|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | | | | | | | | | **SMA type 2** | | **SMA type 3** | | **Overall (n=81)** |
|  | | | | | | | | | **Non-sitter (n=19)** | **Sitter (n=34)** | **Non-Ambulant (n=9)** | **Ambulant (n=19)** |
| **Site (n)** | | | | | | | | |  |  |  |  |  |
|  | | | | | | | | *Paris* | *2* | *10* | *4* | *3* | *19* |
|  | | | | | | | | *Liège* | *5* | *5* | *0* | *4* | *14* |
|  | | | | | | | | *Nantes* | *5* | *4* | *0* | *1* | *10* |
|  | | | | | | | | *Toulouse* | *2* | *5* | *1* | *2* | *10* |
|  | | | | | | | | *Lyon* | *0* | *3* | *1* | *3* | *7* |
|  | | | | | | | | *Lille* | *3* | *2* | *1* | *0* | *6* |
|  | | | | | | | | *Leuven* | *1* | *2* | *1* | *2* | *6* |
|  | | | | | | | | *Strasbourg* | *0* | *2* | *0* | *3* | *5* |
|  | | | | | | | | *Essen* | *1* | *1* | *1* | *1* | *4* |
| **Male (n)** | | | | | | | | | *7* | *14* | *6* | *10* | *37* |
| **Other family member affected (%)** | | | | | | | | | *1* | *3* | *2* | *4* | *10* |
| **Medical history (perinatal period)** | | | | | | | | |  |  |  |  |  |
|  | | | *Birth (week of amenorrhea; n=77)* | | | | | | *40 (38-40)* | *39 (39-41)* | *40 (38-41)* | *40 (39-40)* | *40 (39-40)* |
|  | | | *Birth weight (g; n=80) \** | | | | | | *3040* a  *(2560-3290)* | *3445* b, c  *(3200-3894)* | *3250* a, c  *(2050-3460)* | *3360* a, c  *(2960-3676)* | *3300*  *(3021-3635)* |
|  | | | *Birth length (cm; n=77)\** | | | | | | *48* a *(48-49)* | *50* b, c *(49-52)* | *49* a, c *(47-53)* | *50* a, c *(48-52)* | *48 (48-51)* |
|  | | | *Head circumference (cm; n=65)* | | | | | | *34 (33-35)* | *35 (34-36)* | *35 (33-36)* | *34 (32-35)* | *33 (33-36)* |
| **Medical history (events total number)** | | | | | | | | |  |  |  |  |  |
|  | | | | | *Eye/Ear/Nose/Throat* | | | | *1 (0-2)* | *0 (0-2)* | *0 (0-2)* | *0 (0-1)* | *0 (0-2)* |
|  | | | | | *Neurologic and Psychiatric* | | | | *0 (0-1)* | *0 (0-1)* | *0 (0-1)* | *1 (0-2)* | *0 (0-1)* |
|  | | | | | *Pulmonary \*\** | | | | *3* a *(1-5)* | *1* a, b *(0-3)* | *1* a, b *(0-2)* | *0* b, c *(0-1)* | *1 (0-3)* |
|  | | | | | *Cardiovascular and Blood* | | | | *0 (0-0)* | *0 (0-0)* | *0 (-)* | *0 (0-0)* | *0 (0-0)* |
|  | | | | | *Musculoskeletal \*\** | | | | *3* a *(2-3)* | *1* b *(0-3)* | *1* a, b *(0-2)* | *1* b *(1-2)* | *1 (1-3)* |
|  | | | | | *Hepatic-Gastrointestinal* | | | | *0 (0-0)* | *0 (0-0)* | *0 (0-0)* | *0 (0-0)* | *0 (0-0)* |
|  | | | | | *Metabolic* | | | | *0 (0-0)* | *0 (0-0)* | *0 (0-0)* | *0 (0-0)* | *0 (0-0)* |
|  | | | | | *Genitourinary \** | | | | *0 (0-1)* | *0 (0-0)* | *0 (0-1)* | *0 (0-0)* | *0 (0-1)* |
|  | | | | | *Skin* | | | | *0 (0-1)* | *0 (0-0)* | *0 (0-1)* | *0 (0-1)* | *0 (0-1)* |
| **Heart rate (bpm; n=79) \*\*** | | | | | | | | | *99* a, b *(90-115)* | *115* a *(99-122)* | *79* c *(75-84)* | *93* b, c *(72-115)* | *100 (84-117) Δ* |
| **Systolic blood pressure (mmHg; n=78)** | | | | | | | | | *108 (96-124)* | *103 (95-112)* | *111 (107-127)* | *106 (98-115)* | *106 (98-115) Δ* |
| **Diastolic blood pressure (mmHg; n=78)** | | | | | | | | | *69 (58-85)* | *67 (60-73)* | *70 (66-72)* | *62 (57-69)* | *66 (60-72) Δ* |
| **Organs and systems (abnormal, n)** | | | | | | | | |  |  |  |  |  |
|  | | | | *Eye/Ear/Nose/Throat* | | | | | *2* | *3* | *1* | *0* | *6* |
|  | | | | *Neurologic and Psychiatric* | | | | | *14* | *27* | *5* | *18* | *64* |
|  | | | | *Pulmonary \** □ | | | | | *5* | *3* | *0* | *0* | *8* |
|  | | | | *Cardiovascular and Blood* | | | | | *0* | *1* | *0* | *1* | *2* |
|  | | | | *Musculoskeletal* | | | | | *18* | *33* | *9* | *19* | *79* |
|  | | | | *Hepatic-Gastrointestinal* | | | | | *2* | *3* | *0* | *0* | *5* |
|  | | | | *Metabolic \** □ | | | | | *4* a | *0* b | *0* a, b | *0* a, b | *4* |
|  | | | | *Genitourinary* | | | | | *2* | *2* | *0* | *0* | *4* |
|  | | | | *Skin* | | | | | *6* | *3* | *1* | *3* | *13* |
| **Ventilation details (n)** | | | | | | | | |  |  |  |  |  |
|  |  | | | | | Non Invasive Ventilation | | |  |  |  |  |  |
|  |  | | | | | *CPAP* | | | *4* | *3* | *0* | *0* | *7* |
|  |  | | | | | *BPAP* | | | *10* | *10* | *0* | *0* | *20* |
|  |  | | | | | *IPV* | | | *5* | *5* | *0* | *0* | *10* |
|  |  | | | | | *NPV* | | | *1* | *0* | *0* | *0* | *1* |
|  |  | | | | | *Sip and puff* | | | *1* | *0* | *0* | *0* | *1* |
|  |  | | | | | *IPPV* | | | *8* | *11* | *1* | *1* | *21* |
|  |  | | | | | *Cough assist* | | | *8* | *8* | *0* | *0* | *16* |
|  |  | | | | | Invasive Ventilation | | |  |  |  |  |  |
|  |  | | | | | *Tracheostomy* | | | *0* | *1* | *0* | *0* | *1* |
| **Feeding difficulties details (n)** | | | | | | | | |  |  |  |  |  |
|  | | | | | | | *Sucking* | | *2* | *0* | *0* | *0* | *2* |
|  | | | | | | | *Swallowing* | | *8* | *6* | *1* | *0* | *15* |
|  | | | | | | | *Chewing* | | *10* | *7* | *0* | *0* | *17* |
|  | | | | | | | *Reflux/vomiting* | | *2* | *5* | *0* | *0* | *7* |
| **Assistive devices details (n)** | | | | | | | | |  |  |  |  |  |
|  | | *AFO, Hand & ankle splint \** □ | | | | | | | *18* a | *27* a, b | *8* a, b | *6* b | *59* |
|  | | *Power wheelchair \*\** □ | | | | | | | *17* a | *25* a | *7* a | *2* b | *51* |
|  | | *Manual wheelchair \*\** □ | | | | | | | *11* a, b | *24* b | *9* b | *3* a | *47* |
|  | | *Stander \*\** □ | | | | | | | *14* a | *25* a | *3* a, b | *1* b | *43* |
|  | | *Corset, lumbar belt \*\** □ | | | | | | | *19* a | *18* b | *2* b, c | *1* c | *40* |
|  | | *Walker, cane \*\** □ | | | | | | | *0* a | *7* a, b | *6* b | *2* a, b | *15* |
|  | | *Medicalized bed, positioning mattress* | | | | | | | *5* | *3* | *3* | *0* | *11* |
|  | | *Orthopaedic soles/shoes, heel cup \** □ | | | | | | | *1* a, b | *1* b | *2* a, b | *5* a | *9* |
|  | | *Communication device* | | | | | | | *1* | *1* | *0* | *2* | *4* |
| **Absence of cognitive impairment (n)** | | | | | | | | | *19* | *34* | *9* | *19* | *81* |
| **School attendance (6-18 years old; n)** | | | | | | | | | *n=11* | *n=8* | *n=2* | *n=5* | *n=26* |
|  | | | | | | | | | *11* | *8* | *2* | *5* | *26* |
| **School level (18-30 years old, n)** | | | | | | | | | *n=4* | *n=1* | *n=7* | *n=5* | *n=17* |
|  | | | | | | | | *High school* | *2* | *0* | *3* | *1* | *6* |
|  | | | | | | | | *College* | *1* | *1* | *3* | *0* | *5* |
|  | | | | | | | | *Post Graduate* | *1* | *0* | *1* | *4* | *6* |
| **Employment status (18-30 years old; n)** | | | | | | | | | *n=4* | *n=1* | *n=6* | *n=5* | *n=16* |
|  | | | | | | | | *Full time* | *1* | *0* | *0* | *1* | *12* |
|  | | | | | | | | *Part time* | *0* | *0* | *1* | *0* | *6* |
|  | | | | | | | | *Student* | *2* | *1* | *3* | *3* | *53* |
|  | | | | | | | | *Unemployed* | *1* | *0* | *2* | *1* | *24* |