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| **ID** | **GENIE-P-780** | **HPO terms** |
| **Current age** | 9 years |  |
| **Tags** | Complex myoclonic epilepsy |  |
| (Childhood-onset) epileptic encephalopathy | HP:0200134 |
| Focal epilepsy with language disorder |  |
| Sleep-activated EEG |  |
| Drug-resistant seizures |  |
| Autism | HP:0000717 |
| **Epilepsy syndrome** | Unclassified focal and generalised epilepsy | N/A |
| **Onset of seizures** | 36 months |  |
| **First seizure** | Focal seizure with autonomic features | HP:0011154 |
| **Seizure types** | Focal status epilepticus  Focal seizures  Convulsive status epilepticus  Myoclonic head drops  Typical absence seizures | HP:0012847  HP:0002384  HP:0002133  HP:0002123  HP:0011147 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Poor coordination. Described as “clumsy” | HP:0002312 |
| **Fine motor development** | Normal |  |
| **Vision** | Normal |  |
| **Speech and language development** | Significant deceleration in language acquisition since onset of seizures | HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Mils learning disability | HP:0001256 |
| **Autistic features?** | Yes. Formal diagnosis | HP:0000717 |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Typical absence seizures: once per week |  |
| **Effective treatments** | Clobazam – effective for sleep-transition related seizures |  |
| **Detrimental treatments** | Carbamazepine – onset of myoclonus associated with starting this |  |
| **Other neurological features** | Overactive bladder – on Tolterodine  Left eye esotropia  Pes planus |  |
| **Non neurological features** | None |  |
| **Height** | 131.3cm (-0.26) |  |
| **OFC** | 55.3 (+1.54) |  |
| **Family history** | None |  |
| **Neuroimaging** | 2010 – mild symmetrical prominence of the frontal extra-axial spaces |  |
| **Ictal EEGs** | 2011 – ? Generalised spike-wave activity, with a frontal predominance (not seen actual report) | HP:0010850 |
| **Interictal EEGs** | 2011 – Bisynchronous bursts of irregular spike/polyspike and wave activity  2014 – Generalised spike/polyspike wave activity during sleep. | HP:0001326  HP:0002392 |