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| **ID** | **GENIE-P-152** | **HPO terms** |
| **Current age** | 22 years |  |
| **Tags** | (Infantile onset) epileptic encephalopathy | HP:0200134 |
| Drug-resistant seizures |  |
| “Lennox-Gastaut” syndrome |  |
| Sleep-activated EEG | HP:0011097 |
| Epileptic spasms | HP:0011097 |
| **Epilepsy syndrome** | Unclassified polymorphic epilepsy |  |
| **Onset of seizures** | 9 months |  |
| **First seizure** | Febrile convulsion | HP: 0011171 |
| **Seizure types** | Febrile convulsions  Status epilepticus  Generalised tonic-clonic seizures  Atonic seizures  Absence seizures  Epileptic spasms | HP: 0011171  HP:0002133  HP:0002069  HP:0010819  HP:0002121  HP:0011097 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | Yes | HP:0006834 |
| **Gross motor development** | Poor balance and clumsy gait | HP:0010521 |
| **Fine motor development** | Some difficulties e.g. cannot tie shoelaces | HP:0002370 |
| **Vision** | Good with glasses |  |
| **Speech and language development** | Verbal IQ on WISC IV (2006) = 93 (low normal range) | HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Mild intellectual disability | HP:0001256 |
| **Autistic features?** | Yes but no formal diagnosis | HP:0000729 |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Nocturnal GTCS: once every 2 weeks |  |
| **Effective treatments** | Pregabalin  Sodium valproate/Lamotrigine combination |  |
| **Detrimental treatments** | Levetiracetam (behaviour)  Ketamine (behaviour ++) |  |
| **Other neurological features** | Right leg weakness (aetiology unclear) |  |
| **Non neurological features** | Asthma  Recurrent shoulder dislocation  Brittle dysplastic finger and toe nails | HP:0002099  HP:0003834  HP:0002164 |
| **Height** | 159.2cm (-0.65) |  |
| **OFC** |  |  |
| **Family history** | Half brother to mother – congenital Horner’s syndrome plus anhydrosis  Mother – epilepsy  Father – recurrent febrile seizures |  |
| **Neuroimaging** | MRI 2010 - normal |  |
| **Ictal EEGs** | 2003: Runs of high amplitude bisynchronous 2-3Hz polyspike and wave activity | HP:0002392 |
| **Interictal EEGs** | 2003: Frequent bursts of high amplitude bisynchronous polyspike and wave during sleep | HP:0002392 |