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| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient No** | **Sex** | **Age**  **(**months) | **Age of seizure onset** | **Consanguinity** | **Clinic features at presentation** | **Brain MRI** | **EEG** | **Genetic analysis** | **Treatment** | **Neurologic**  **outcome** |
| 1 | F | 11 | 5  months | No | Visual impairment, generalized tonic, and myoclonic seizures, hypotonicity, hyperreactive deep tendon reflexes | Normal | İrregular background rhythm, and epileptic activity at both occipital regions | CDKL5 gene (c.2222C>G) heterozygote variant | Levetiracetam, valproic acid, clobazam, ketogenic diet, cannabidiol | Short-term seizures continue, increase in social development |
| 2 | F | 36 | 2 months | No | Epileptic spasm, can sit with assistance, not walking. Uses 20-30 words, no sentences | Normal | İrregular background rhythm, multifocal and generalized epileptic activity | CDKL5 gene p.E21K (c.61G>A)  heterozygote variant | Vigabatrin,  [Adrenocorticotropic hormone (ACTH),](https://medlineplus.gov/lab-tests/adrenocorticotropic-hormone-acth/)  ketogenic diet | Seizures continue, she started to say words |
| 3 | M | 30 | 2 months | No | Generalized tonic, and myoclonic seizures, can sit with assistance, no walking, no talking | Mild cerebral atrophy, and enlargement of ventricles due to central atrophy | İrregular background rhythm, multifocal and generalized epileptic activity | CDKL5 gene  p.R735fs (c.2205\_2206del)  hemizygous variant | Vigabatrin, clobazam, valproic acid, ACTH | Seizures continue |
| 4 | F | 10 | 1 month | Yes | Generalized clonic seizures, hypotonic, hyperreactive deep tendon reflexes | Normal | İrregular background rhythm, multifocal and generalized epileptic activity | CDKL5 gene  c.554+4A>G  heterozygote variant | Levetiracetam, valproic acid, clobazam, lamotrigine | Reduction in seizures |
| 5 | F | 70 | 5 months | No | Epileptic spasm,and generalized tonic hypotonic, walks unsteadily | Normal | İrregular background rhythm, multifocal and generalized epileptic activity | CDKL5 gene  c.282+3A>T  heterozigot | Phenobarbital, vigabatrin, valproic acid, topiramate | Reduction in seizures |
| 6 | F | 12 | 1 month | No | Generalized clonic seizures, no object tracking  hypotonic, no walking | Normal | İrregular background rhythm, and epileptic activity at both occipital regions | CDKL5 gene p.L220P; c.659T>C) | Levetiracetam, clobazam, lamotrigine, ketogenic diet | Significant decrease in seizures, increase in social development |
| 7 | F | 18 | 3 months | No | Focal clonic | Normal | Normal background rhythm, and temporal spikes | CDKL5 gene p.(His844Thrfs\*66  heterezigot | Levetiracetam | Seizures continue |

**Table 1. The demographic and clinical data, brain MRI and EEG features, and prognosis of patients with a CDKL5 mutation**