**Table 1. Clinical description of the four reported individuals**

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | **Family I** | | | **Family II** |
|  | **Individual I** | **Individual II** | **Individual III** | **Individual IV** |
| Antenatal findings | none | none | none | none |
| Geographical origin | Turkısh | Turkısh | Turkısh | Turkısh |
| Consanquinity | yes | yes | yes | yes |
| Gender | male | female | female | male |
|  |  |  |  |  |
| **Neonatal Period** |  |  |  |  |
| Term (weeks) | full term | full term | full term | full term |
| Birth weight (g) | 3,400 | 3,090 | 3,000 | 3,570 |
| Birth length (cm) | 51 | 50 | 49 | 52 |
| Birth OFC (cm) | 34 | 34 | 34 | 35 |
| APGAR score (1/3/5 min) | 10/10/10 | 10/10/10 | 10/10/10 | 10/10/10 |
| **Disease Onset** |  |  |  |  |
| Age at first symptoms | 4months | 4months | 4months | 3months |
| First symptom type | Hypotonia,  nystagmus  seizures, | Hypotonia,  nystagmus  seizures, | Hypotonia,  nystagmus  seizures, | Hypotonia,  seizures |
| Seizure type | infantile spasms, generalized tonic clonic, focal myoclonic | infantile spasms, myoclonic | infantile spasms, myoclonic | infantile spasms, generalized tonic clonic, focal myoclonic |
| First status epilepticus | 4 months | 6 months | 6 months | NA |
| **Evolution** |  |  |  |  |
| Age at last visit | 28 months | 7 years | 7years | 123 months |
| **Neurodevelopmental**  **Evolution** |  |  |  |  |
| Seizures | pharmaco-  resistant | stopped | stopped | pharmaco-  resistant |
| **Motor development** |  |  |  |  |
| Eye contact | poor | none | poor | none |
| Sitting position | no | no | Sitting without support, knees by holding on | no |
| Walking | no | no | no | no |
| Speech | no | no | no | no |
| **Clinical Evaluation** |  |  |  |  |
| Facial dysmorphism | - | - | - | -­ |
| Dermatological  findings | - | - | - | - |
| Extremities | - | - | - | Flexion contractures in the hands, drop in the feet (club foot deformites) |
| **Neurological Examination** |  |  |  |  |
| Axial hypotonia | + | + | + | + |
| Peripheral hypotonia | - | + | + | - |
| Weak deep tendon reflexes | - | + | + | - |
| Median stereotypies | + | + | + | + |
| Dyskinesia | + | + | + | + |
| Sleep disorders | - | - | - | - |
| Behavioral disorder | + | + | + | + |
| **Ophthalmological Examination** |  |  |  |  |
| Fundus | normal | normal | normal | Optic nerve pallor and hypoplasia |
| ERG, VEP | NA | NA | NA | NA |
| **Malformative Workup** |  |  |  |  |
| Brain MRI | Myelination markedy reversed | Cerebral atrophy,  Expansion in  extraaxial distance | Cerebral atrophy,  Expansion in  extraaxial distance | Normal |
| Brain CT scan | Normal | Normal | Normal | Normal |
| Cardiac USG (ECHO) | Normal | Normal | Normal | Normal |
| Abdominl USG | Normal | Normal | Normal | Normal |
| Metabolic Screening | normal | Normal | Normal | Normal |
| EEG | Hypsarrhythmia | Hypsarrhythmia | Hypsarrhythmia | Burst- suppression patern |

Abbreviations are as follows: + , present; - , absent; NA, not available; OFC, occipito-frontal circumference; CT, computed tomography; MRI, magnetic resonance image; ERG, electroretinography; VEP, visual evoked potential; USG, ultrasound; ECHO, echocardiography; EEG, electroencephalography