|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Table e-1.** Detailed *SMN1*:*SMN2* copy number configuration in controls and patients with PMA. | | | | | |
| ***SMN1*** | ***SMN2*** | **Controls (n=956)** | **PMA (n=150)** | **p** | **OR (95% CI)** |
| 1 | 1 | 8 (0.8%) | 1 (0.7%) | 1.00 | - |
|  | 2 | 10 (1.0%) | 2 (1.3%) | 0.67 | - |
|  | 3 | 1 (0.1%) | 1 (0.7%) | .25 | - |
|  | 4 | 2 (0.2%) | 0 (0.0%) | 1.00 | - |
|  |  |  |  |  |  |
| 2 | 0 | 71 (7.4%) | 8 (5.3%) | .49 | - |
|  | 1 | 372 (38.9%) | 67 (44.7%) | .21 | - |
|  | 2 | 424 (44.4%) | 51 (34.0%) | .022 | 0.65 (0.44 – 0.94) |
|  | 3 | 20 (2.0%) | 2 (1.3%) | .76 | - |
|  |  |  |  |  |  |
| 3 | 0 | 6 (0.6%) | 2 (1.3%) | .30 | - |
|  | 1 | 20 (2.0%) | 4 (2.7%) | .55 | - |
|  | 2 | 16 (1.7%) | 8 (5.3%) | .010 | 3.30 (1.20 -8.36) |
|  | 3 | 4 (0.4%) | 1 (1.3%) | .52 | - |
|  |  |  |  |  |  |
| 4 | 0 | 0 (0.0%) | 1 (0.7%) | 0.14 | - |
|  | 1 | 1 (0.1%) | 2 (1.3%) | 0.05 | - |
|  |  |  |  |  |  |
| 5 | 0 | 1 (0.1%) | 0 (0.0%) | 1.00 | - |
| CI = confidence interval, OR = odds ratio, PMA = progressive muscular atrophy, SMN = survival motor neuron | | | | | |