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| **ID** | **GENIE-P-22** | **HPO terms** |
| **Current age** | 19 years |  |
| **Tags** | (Early infantile-onset) epileptic encephalopathy |  |
| Autism | HP:0000717 |
| Drug-resistant seizures |  |
| Focal epilepsy with language disorder |  |
| Movement disorder (stooped gait) | HP:0001288 |
| **Epilepsy syndrome** | Unclassified focal epilepsy with startle-triggered seizures |  |
| **Onset of seizures** | 2 months |  |
| **First seizure** | Focal seizures | HP:0007359 |
| **Seizure types** | Focal seizures with impairment of awareness | HP:0002384 |
| **Developmental concerns prior to epilepsy onset?** | No |  |
| **Regression associated with epilepsy onset?** | Yes |  |
| **Gross motor development** | Walked at 22 months. Mobile but with stooped gait. | HP:0002194  HP:0001288 |
| **Fine motor development** | Has difficulties but can write own name and use an ipad | HP:0010862 |
| **Vision** | Normal |  |
| **Speech and language development** | Stammer. Good vocab but little use of sentences. Good understanding | HP:0009088  HP:0000750 |
| **Hearing** | Normal |  |
| **Cognition** | Severe learning disability  Cannot read or write (apart form own name) | HP:0010864 |
| **Autistic features?** | Formal diagnosis from neuropsychology | HP:0000717 |
| **Attention/concentration/ behaviour difficulties?** | No |  |
| **Current seizure frequency** | Focal seizures: average 2 per night |  |
| **Effective treatments** | Sodium valproate  Clobazam  Sulthiame  Lamotrigine |  |
| **Detrimental treatments** | None |  |
| **Other neurological features** | Stooped gait  Lower limb hypertonia  Clinodactyly little finger left hand | HP:0001288  HP:0006895  HP:0004209 |
| **Non neurological features** | Chronic constipation | HP:0002019 |
| **Height** | 157cm (-1.1) |  |
| **OFC** | 55.5 (-0.01) |  |
| **Family history** | None |  |
| **Neuroimaging** | 2006 – MRI normal |  |
| **Ictal EEGs** | 1997 – left temporal sharp waves | HP:0011296 |
| **Interictal EEGs** | 1997 – rhythmic build up of moderate voltage 4-5Hz theta over left temporal area, spreading to all areas and becoming high voltage regular fast spiking | HP:0012018  HP:0011188 |