|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Table e-3.** Clinical features in ISOD and MoCD class-1 and class-2 patient subgroups | | | | | | | | |
| Disease feature | Class-1 | | | | Class-2 | | | |
|  | ISOD (N=39) | | MoCD (N=82) | | ISOD (N=13) | | MoCD (N=12) | |
|  | N | % | N | % | N | % | N | % |
| Motor/Tone |  |  |  |  |  |  |  |  |
| Axial hypotonia | 21 | 54 | 34 | 41 | 6 | 46 | 7 | 58 |
| Appendicular hypertonia | 28 | 72 | 48 | 59 | 6 | 46 | 8 | 67 |
| Quadriparesis | 4 | 10 | 11 | 13 | 2 | 15 | 1 | 8 |
| Acute hemiparesis | 0 | 0 | 0 | 0 | 3 | 23 | 1 | 8 |
| Extrapyramidal |  |  |  |  |  |  |  |  |
| Opisthotonos | 8 | 21 | 10 | 12 | 2 | 15 | 0 | 0 |
| Choreoathetosis | 0 | 0 | 0 | 0 | 7 | 54 | 3 | 25 |
| Dystonia | 1 | 2 | 0 | 0 | 4 | 31 | 4 | 33 |
| Extrapyramidal NOS | 3 | 8 | 5 | 6 | 0 | 0 | 2 | 17 |
| Cerebellar |  |  |  |  |  |  |  |  |
| Ataxia | 0 | 0 | 0 | 0 | 5 | 38 | 2 | 17 |
| Nystagmus | 2 | 5 | 2 | 2 | 1 | 8 | 0 | 0 |
| Paroxysmal events |  |  |  |  |  |  |  |  |
| Seizures | 33 | 85 | 76 | 93 | 6 | 46 | 4 | 33 |
| Infantile spasms | 0 | 0 | 3 | 4 | 0 | 0 | 0 | 0 |
| Hyperekplexia | 3 | 8 | 7 | 9 | 0 | 0 | 0 | 0 |
| Ocular |  |  |  |  |  |  |  |  |
| Ectopic Lentis | 9 | 23 | 13 | 16 | 6 | 46 | 5 | 42 |
| Enophthalmos | 4 | 10 | 7 | 9 | 0 | 0 | 0 | 0 |
| NOS=Not otherwise specified. | | | | | | | | |