

**Supplementary Table S1.** Details of number of assessments/patients included in the *Exon skipping class* analysis. *Key to table: Only completed data were used for the analysis. Patients with missing data at baseline or 24 months were excluded from the longitudinal analysis.*

|                                 | Number of patients | Number of paired assessments (baseline-24 months) |
|---------------------------------|--------------------|---|
| <b>Deletions</b>                | <b>215</b>         | <b>553</b>  |
| <b>Duplications</b>             | <b>26</b>          | <b>60</b>   |
| <b>Small mutations</b>          | <b>70</b>          | <b>195</b>  |
| <b>Amenable to skip exon 44</b> | <b>27</b>          | <b>66</b>   |
| <b>Amenable to skip exon 45</b> | <b>25</b>          | <b>59</b>   |
| <b>Amenable to skip exon 51</b> | <b>34</b>          | <b>103</b>  |
| <b>Amenable to skip exon 43</b> | <b>24</b>          | <b>76</b>   |

**Supplementary Table S2.** 12 and 24 months PUL changes subdivided in exon skip classes and ambulatory status. \*= N represent the number of 24-months paired visits.

|                  |                         | AMBULANT          |                   |                   |                      | NON AMBULANT        |                     |                     |                     |
|------------------|-------------------------|-------------------|-------------------|-------------------|----------------------|---------------------|---------------------|---------------------|---------------------|
|                  |                         | 44 (N=48*)        | 45 (N=42*)        | 51 (N=48*)        | 53 (N=30*)           | 44 (N=18*)          | 45 (N=17*)          | 51 (N=55*)          | 53 (N=46*)          |
| BASELINE         | <b>Age</b>              |                   |                   |                   |                      |                     |                     |                     |                     |
|                  | Mean (SD)               | 9.60 (4.30)       | 8.22 (2.18)       | 8.58 (2.29)       | 8.64 (3.72)          | 19.1 (5.19)         | 15.4 (4.14)         | 15.9 (4.06)         | 17.0 (4.64)         |
|                  | Median [Min, Max]       | 8.53 [3.81, 22.1] | 8.35 [3.80, 13.2] | 8.36 [4.59, 13.6] | 8.19 [4.12, 21.7]    | 20.3 [9.99, 26.5]   | 14.2 [10.0, 25.3]   | 15.7 [7.87, 25.7]   | 17.1 [8.83, 24.8]   |
|                  | <b>TRANSITIONING</b>    |                   |                   |                   |                      |                     |                     |                     |                     |
|                  | NO                      | 44 (91.7%)        | 37 (88.1%)        | 31 (64.6%)        | 18 (60.0%)           | N/A                 | N/A                 | N/A                 | N/A                 |
|                  | YES                     | 4 (8.3%)          | 5 (11.9%)         | 17 (35.4%)        | 12 (40.0%)           | N/A                 | N/A                 | N/A                 | N/A                 |
|                  | <b>TOTAL PUL 2.0</b>    |                   |                   |                   |                      |                     |                     |                     |                     |
|                  | Mean (SD)               | 40.1 (1.51)       | 38.5 (3.17)       | 36.7 (4.51)       | 36.8 (5.01)          | 23.6 (8.43)         | 19.2 (10.3)         | 18.3 (9.25)         | 24.6 (9.83)         |
|                  | Median [Min, Max]       | 40.0 [37.0, 42.0] | 39.5 [29.0, 42.0] | 37.5 [23.0, 42.0] | 37.5 [24.0, 42.0]    | 22.5 [8.00, 38.0]   | 17.0 [3.00, 34.0]   | 16.0 [3.00, 39.0]   | 22.0 [10.0, 40.0]   |
| 12 MONTH changes | <b>TOTAL PUL 2.0</b>    |                   |                   |                   |                      |                     |                     |                     |                     |
|                  | Mean (SD)               | 0.104 (1.97)      | -0.381 (4.06)     | -0.458 (3.32)     | 0.0167 (3.18)        | -2.75 (3.61)        | -3.06 (3.12)        | -2.33 (3.20)        | -3.23 (4.06)        |
|                  | Median [Min, Max]       | 0 [-7.00, 5.00]   | 0 [-17.0, 11.0]   | 0 [-8.00, 8.00]   | -0.250 [-10.0, 8.00] | -1.50 [-12.0, 3.00] | -2.00 [-10.0, 2.00] | -2.00 [-13.0, 3.00] | -3.00 [-19.0, 3.00] |
|                  | <b>SHOULDER PUL 2.0</b> |                   |                   |                   |                      |                     |                     |                     |                     |
|                  | Mean (SD)               | 0.0208 (1.35)     | -0.214 (2.09)     | -0.521 (2.02)     | -0.200 (2.14)        | -1.11 (1.74)        | -1.29 (2.54)        | -0.718 (1.67)       | -1.39 (2.07)        |
|                  | Median [Min, Max]       | 0 [-4.00, 3.00]   | 0 [-7.00, 6.00]   | 0 [-5.00, 4.00]   | 0 [-4.00, 6.00]      | 0 [-5.00, 0]        | 0 [-8.00, 0]        | 0 [-6.00, 3.00]     | 0 [-9.00, 0]        |
|                  | <b>ELBOW PUL 2.0</b>    |                   |                   |                   |                      |                     |                     |                     |                     |
|                  | Mean (SD)               | -0.0104 (0.789)   | -0.274 (1.68)     | -0.125 (1.67)     | 0.0667 (0.971)       | -1.42 (1.70)        | -1.03 (1.89)        | -1.24 (1.84)        | -1.63 (2.37)        |

|                     |                         |                    |                         |                        |                         |                        |                         |                        |                        |
|---------------------|-------------------------|--------------------|-------------------------|------------------------|-------------------------|------------------------|-------------------------|------------------------|------------------------|
| 24 MONTH<br>changes | Median [Min,<br>Max]    | 0 [-4.00,<br>1.00] | 0 [-8.00, 4.00]         | 0 [-5.00,<br>5.00]     | 0 [-2.00, 3.00]         | -1.00 [-6.00,<br>1.00] | -0.500 [-5.00,<br>2.00] | -1.00 [-7.00,<br>2.00] | -1.00 [-10.0,<br>2.00] |
|                     | <b>DISTAL PUL 2.0</b>   |                    |                         |                        |                         |                        |                         |                        |                        |
|                     | Mean (SD)               | 0.0938<br>(0.607)  | 0.107 (1.15)            | 0.188 (1.01)           | 0.150 (1.29)            | -0.222 (1.23)          | -0.735 (1.09)           | -0.373 (1.07)          | -0.207<br>(0.873)      |
|                     | Median [Min,<br>Max]    | 0 [-1.00,<br>1.00] | 0 [-2.00, 5.00]         | 0 [-2.00,<br>3.00]     | 0 [-4.00, 3.00]         | 0 [-4.00,<br>2.00]     | -0.500 [-3.00,<br>1.00] | 0 [-3.00,<br>2.00]     | 0 [-3.00,<br>2.00]     |
|                     | <b>TOTAL PUL 2.0</b>    |                    |                         |                        |                         |                        |                         |                        |                        |
|                     | Mean (SD)               | -0.229<br>(2.58)   | -1.71 (5.93)            | -1.31 (4.82)           | -1.67 (5.89)            | -4.78 (4.35)           | -5.41 (4.91)            | -4.38 (3.81)           | -6.39 (5.59)           |
|                     | Median [Min,<br>Max]    | 0 [-8.00,<br>5.00] | 0 [-18.0, 11.0]         | -1.00 [-12.0,<br>8.00] | -1.00 [-25.0,<br>11.0]  | -3.50 [-13.0,<br>2.00] | -4.00 [-15.0,<br>1.00]  | -4.00 [-16.0,<br>1.00] | -6.00 [-21.0,<br>1.00] |
|                     | <b>SHOULDER PUL 2.0</b> |                    |                         |                        |                         |                        |                         |                        |                        |
|                     | Mean (SD)               | -0.229<br>(1.49)   | -1.26 (3.25)            | -1.13 (2.83)           | -1.27 (2.68)            | -1.78 (2.65)           | -2.35 (3.77)            | -1.29 (2.39)           | -2.57 (3.29)           |
|                     | Median [Min,<br>Max]    | 0 [-4.00,<br>3.00] | -0.500 [-9.00,<br>6.00] | 0 [-7.00,<br>5.00]     | -0.500 [-9.00,<br>4.00] | 0 [-8.00, 0]           | 0 [-9.00, 0]            | 0 [-9.00, 0]           | 0 [-11.0, 0]           |
| 24 MONTH<br>changes | <b>ELBOW PUL 2.0</b>    |                    |                         |                        |                         |                        |                         |                        |                        |
|                     | Mean (SD)               | -0.146<br>(1.25)   | -0.571 (2.37)           | -0.417 (2.10)          | -0.333 (2.66)           | -2.44 (1.79)           | -1.82 (2.21)            | -2.33 (2.26)           | -3.33 (3.00)           |
|                     | Median [Min,<br>Max]    | 0 [-5.00,<br>1.00] | 0 [-8.00, 4.00]         | 0 [-5.00,<br>4.00]     | 0 [-10.0, 6.00]         | -2.50 [-6.00,<br>0]    | -2.00 [-6.00,<br>2.00]  | -2.00 [-8.00,<br>1.00] | -3.00 [-12.0,<br>1.00] |
|                     | <b>DISTAL PUL 2.0</b>   |                    |                         |                        |                         |                        |                         |                        |                        |
|                     | Mean (SD)               | 0.146<br>(0.743)   | 0.119 (1.25)            | 0.229 (1.31)           | -0.0667 (1.91)          | -0.556 (1.69)          | -1.24 (1.48)            | -0.764 (1.35)          | -0.500 (1.19)          |
|                     | Median [Min,<br>Max]    | 0 [-1.00,<br>2.00] | 0 [-3.00, 5.00]         | 0 [-2.00,<br>4.00]     | 0 [-6.00, 6.00]         | 0 [-6.00,<br>2.00]     | -1.00 [-4.00,<br>1.00]  | -1.00 [-4.00,<br>3.00] | -1.00 [-4.00,<br>3.00] |