Family history** | Younger brother: Single complex febrile seizure, aged 3 years  Father: migraine (occasionally hemiplegic) |  |
| **Neuroimaging** | 2015 – MRI brain normal |  |
| **Ictal EEGs** | 2014 – brief bisynchronous bursts of spike-wave activity | HP:0010850 |
| **Interictal EEGs** | 2014 – Brief bisynchronous episodes of frontally dominant mixed spikes and slow waves are seen. Increase during periods of sleep  2014 – several episodes of 3-4 Hz bilateral spike and wave bursts of varying duration | HP:0010849 |