Supplementary Table 1 – Characteristics of patients

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **M** | **N** | **B** | **G** | **I** | **P** | **Q** | **A** | **C** | **D** | **J** | **O** | **H** | **K** | **L** | **E** | **F** |
| Sex | Male | Female | Male | Female | Male | Male | Male | Female | Male | Male | Female | Female | Female | Female | Male | Female | Male |
| SMA type | 3 | 3 | 3 | 3 | 3 | 3 | 3 | 3 | 3 | 3 | 3 | 3 | 3 | 2 | 2 | 2 | 2 |
| *SMN2* copy number | 4 | 3 | 3 | 4 | 3 | 4 | 4 | 3 | 4 | 3 | 3 | 4 | 3 | 3 | 3 | 3 | 3 |
| Age at symptom onset | N/A | 18mo | 4y | 3y | N/A | 30mo | 13.5y | 8y | 2y | 12mo | 2y | 18mo | 2y | 10mo | 2mo | 6mo | 3mo |
| Age at diagnosis (y) | 9 | 3 | 5 | 4 | N/A | 4 | 15 | 8 | 2 | 5 | 2 | 2 | 2 | 1.5 | 1 | 1 | 2 |
| Age at first dose (y) | 23 | 33 | 18 | 16 | 16 | 14 | 18 | 54 | 23 | 26 | 36 | 16 | 52 | 40 | 20 | 24 | 24 |
| Functional status | Walker | Walker | Walker | Walker | Walker | Walker | Walker | Sitter | Sitter | Sitter | Sitter | Sitter | Non-sitter | Non-sitter | Non-sitter | Non-sitter | Non-sitter |

Abbreviations: mo: month; N/A: not available; y: year.

Supplementary Table 2 - Individual variations of motor function outcomes between time points and baseline

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Patient ID** | **Functional Group** | **HFMSE** (pt) | **RULM** (pt) | **6MWT** (m) | **CHOP-ATEND** (pt)‡ |
| 12-mo | 24-mo | 36-mo | 12-mo | 24-mo | 36-mo | 12-mo | 24-mo | 36-mo | 12-mo | 24-mo | 36-mo |
| M | Walker | -1 | -1 | -1 | 0 | 0 | 0 | 40\* | 19 | -4 | N/A | N/A | N/A |
| N | Walker | -2 | 5\* | 3\* | -4\*\* | -1 | -2\*\* | 0 | 30\* | 31\* | N/A | N/A | N/A |
| B | Walker | 0 | -4\*\* | -5\*\* | N/A | N/A | N/A | -35\*\* | -20 | -28 | N/A | N/A | N/A |
| G | Walker | 1 | 1 | 1 | N/A | N/A | N/A | 86\* | 93\* | 113\* | N/A | N/A | N/A |
| I | Walker | N/A | N/A | N/A | N/A | N/A | N/A | -10 | 55\* | 143\* | N/A | N/A | N/A |
| P | Walker | 0 | 0 | 0 | 0 | 0 | 0 | 65\* | N/A | 109\* | N/A | N/A | N/A |
| Q | Walker | 2 | 1 | 0 | 0 | 0 | 0 | 51\* | N/A | 45\* | N/A | N/A | N/A |
| A | Sitter | -1 | -1 | N/A | 7\* | 6\* | 7\* | N/A | N/A | N/A | 0† | -1† | N/A |
| C | Sitter | 7\* | 9\* | 10\* | 4\* | 3\* | 3\* | N/A | N/A | N/A | 0 | 1 | N/A |
| D | Sitter | 2 | 2 | 2 | 1 | 1 | 1 | N/A | N/A | N/A | 4 | 4 | 9 |
| J | Sitter | 6\* | 8\* | 8\* | -2\*\* | -2\*\* | -6\*\* | N/A | N/A | N/A | 0† | 0† | N/A |
| O | Sitter | 4\*† | 3\*† | 3\*† | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| H | Non-sitter | 2 | 2 | N/A | 0 | 1 | 3\* | N/A | N/A | N/A | 3† | 3† | N/A |
| K | Non-sitter | 1 | 0 | N/A | 7\* | 7\* | 6\* | N/A | N/A | N/A | 0† | 1† | N/A |
| L | Non-sitter | 0 | N/A | N/A | 0 | 0 | 0 | N/A | N/A | N/A | 3 | 4 | N/A |
| E | Non-sitter | 0 | 0 | 0 | 2\* | 2\* | 1 | N/A | N/A | N/A | 1 | 1 | 3 |
| F | Non-sitter | 0 | 0 | 0 | 0 | 1 | 1 | N/A | N/A | N/A | -3 | -3 | -5 |

† For those patients, baseline data were missing and replaced by 6 months data.

‡ No MCID is available for the CHOP-ATEND.

\* Difference between time point and baseline above the MCID (HFMSE = ≥3; RULM ≥2; 6MWT ≥30).

\*\* Difference between time point and baseline below the MCID.

Abbreviations: 6MWT: 6 Minutes Walk Test; CHOP-ATEND: Children's Hospital of Philadelphia Adult Test of Neuromuscular Disorders; HFMSE: Hammersmith Functional Motor Scale Expanded for SMA; m: meter; mo: month; N/A: not available; pt: point; RULM: Revised Upper Limb Module.

Supplementary Table 3 - Individual variations of motor and pulmonary functions and patient-reported outcomes between time points and baseline

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **Patient ID** | **Functional Group** | **SMAFRS** (pt) | **FVC** (%) | **PCF** (L/min) |
| 12-mo | 24-mo | 36-mo | 12-mo | 24-mo | 36-mo | 12-mo | 24-mo | 36-mo |
| M | Walker | 0 | N/A | 3 | 2 | 2 | 3 | -17 | -21 | 38 |
| N | Walker | 1 | 1 | 2 | 10 | -1 | 5 | -35 | -6 | 11 |
| B | Walker | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| G | Walker | N/A | N/A | N/A | 22 | 2 | 6 | N/A | N/A | N/A |
| I | Walker | N/A | N/A | N/A | -8 | -15 | -5 | N/A | N/A | N/A |
| P | Walker | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| Q | Walker | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A | N/A |
| A | Sitter | 4 | 8 | 8 | -4 | -2 | -3 | 13 | -8 | -5 |
| C | Sitter | 0 | 0 | 0 | 7 | 10 | 11 | 30 | 22 | 45 |
| D | Sitter | 0 | 0 | 0 | -12 | -14 | -2 | -20 | -19 | 34 |
| J | Sitter | N/A | N/A | N/A | 9 | 13 | 12 | -2 | 22 | -21 |
| O | Sitter | N/A | N/A | N/A | N/A | N/A | -2 | N/A | N/A | N/A |
| H | Non-sitter | 1 | 1 | 1 | 5 | 2 | 8 | -40 | -22 | -26 |
| K | Non-sitter | 11 | 11 | 11 | 9 | 9 | 2 | 11 | 11 | 2 |
| L | Non-sitter | 0 | 0 | 1 | -1 | -1 | -2 | N/A | 56 | 14 |
| E | Non-sitter | 1 | 1 | 3 | 3 | 2 | 2 | 27 | 18 | N/A |
| F | Non-sitter | 0 | 0 | 0 | -5 | N/A | 2 | -33 | -86 | -12 |

Abbreviations: pt: points; FVC: forced vital capacity; mo: month; N/A: not available; PCF: peak cough flow; SMAFRS: Spinal Muscular Atrophy Functional Rating Scale.