|  |  |  |  |
| --- | --- | --- | --- |
|  | SMA + SPG4 | SMA + Noonan syndrome | SMA + CMT1A |
| *SMA type* | 3b | 3b | 3a |
| *SMN2 copies* | 3 | 3 | 3 |
| *Current age of the patient (years)* | 17 | 20 | 20 |
| *Clinical features* | | | |
| Upper limbs weakness | Proximal | Proximal | Proximal > Distal |
| Upper limbs wasting | Proximal | Proximal | Proximal > Distal |
| Lower limb weakness | Proximal > distal | Proximal > distal | Proximal = distal |
| Lower limbs wasting | Proximal = distal | Proximal > distal | Proximal = distal |
| Knee reflex | Absent | Absent | Absent |
| Ankle reflex | Present | Decreased | Absent |
| Foot deformities | Pes cavus | - | - |
| Scoliosis | Mild | Mild | Severe  (Surgery) |
| *6MWT (meters)* | 365 | 273 | Loss of ambulation |
| *HFMSE* | 56 | 48 | 8 |
| *RULM* | 35 | 36 | Not assessed |
| *MFM* | 70 | 75 | 48 |

**Table 2.** Summary of the patients’ clinical details

SMA — spinal muscular atrophy, SPG4 — spastic paraplegia 4, CMT1A — Charcot-Marie Tooth type 1A disease, 6MWT — Six Minutes Walking Test, HFMSE — Hammersmith Functional Motor Scale – Expanded, RULM — Revised Upper Limb Module, MFM — Motor Function Measure