|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Parameter | I:1 | II:1 | II:2 | III:1 | IV:1 |
| Perinatal stiffness | NR | NR | NR | Y | N |
| Age of onset | 40 | Childhood | Childhood | Childhood | Childhood |
| Treatment | NR | Phenobarbital | Phenobarbital | Phenobarbital first Clonazepam later | Clonazepam |
| Spontaneous remission periods | NR\* | Y | N | Y | N |
| Other movement disorders | NR | Hypnagogic myoclonus | Hypnagogic myoclonus | Hypnagogic myoclonus | Functional tremor |
| Current age/Age of death | 67 | 87 | 76 | 66 | 42 |

**Table e-1:** Summary of the clinical features in the first reported hyperekplexia family harboring the R271Q mutation in *GLRA,* remarkable is the long spontaneous remission in patients III:1 and II:1. Patients II:1 and II:2 suffered skull fractures after startle induced falls. The startle response was followed by short-lasting generalized stiffness in all the affected. \*Nevertheless the low frequency of falls (once or twice per year) raises a suspicion of probable remissions. NR: not reported, Y: yes. N: no.