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| **ID** | **GENIE-P-830** | **HPO terms** |
| **Current age** | 3 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| Drug-resistant seizures |  |
| “Lennox-Gastaut” syndrome |  |
| Clusters of seizures |  |
| Movement disorder (intermittent ataxia) | HP:0002131 |
| **Epilepsy syndrome** | Unclassified focal and generalised epilepsy | N/A |
| **Onset of seizures** | Febrile: 4 months  Afebrile: 24 months |  |
| **First seizure** | Complex febrile convulsion | HP:0011172 |
| **Seizure types** | Complex febrile  Generalised tonic-clonic  Focal with impaired awareness  Myoclonic  Atonic  Typical absence | HP:0011172  HP:0007334  HP:0002384  HP:0002123  HP:0010819  HP:0002121 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | No |  |
| **Gross motor development** | Walked at one year, but now has intermittent episodes of ataxia (once per week) | HP:0002131 |
| **Fine motor development** | Difficult with coordination. Cannot copy a circle (aged 3.5y) | HP:0002370  HP:0010862 |
| **Vision** | Good with glasses |  |
| **Speech and language development** | Normal |  |
| **Hearing** | Normal |  |
| **Cognition** | Normal |  |
| **Autistic features?** | No |  |
| **Attention/concentration/ behaviour difficulties?** | Yes | HP:0000736 |
| **Current seizure frequency** | Focal with impaired awareness: 4 times per week  Clusters of GTCS: Every few months  Myoclonic: Daily  Atonic: Sporadically  Typical absence: daily |  |
| **Effective treatments** | Sodium valproate |  |
| **Detrimental treatments** |  |  |
| **Other neurological features** | Episodes ataxia (lasting 2 mins to all day) with accompanying “moodiness” once per week | HP:0002131 |
| **Non neurological features** | Left bronchomalacia  Asthma | HP:0002780  HP:0002099 |
| **Height** | 100.8cm (-0.2) |  |
| **OFC** | 52.2cm (+1.71) |  |
| **Family history** | None |  |
| **Neuroimaging** | 2015 – MRI: Incidental dominant left vertebral artery, with a small right vertebral artery. |  |
| **Ictal EEGs** | 2016 – no events captured |  |
| **Interictal EEGs** | 2016 – normal background rhythms |  |