

Supplementary Information

Table S1. Individual patient characteristics and outcomes for pediatric patients 1-14.

| Case number | Gender | SMA type | Number of SMN2 gene copies | Family history of SMA | CHOP-INTEND (baseline) | | CHOP-INTEND (most recent follow-up) | | Treatment response | Adverse events |
|-------------|--------|----------|----------------------------|-----------------------|-----------------------------------|-------|-------------------------------------|-------|-----------------------|--|
| | | | | | Months after treatment initiation | Score | Months after treatment initiation | Score | | |
| 1 | Female | I | 1 | (+) | 0 | 20 | 19 | 24 | Clinically meaningful | Chest infections, scoliosis, elevations of PT and aPPT |
| 2 | Male | II | 1 | (+) | 0 | 7 | 19 | 8 | Considerable | Chest infections, scoliosis |
| 3 | Female | II | 2 | (+) | 0 | 19 | 19 | 21 | Considerable | Chest infections, scoliosis |
| 4 | Female | I | 2 | (+) | 0 | 27 | 80 | 53 | Clinically meaningful | None |
| 5 | Female | I | 2 | (+) | 0 | 23 | 21 | 29 | Clinically meaningful | None |
| 6 | Male | I | 2 | (-) | 5 | 4 | 77 | 33 | Clinically meaningful | None |
| 7 | Male | I | 2 | (-) | 0 | 5 | 52 | 8 | Considerable | None |
| 8 | Female | I | 2 | (-) | 0 | 3 | 26 | 3 | Stabilization | None |
| 9 | Female | I | Unknown | (-) | 0 | 0 | 19 | 18 | Clinically meaningful | None |
| 10 | Male | II | Unknown | (-) | 0 | 12 | 78 | 43 | Clinically meaningful | None |
| 11 | Male | II | 2 | (-) | 0 | 36 | 33 | 48 | Clinically meaningful | None |
| 12 | Female | II | Unknown | (-) | 0 | 50 | 49 | 63 | Clinically meaningful | None |
| 13 | Male | II | Unknown | (-) | 0 | 22 | 58 | 52 | Clinically meaningful | None |
| 14 | Male | I | 2 | (-) | 0 | 18 | 23 | 34 | Clinically meaningful | Scoliosis |

aPPT, activated partial thromboplastin time. CHOP-INTEND, Children's Hospital of Philadelphia Infant Test of Neuromuscular Disorders. PT, prothrombin time. SMA, spinal muscular atrophy.

Table S2. Individual patient characteristics and outcomes for pediatric patients 15-20.

| Case number | Gender | SMA type | Number of SMN2 gene copies | Family history of SMA | HFMSE (baseline) | | HFMSE (most recent follow-up) | | Treatment response | Adverse events |
|-------------|--------|----------|----------------------------|-----------------------|-----------------------------------|-------|-----------------------------------|-------|-----------------------|---|
| | | | | | Months after treatment initiation | Score | Months after treatment initiation | Score | | |
| 15 | Male | II | 2 | (-) | 0 | 4 | 51 | 4 | Stabilization | Scoliosis |
| 16 | Female | III | Unknown | (+) | 0 | 54 | 20 | 57 | Clinically meaningful | Scoliosis |
| 17 | Male | II | 2 | (+) | 5 | 44 | 54 | 46 | Considerable | Scoliosis |
| 18 | Male | III | 3 | (+) | 4 | 60 | 43 | 65 | Clinically meaningful | Scoliosis |
| 19 | Female | III | Unknown | (+) | 0 | 52 | 20 | 57 | Clinically meaningful | Scoliosis, headache |
| 20 | Male | III | 3 | (+) | 5 | 48 | 54 | 54 | Clinically meaningful | Scoliosis, lumbar region fibrous tissue formation |

HFMSE, Hammersmith Functional Motor Scale – Expanded. SMA, spinal muscular atrophy.

Table S3. Individual patient characteristics and outcomes for adult patients 21-38.

| Case number | Gender | SMA type | Number of SMN2 gene copies | Family history of SMA | Parental consanguinity | RULM (baseline) | | RULM (most recent follow-up) | | Treatment response | Adverse events |
|-------------|--------|----------|----------------------------|-----------------------|------------------------|-----------------------------------|-------|-----------------------------------|-------|-----------------------|--------------------------|
| | | | | | | Months after treatment initiation | Score | Months after treatment initiation | Score | | |
| 21 | Male | III | 3 | (+) | (+) | 6 | 23 | 22 | 32 | Clinically meaningful | None |
| 22 | Male | III | Unknown | (+) | (+) | 13 | 19 | 34 | 25 | Clinically meaningful | None |
| 23 | Male | III | 4 | (+) | (+) | 0 | 27 | 22 | 32 | Clinically meaningful | None |
| 24 | Male | III | 2 | (+) | (+) | 0 | 16 | 30 | 20 | Clinically meaningful | Restrictive lung disease |
| 25 | Female | III | 3 | (+) | (+) | 3 | 36 | 10 | 32 | Motor decline | None |
| 26 | Female | III | 4 | (-) | (+) | 0 | 34 | 13 | 36 | Clinically meaningful | None |
| 27 | Male | III | Unknown | (-) | (+) | 14 | 24 | 38 | 34 | Clinically meaningful | None |
| 28 | Female | III | Unknown | (+) | (+) | 0 | 18 | 18 | 19 | Considerable | None |
| 29 | Male | III | 2 | Unknown | Unknown | 4 | 37 | 36 | 37 | Stabilization | None |
| 30 | Male | III | Unknown | (+) | (+) | 0 | 33 | 18 | 35 | Clinically meaningful | None |
| 31 | Male | III | 3 | (-) | (+) | 0 | 36 | 52 | 37 | Considerable | None |
| 32 | Male | III | 3 | (+) | (+) | 0 | 32 | 50 | 37 | Clinically meaningful | None |
| 33 | Male | III | Unknown | (+) | (+) | 0 | 16 | 14 | 23 | Clinically meaningful | None |
| 34 | Male | III | 2 | (+) | (+) | 4 | 25 | 47 | 28 | Clinically meaningful | None |
| 35 | Male | III | 3 | (+) | (+) | 0 | 19 | 34 | 27 | Clinically meaningful | None |
| 36 | Male | II | 3 | (+) | (+) | 0 | 27 | 35 | 30 | Clinically meaningful | None |
| 37 | Female | III | 3 | (+) | (+) | 0 | 19 | 37 | 20 | Considerable | None |
| 38 | Female | II | 3 | (-) | (+) | 6 | 33 | 44 | 37 | Clinically meaningful | None |

RULM, Revised Upper Limb Module. SMA, spinal muscular atrophy.