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| **ID** | **GENIE-P-951** | **HPO terms** |
| **Current age** | 7 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| Epilepsy with myoclonic atonic seizures |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| Drug-resistant seizures |  |
| “Lennox-Gastaut” syndrome |  |
| Sleep-activated EEG |  |
| Movement disorder (ataxia) | HP:0002066 |
| **Epilepsy syndrome** | Epilepsy with myoclonic atonic seizures |  |
| **Onset of seizures** | 32 months |  |
| **First seizure** | Cluster of generalised tonic-clonic seizures | HP:0007334 |
| **Seizure types** | Convulsive status epilepticus  Generalised tonic-clonic  Myoclonic  Myoclonic-atonic  Typical absence | HP:0002133  HP:0007334  HP:0002123  HP:0011170  HP:0002121 |
| **Developmental concerns prior to epilepsy onset?** | Yes – referred to SALT |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Walked at 15 months, but walks with a wide-based hip-dropping gait | HP:0002066 |
| **Fine motor development** | Has seen OT for difficulties with use of cutlery | HP:0010862 |
| **Vision** | Normal |  |
| **Speech and language development** | Was referred to SALT for concerns about speech delay (before epilepsy onset) | HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Moderate intellectual disability | HP:0002342 |
| **Autistic features?** | Yes, ASD assessment ongoing | HP:0000729 |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Myoclonic: 1-2 episodes per week  Myoclonic-atonic: 1-2 per week |  |
| **Effective treatments** | Nitrazepam – effective for myoclonic-atonics  Levetiractem – very effective for GTCS and myoclonic-atonics |  |
| **Detrimental treatments** |  |  |
| **Other neurological features** | Postural hand tremor (bilateral)  Ataxic gait | HP:0007351  HP:0002066 |
| **Non neurological features** | Irritable hip |  |
| **Height** | 121.0cm (-0.55) |  |
| **OFC** | 51.5cm (-1.38) |  |
| **Family history** | None |  |
| **Neuroimaging** | 2009 – MRI brain 2 small areas of deep white matter abnormality posteriorly adjacent to the ventricles (3mm diameter) – suggestive of ectopic grey matter, though location is unusual |  |
| **Ictal EEGs** | 2012 – Myoclonic-atonic seizure: burst of generalised irregular spike/polyspike and wave activity (about 2.5Hz). | HP:0002392  HP:0001326  HP:0010848 |
| **Interictal EEGs** | 2012 – Photic stimulation at 3 and 18 Hz elicits bursts of spike wave activity anteriorly and very high amplitude posterior slow wave complexes similar to those seen interictally (? giant VEPs) | HP:0010843 |