

Duchenne Muscular Dystrophy in Europe

Results of the CARE-NMD cross-sectional survey

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Introduction

Although international consensus guidelines¹ exist for optimal multidisciplinary care for Duchenne muscular dystrophy, their level of implementation and impact on health outcomes remains unclear. The EU-funded CARE-NMD project analysed the current care situation and health impact of patients with Duchenne muscular dystrophy (DMD) in 7 European countries (Bulgaria (BG), the Czech Republic (CZ), Denmark (DK), Germany (DE), Hungary (HU), Poland (PL) and the United Kingdom (UK)).

Methods

Questionnaires, translated into each local language, were distributed via the national patient registries in autumn 2011. All registered patients with DMD received either an online or paper version of the questionnaire. It contained 42 questions covering a wide range of care aspects. Data collection was completed on 15.04.2012. An overall response rate of 64% was achieved.

Table 1. Number of evaluated questionnaires (sent questionnaires)

BG	CZ	DK	DE	HU	PL	UK	Total
40 (73)	90 (191)	90 (131)	424 (545)	57 (70)	142 (246)	228 (421)	1071 (1677)

Results I

Patient cohort

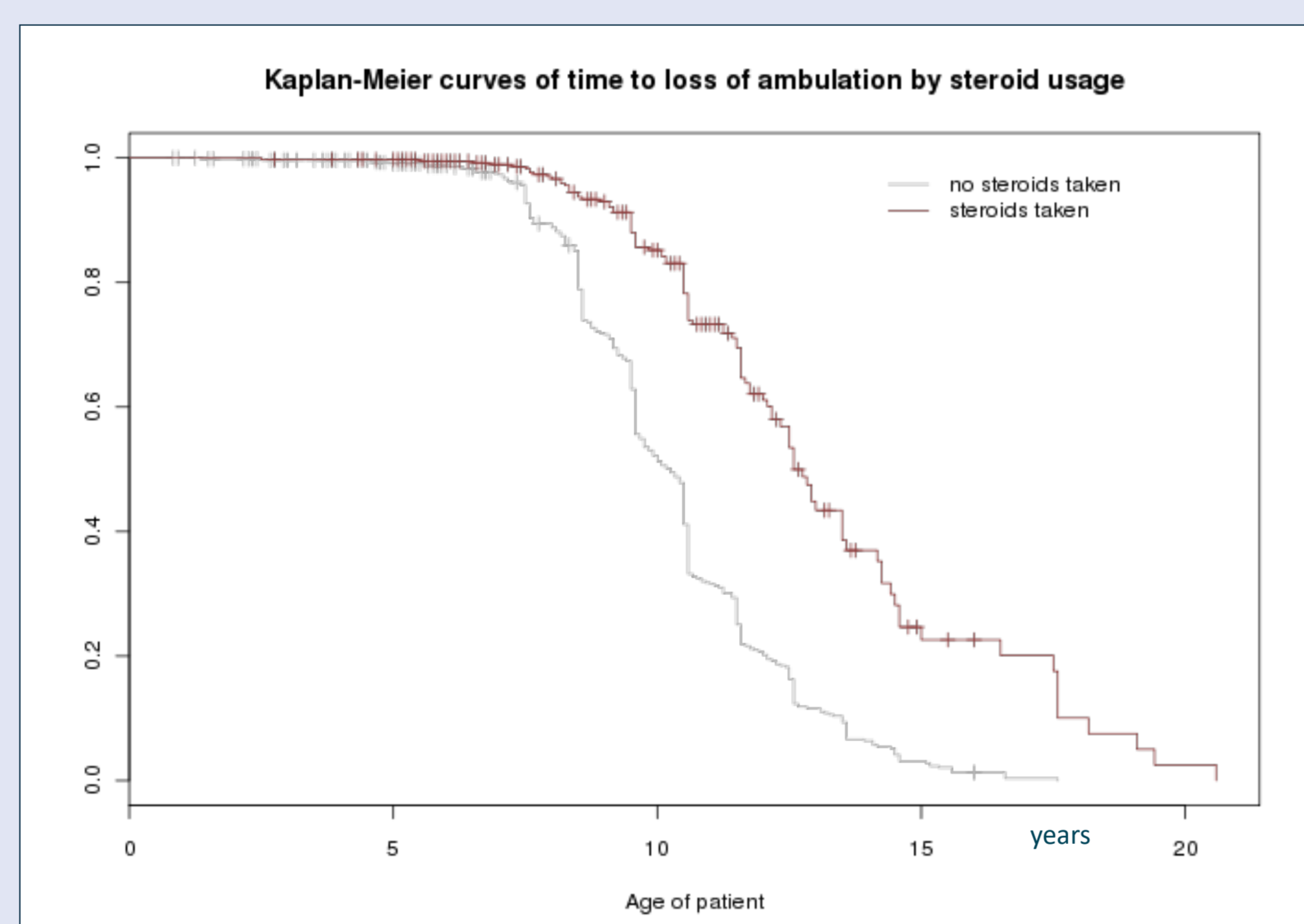
- The mean patient age was 13.1 years (range 0.8 – 46.2 years).
- 179 (17%) patients were aged >18 years.
- 498 (47%) patients were member of patient advocacy groups.
- 483 (45%) patients were ambulatory, 575 (54%) were not.

Diagnosis

- Mean delay between first concerns expressed by the parents and the established diagnosis was 1.4 years.
- Diagnosis was confirmed by genetic testing in 72% and/or muscle biopsy in 49%. However, 12 % had neither of them.

Steroid use

- Ambulation of patients with current or past steroid use (n=642, 60%) was significantly longer than of those without steroid medication (n=389, 36%).



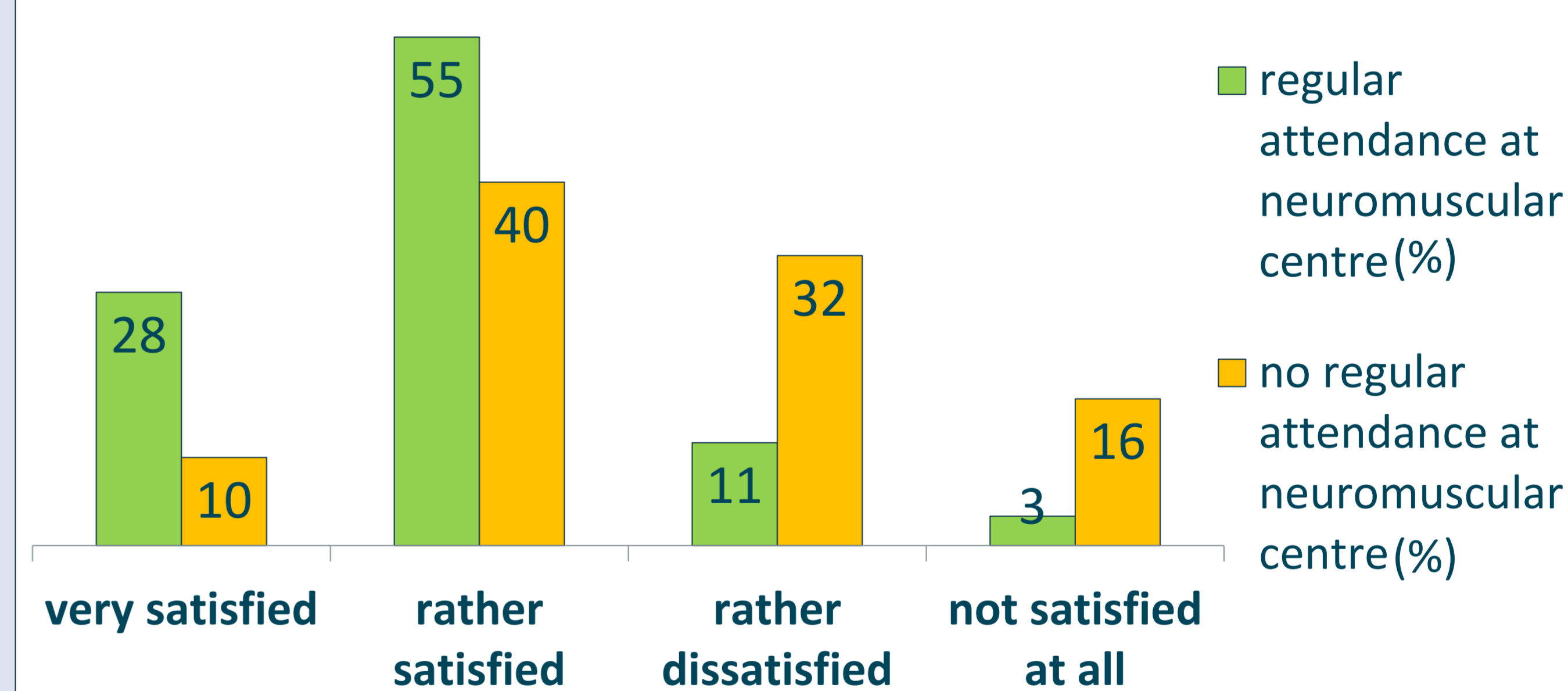
- Main reasons for not taking steroids were “not proposed by doctor” (46%) and family refusal (38%). 67% of patients regularly attending neuromuscular centres are/were steroid users versus 37% of those who don't.

Results II

Attendance of neuromuscular specialist

- 79% of all patients regularly attended a neuromuscular specialist (≥ once a year), but access varied by country and age: Of those patients aged >18 years, only 57% against 84% of paediatric patients regularly attended a neuromuscular specialist.
- Long distance was mentioned most frequently as the reason for not attending.

Satisfaction with medical treatment and attendance of neuromuscular centre



Assisted ventilation

- 41 patients received invasive and 115 non-invasive ventilation.

Assisted ventilation in patients with known FVC < 20% (n=35)

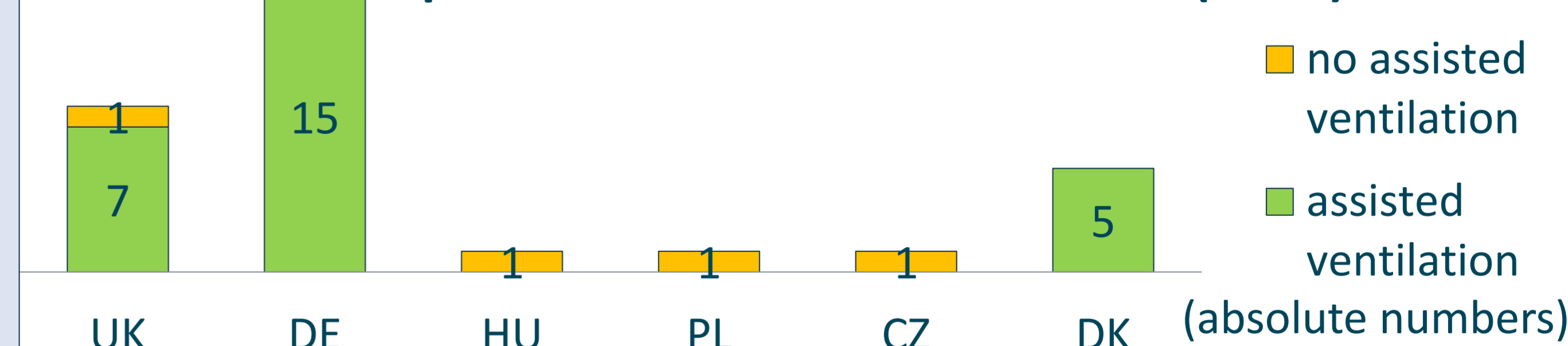


Table 2. summary of results

	BG	CZ	DK	DE	HU	PL	UK	overall
Mean patient age [years] at completing questionnaire (SD)	13.7 (6.6)	10.5 (5.3)	19.8 (10.4)	12.3 (6.9)	10.1 (5.5)	12.1 (6.1)	12.6 (6.9)	13.1 (7.3)
Mean age [years] at diagnosis (SD)	6.4 (4.0)	3.7 (2.0)	4.6 (2.5)	3.9 (2.4)	4.6 (2.7)	5.3 (2.5)	4.1 (2.0)	4.3 (2.5)
Mean age [years] at loss of ambulation	10.2 (2.0)	9.9 (1.82)	9.8 (2.4)	10.9 (2.7)	9.8 (3.1)	10.2 (1.8)	10.3 (2.1)	10.4 (2.4)
Current or past steroid users	32.5%	28.9%	36.7%	63.4%	45.6%	73.2%	75%	59.9%
Mean age [years] at start of steroids (SD)	6.4 (1.9)	7.0 (2.3)	5.9 (2.1)	6.0 (3.2)	6.5 (2.2)	5.9 (2.6)	6.4 (2.4)	6.2 (2.6)
Patients who have never taken steroids of those in late-ambulatory phase, absolute number and %	0 of 2 (0%)	6 of 10 (60%)	1 of 11 (9.1%)	8 of 64 (12.5%)	0 of 2 (0%)	2 of 13 (15.4%)	3 of 45 (6.7%)	20 of 147 (13.6%)

Conclusion: This is the largest cross-sectional study ever performed on people with DMD, describing their current functional status and care situation in Europe. It serves as a basis for further activities to improve implementation of Standards of Care (SoC). Compliance with standards of care on diagnosis, steroid use and assisted ventilation varies between European countries. The data presented here indicate that patients seen at neuromuscular centres receive better care according to SoC and are more satisfied with their treatment. Transition to adulthood is still problematic.