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| **ID** | **GENIE-P-958** | **HPO terms** |
| **Current age** | 14 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| “Lennox-Gastaut” syndrome |  |
| Nonconvulsive status epilepticus | HP:0012847 |
| Drug-resistant seizures |  |
| Ketogenic diet responder |  |
| Autism | HP:0000717 |
| **Epilepsy syndrome** | Epilepsy with myoclonic atonic seizures |  |
| **Onset of seizures** | 12 months (febrile)  3.5 years (afebrile) |  |
| **First seizure** | Febrile convulsion | HP:0011171 |
| **Seizure types** | Simple febrile convulsions  Non-convulsive status epilepticus  Focal (plus impaired awareness)  Generalised tonic-clonic  Myoclonic  Myoclonic-atonic  Atonic  Absence-atonic (atypical absence)  Tonic  Typical absence | HP:0011171  HP:0012847  HP:0002384  HP:0007334  HP:0002123  HP:0011170  HP:0010819  N/A (HP:0007270)  HP:0010818  HP:0002121 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | Yes, after focal seizures began at 3.5 years | HP:0006834 |
| **Gross motor development** | Walks with a clumsy gait | HP:0002317 |
| **Fine motor development** | Can draw to the standard of a 7 year-old  Limb dyspraxia | HP:0010862  HP:0007057 |
| **Vision** | Normal |  |
| **Speech and language development** | Immature language | HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Moderate learning disability | HP:0002342 |
| **Autistic features?** | Yes. Formal diagnosis | HP:0000717 |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Focal: daily  Bilateral convulsive: every night  Tonic: 1-2 per week |  |
| **Effective treatments** | Clobazam  Ketogenic diet  Sodium valproate  Vagus nerve stimulation |  |
| **Detrimental treatments** | Carbamazepine – increased all seizure types |  |
| **Other neurological features** | None |  |
| **Non neurological features** | Cow’s milk allergy | HP:0100327 |
| **Height** | Unknown |  |
| **OFC** | 57.0cm (+0.63 SD) |  |
| **Family history** | None |  |
| **Neuroimaging** | 2006 – MRI brain normal |  |
| **Ictal EEGs** | 2006 - 2-2.5/second spike/polyspike bifrontally associated with absences  2007 – Slow spike and wave 2-2.4/second associated with eye flickering  2007 – generalised spike and wave associated with absences  2007 – frequent high voltage bisynchronous spike-wave activity at 1.5-2 per second more marked over left hemisphere | HP:0012001  HP:0010847 |
| **Interictal EEGs** | 2006 – frontally dominant slow, left sided spike discharges and, spike-wave bursts  2006 – poorly organised and markedly slow  2007 – frequent bursts of irregular spike and wave activity of variable frequencies  2007 – high amplitude spike and wave, slow background with little organisation (NCS)  2009 – continuous slow spike and wave discharge with frontal emphasis | HP:0011193/HP:0011197  HP:0010845  HP:0001236  HP:0012010 |