**Supplementary Table 1. Overview on missing data**

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **ID** | **Neurological examination, HHD\*** | **MFM-20/32** | **HFMS** | **PBS/**  **miniBEST** | **GTFT** | **Respiratory function (**Spirometry, MIP/MEP/SNIP, US diaphragm) | **Cardiac assessment** (ECG, LVEF, GLS) | **Bone quality (L, F)** | **Questionnaires** |
| 1 | + | + | + | - (non-ambulant) | + (except for 30 seconds sit to stand, descend 4 stairs, and raise from the floor; 10MWT and 6MWT with walking aid) | + | +, but no GLS because technically not possible | F (L not possible due to scoliosis surgery osteosynthesis material) | PedsQL, ACTIVLIM, Wong-Baker Faces  Pain Rating Scale, McGill pain questionnaire, EK2 |
| 2 | + | + | - (ambulant) | + | + (except for 30 seconds sit to stand) | + | + | F (L not possible due to scoliosis surgery osteosynthesis material) | RAND36, INQoL, ACTIVLIM, IPA, CIS, Wong-Baker Faces Pain Scale, McGill pain questionnaire, EK2 |
| 3 | - (too young to perform these tasks) | + | - (ambulant) | + | + (except for TUG with cognitive task) | - (too young to perform these tasks) | +, but no GLS because technically not possible | L (no normal values for F available for this age) | PedsQL (only parent-proxy), Wong-Baker Faces Pain Scale |
| 4 | + | + | - (ambulant) | + | + | + | + | + | RAND36, INQoL, ACTIVLIM, IPA, CIS, Wong-Baker Faces Pain Scale, McGill pain questionnaire, EK2 |
| 5 | + | + | - (ambulant) | + | + | + | + | + | PedsQL ACTIVLIM, Wong-Baker Faces Pain Scale |
| 6 | + | + | - (ambulant) | + | + (except for 30 seconds sit to stand) | + | + | F (L not possible due to scoliosis surgery osteosynthesis material) | RAND36, INQoL, ACTIVLIM, IPA, CIS, Wong-Baker Faces Pain Scale, McGill pain questionnaire, EK2 |
| 7 | + | + | + | - (non-ambulant) | - | + | + | F (L not possible due to scoliosis surgery osteosynthesis material) | RAND36, INQoL, ACTIVLIM, IPA, CIS, Wong-Baker Faces Pain Scale, McGill pain questionnaire, EK2 |
| 8 | + | + | - (ambulant) | + | + | + | + | + | PedsQL ACTIVLIM, Wong-Baker Faces Pain Scale |
| 9 | + | + | - (ambulant) | + | + (except for 30 seconds sit to stand) | + | + | + | PedsQL, ACTIVLIM, . Wong-Baker Faces  Pain Rating Scale, McGill pain questionnaire, EK2 |
| 10 | + | + | - (ambulant) | + | + | + | - (technical issues of ultrasound machine) | + | PedsQL ACTIVLIM, Wong-Baker Faces Pain Scale |
| 11 | + | + | + | - (physical limitations) | + (except for 30 seconds sit to stand, climb and descend 4 stairs, raise from the floor, 10MWT and 6MWT) | Only MIP/ MEP/ SNIP because of home visit | - (home visit) | - (home visit) | RAND36, INQoL, ACTIVLIM, IPA, CIS, Wong-Baker Faces Pain Scale, McGill pain questionnaire, EK2 |

\* HHD was only performed in individual muscles that have a MRC score of 4 or higher.

HHD = Hand-Held Dynamometry; MFM-20/32 = Motor Function Measure 20/32; HFMS = Hammersmith Functional Motor Scale; PBS = Pediatric Balance Scale; MiniBEST = Mini Balance Evaluation Systems Test; GTFT = graded and timed function tests; 6MWT = 6-Minute Walk Test; TUG = Timed Up and Go-test; MIP = Maximum Inspiratory Pressure; MEP = Maximum Expiratory Pressure; SNIP = Sniff Nasal Inspiratory Pressure; US diaphragm = ultrasound of the diaphragm; LVEF = left ventricular ejection fraction; GLS = global longitudinal strain; L = lumbar bone mineral density; F = femoral bone mineral density.

**Supplementary Table 2. Overview on questionnaires in SELENON-RM patients**

|  |  |  |  |
| --- | --- | --- | --- |
| **Questionnaire** | **N** | **Mean±SD (range)** | **Cut – off or range** |
| RAND36(36) |  |  |  |
| * Physical functioning (%) | 5 | 15±11.7 (5 – 30) | 81.9±23.2 |
| * Role limitations due to physical health (%) | 5 | 25±35.4 (0 to 75) | 79.4±35.5 |
| * Role limitation due to emotional problems (%) | 5 | 40±54.8 (0 to 100) | 84.1±32.3 |
| * Energy/fatigue (%) | 5 | 52±10.4 (40 to 65) | 67.4±19.9 |
| * Emotional well-being (%) | 5 | 80±13.0 (60 to 96) | 76.8±18.4 |
| * Social functioning (%) | 5 | 82.5±25.9 (37.5 to 100) | 86.9±20.5 |
| * Pain (%) | 5 | 72.2±22.0 (47.5 to 100) | 79.5±25.6 |
| * General health (%) | 5 | 38±16.4 (20 to 55) | 72.7±22.7 |
| INQoL |  |  | Range |
| * Weakness (%) | 5 | 74.7±8.6 (68.4 to 89.5) | 0 – 100% |
| * Muscle locking (%) | 5 | 4.2±9.4 (0 to 21.1) | 0 – 100% |
| * Pain (%) | 5 | 36.8±35.3 (0 to 84.2) | 0 – 100% |
| * Fatigue (%) | 5 | 51.6±20.5 (36.8 to 78.9) | 0 – 100% |
| * Activities (%) | 5 | 55.9±19.0 (28.7 to 73.1) | 0 – 100% |
| * Independence (%) | 5 | 37.8±27.9 (5.6 to 72.2) | 0 – 100% |
| * Social relationships (%) | 5 | 14.6±14.4 (0 to 32.4) | 0 – 100% |
| * Emotions (%) | 5 | 29.4±15.3 (8.3 to 50) | 0 – 100% |
| * Body image (%) | 5 | 57.2±20.1 (30.6 to 77.8) | 0 – 100% |
| * QoL score (%) | 5 | 42.8±11.4 (30.6 to 59.4) | 0 – 100% |
| CIS(31) |  |  |  |
| * Subjective fatigue | 5 | 34.6±3.05 (31 to 38) | Increased if ≥ 27  Severely increased if ≥ 35 |
| * Total | 5 | 80.2±3.4 (76 to 85) | ≥ 76 |
| PedsQL Generic Core Scale(37) |  |  |  |
| * Self-report | 4 | 55.7±15.1 (40.2 to 73.9) | 82.87±13.16 |
| * Proxy-parent mother | 5 | 52.4±8.4 (43.5 to 62.0) | 81.34±15.92 |
| * Proxy-parent father | 5 | 52.9±10.0 (41.3 to 70.7) | 81.34±15.92 |
| PedsQL Neuromuscular Module(25) |  |  | In SMA |
| * Self-report | 4 | 61.8±11.4 (48.0 to 74.0) | 59.74±16.75 |
| * Proxy-parent mother | 5 | 57.7±10.1 (43.0 to 70.0) | 67.54±15.57 |
| * Proxy-parent father | 5 | 53.8±13.8 (29.0 to 68.0) | 67.54±15.57 |
| PedsQL Mutidimensional Fatigue Scale(38) |  |  |  |
| * Self-report | 4 | 53.6±21.7 (29.2 to 77.8) | 80.49±13.33 |
| * Proxy-parent mother | 5 | 59.5±15.1 (34.7 to 79.2) | 89.63±11.38 |
| * Proxy-parent father | 5 | 60.6±13.3 (38.9 to 79.2) | 89.63±11.38 |
| ACTIVLIM total score (median) | 10 | 28.5 [19 – 31] | Total score: 36 |
| * Activities impossible to perform |  | 2.6±3.5 (0 to 10) | 0 |
| * Activities difficult to perform |  | 5.4±3.8 (1 to 14) | 0 |
| * Activities easy to perform |  | 9.9±5.0 (2 to 17) | 18 |
| IPA (median [IQR]) |  |  | Median |
| * Autonomy indoors | 5 | 0 [0 – 0] | 0 [0 – 0.1] |
| * Family role | 5 | 1 [0 – 1] | 0.1 [0 – 0.4] |
| * Autonomy outdoors | 5 | 2 [2 – 2] | 0.2 [0 – 0.7] |
| * Social life | 5 | 0 [0 – 0.5] | 0.0 [ 0 – 0.3] |
| * Work and education | 5 | 1 [1 – 1] | 0.2 [0 – 3.4] |

INQoL = Individualized Quality of Life questionnaire; CIS = Checklist Individual Strength; PedsQL = Pediatric Quality of Life Inventory; IPA = Impact on Participation and Autonomy; IQR = interquartile range.