

## **Supplementary Material**

### **Management of Duchenne Muscular Dystrophy in clinical practice: A survey-based study in Spain**

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## Supplementary Material

**Table 1** Most common type of single point mutation

| N=41                                             |           |
|--------------------------------------------------|-----------|
| Most common type of single point mutation, n (%) |           |
| Nonsense                                         | 21 (51.2) |
| Missense                                         | 12 (29.3) |
| Not available                                    | 7 (17.1)  |
| Other                                            | 1 (2.4)   |

**Table 2** DMD diagnostic tests commonly performed and their availability at participants' hospitals

| N=41                                                                         |            |
|------------------------------------------------------------------------------|------------|
| Assessment of creatine kinase levels, n (%)                                  | 41 (100.0) |
| DMD diagnostic tests commonly performed, n (%)                               |            |
| Multiplex Ligation-dependent Probe Amplification                             | 38 (92.7)  |
| Performed at the participant's hospital                                      | 23 (60.5)  |
| Sequencing of the dystrophin gene                                            | 33 (80.5)  |
| Performed at the participant's hospital                                      | 14 (42.4)  |
| Clinical exome or genetic panel for single point mutations/microduplications | 31 (75.6)  |
| Performed at the participant's hospital                                      | 19 (61.3)  |
| DNA arrays                                                                   | 15 (36.6)  |
| Performed at the participant's hospital                                      | 13 (86.7)  |
| Muscle biopsy                                                                | 14 (34.1)  |
| Performed at the participant's hospital                                      | 10 (71.4)  |

Abbreviations: DMD, Duchenne muscular dystrophy.

**Table 3** Pediatric neurologists' estimates of the age at which patients with DMD developed loss of ambulation

|                 | N=41      |
|-----------------|-----------|
| Mean age, n (%) |           |
| <8 years        | 2 (4.9)   |
| 8-10 years      | 5 (12.2)  |
| 11-13 years     | 22 (53.7) |
| 14-16 years     | 12 (29.3) |
| 17-19 years     | 0 (0.0)   |
| >19 years       | 0 (0.0)   |

Abbreviations: DMD, Duchenne muscular dystrophy.

**Table 4** Age at which patients with DMD developed scoliosis, cardiac involvement, respiratory involvement, and cognitive impairment

|             | Participants (N=41) |                     |                         |                                                               |
|-------------|---------------------|---------------------|-------------------------|---------------------------------------------------------------|
|             | Scoliosis           | Cardiac involvement | Respiratory involvement | Age at which cognitive impairment was first detected/assessed |
| Age, n (%)  |                     |                     |                         |                                                               |
| 0-3 years   | 0 (0.0)             | 0 (0.0)             | 0 (0.0)                 | 6 (14.6)                                                      |
| 4-7 years   | 5 (12.2)            | 4 (9.8)             | 1 (2.4)                 | 17 (41.5)                                                     |
| 8-11 years  | 16 (39.0)           | 13 (31.7)           | 13 (31.7)               | 11 (26.8)                                                     |
| 12-17 years | 18 (43.9)           | 19 (46.3)           | 21 (51.2)               | 4 (9.8)                                                       |
| ≥18 years   | 2 (4.9)             | 5 (12.2)            | 6 (14.6)                | 3 (7.3)                                                       |

Abbreviations: DMD, Duchenne muscular dystrophy.

**Table 5** Clinical management of patients with DMD

|                                                                                    | N=41      |
|------------------------------------------------------------------------------------|-----------|
| Corticosteroids prescribed as first-line treatment, n (%)                          | 39 (95.1) |
| Age at treatment initiation, n (%) <sup>a</sup>                                    |           |
| <4 years                                                                           | 9 (23.1)  |
| 4-5 years                                                                          | 26 (66.7) |
| 6-7 years                                                                          | 3 (7.7)   |
| 8-9 years                                                                          | 1 (2.6)   |
| Corticosteroid regimen, n (%) <sup>a</sup>                                         |           |
| 0.9 mg/kg/day of deflazacort                                                       | 35 (89.7) |
| 0.75 mg/kg/day of prednisone                                                       | 4 (10.3)  |
| 0.75 mg/kg/day of prednisone on alternate days                                     | 0 (0.0)   |
| 0.75 mg/kg/day of prednisone for 10 days, followed by 10 days of resting           | 0 (0.0)   |
| Other regimen                                                                      | 0 (0.0)   |
| Gene therapy recommendation for eligible DMD patients, n (%)                       |           |
| No                                                                                 | 0 (0.0)   |
| I would need more scientific evidence                                              | 5 (12.2)  |
| Yes                                                                                | 36 (87.8) |
| Other disease-modifying therapies in patients not eligible for gene therapy, n (%) | 33 (80.5) |
| Exon-skipping                                                                      | 8 (24.2)  |
| Reading frame restoring                                                            | 3 (9.1)   |
| Both                                                                               | 22 (66.7) |

Abbreviations: DMD, Duchenne muscular dystrophy.

Available data: <sup>a</sup> 39

**Table 6** Questionnaires of quality of life and functional assessment used in patients with DMD

|                                                                                  | N=41      |
|----------------------------------------------------------------------------------|-----------|
| Quality of life questionnaires used for patients with DMD, n (%)                 |           |
| PedsQL 3.0 NMM                                                                   | 12 (29.3) |
| PedsQL 4.0 GCS                                                                   | 2 (4.9)   |
| SF-36 Health Survey                                                              | 2 (4.9)   |
| PROMIS                                                                           | 1 (7.3)   |
| None                                                                             | 24 (58.5) |
| Others                                                                           | 2 (7.3)   |
| Frequency of administration, n (%)                                               |           |
| Every 6 months                                                                   | 4 (9.8)   |
| Once a year                                                                      | 12 (29.3) |
| Not applicable                                                                   | 25 (61.0) |
| Scales and tests used in hospitals for functional assessment, n (%) <sup>a</sup> | 31 (77.5) |
| Most commonly used motor scale, n (%) <sup>b</sup>                               |           |
| North Star Ambulatory Assessment Scale                                           | 16 (51.6) |
| 6-minute walk test                                                               | 10 (32.3) |
| Functional timed tests                                                           | 3 (9.7)   |
| Performance of Upper Limb                                                        | 0 (0.0)   |
| Others                                                                           | 2 (6.5)   |

Abbreviations: DMD, Duchenne muscular dystrophy; GCS, Generic Core Scales; NMM, Neuromuscular Module; SF-36, Short Form-36; PedsQL, Pediatric Quality of Life Inventory; PROMIS, Patient-Reported Outcomes Measurement Information System.

Available data: <sup>a</sup> 40; <sup>b</sup> 31.

**Table 7** Multidisciplinary care team for managing patients with DMD

| N=41                                               |           |
|----------------------------------------------------|-----------|
| Hospitals with multidisciplinary care teams, n (%) | 28 (68.3) |
| Type of specialists, n (%) <sup>a</sup>            |           |
| Cardiologist                                       | 27 (96.4) |
| Rehabilitation physician                           | 27 (96.4) |
| Pulmonologist                                      | 26 (92.9) |
| Endocrinologist                                    | 24 (85.7) |
| Physiotherapists                                   | 24 (85.7) |
| Nutritionist/Dietitian                             | 23 (82.1) |
| Orthopedic surgeon/Traumatologist                  | 22 (78.6) |
| Gastroenterologist                                 | 22 (78.6) |
| Respiratory physiotherapy specialist               | 20 (71.4) |
| Speech therapist                                   | 20 (71.4) |
| Psychologist                                       | 19 (67.9) |
| Psychiatrist                                       | 13 (46.4) |

Abbreviations: DMD, Duchenne muscular dystrophy.

Available data: <sup>a</sup> 28

**Table 8.** Referral of patients with DMD to specialists

|                                                                                      | N=41      |
|--------------------------------------------------------------------------------------|-----------|
| Referral of patients to a hospital rehabilitation physician, n (%)                   | 40 (97.6) |
| Frequency of the rehabilitation attendance, n (%) <sup>a</sup>                       |           |
| Every 6 months                                                                       | 37 (92.5) |
| Referral of patients to the hospital physiotherapy, n (%)                            | 33 (80.5) |
| Frequency of physiotherapy attendance, n (%) <sup>b</sup>                            |           |
| Every 6 months                                                                       | 31 (93.9) |
| Referral of patients to the hospital's orthopedic surgeon or traumatologist, n (%)   | 33 (80.5) |
| Frequency of the orthopedic surgeon or traumatologist attendance, n (%) <sup>b</sup> |           |
| Every 6 months                                                                       | 12 (36.4) |
| Once a year                                                                          | 21 (63.6) |
| Referral of patients to the hospital's endocrinologist, n (%)                        | 35 (85.4) |
| Frequency of the endocrinologist attendance, n (%) <sup>c</sup>                      |           |
| Every 6 months                                                                       | 19 (54.3) |
| Once a year                                                                          | 16 (45.7) |
| Referral of patients to the hospital's pulmonologist, n (%)                          | 39 (95.1) |
| Frequency of the pulmonologist attendance, n (%) <sup>d</sup>                        |           |
| Every 6 months                                                                       | 31 (79.5) |
| Once a year                                                                          | 8 (20.5)  |
| Referral of patients to the hospital's cardiologist, n (%)                           | 40 (97.6) |
| Frequency of the cardiologist attendance, n (%) <sup>a</sup>                         |           |
| Every 6 months                                                                       | 16 (40.0) |
| Once a year                                                                          | 24 (60.0) |
| Referral of patients to the hospital's gastroenterologist, n (%)                     | 25 (61.0) |
| Frequency of the gastroenterologist attendance, n (%) <sup>e</sup>                   |           |
| Every 6 months                                                                       | 13 (52.0) |
| Once a year                                                                          | 12 (48.0) |

|                                                                                          |           |
|------------------------------------------------------------------------------------------|-----------|
| Referral of patients to the hospital's nutritionist or dietitian, n (%)                  | 28 (68.3) |
| Frequency of the nutritionist or dietitian attendance, n (%) <sup>f</sup>                |           |
| Every 6 months                                                                           | 17 (60.7) |
| Once a year                                                                              | 11 (39.3) |
| Referral of patients to the hospital's psychologist, n (%)                               | 24 (58.5) |
| Frequency of the psychologist attendance, n (%) <sup>g</sup>                             |           |
| Every 6 months                                                                           | 15 (62.5) |
| Once a year                                                                              | 9 (37.5)  |
| Referral of patients to the hospital's neurologist, n (%)                                | 38 (92.7) |
| Mean referral age to a neurologist, n (%) <sup>h</sup>                                   |           |
| <14 years                                                                                | 0 (0.0)   |
| 14-15 years                                                                              | 5 (13.2)  |
| 16-17 years                                                                              | 16 (42.1) |
| 18-19 years                                                                              | 14 (36.8) |
| >19 years                                                                                | 3 (7.9)   |
| Specific program available to refer patients to an adult neurologist, n (%) <sup>h</sup> | 14 (36.8) |

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Abbreviations: DMD, Duchenne muscular dystrophy.

Available data: <sup>a</sup> 40; <sup>b</sup> 33; <sup>c</sup> 35; <sup>d</sup> 39; <sup>e</sup> 25; <sup>f</sup> 28; <sup>g</sup> 24; <sup>h</sup> 38