Supplemental material

Supplemental methods for questionnaire development

A panel of neuromuscular specialists from all seven participating countries developed the 42-question self-reporting questionnaire covering age, socio-demographic variables, and medical and psychosocial care history. Six health care domains were selected as the basis for questions according to consensus care recommendations (1;2). These included diagnosis as well as neuromuscular, rehabilitative/orthopaedic, cardiac, pulmonary, and psychosocial management. Clinical indicators describing health care performance (process indicators) and, where possible, the effect on patient health and well-being (outcome indicators) were defined for each domain (supplemental table 1). Selected outcomes included information about diagnosis, functional status, morbidity, occupation, patient satisfaction, and quality of life, but not mortality since the study was designed as a patient-report survey. Process indicators described care activities relevant for certain outcomes or patient well-being and in accordance with clinical care guidelines. Survey questions were designed to ensure provision of sufficient and specific information about health care and to be independently answered by and comprehensible to respondents, irrespective of nationality or education level. The first version of the multiple-choice self-reporting questionnaire was generated in English, then translated by the project partners into the national languages. Pilot surveys were conducted with five or ten patients and their families in each country to check comprehensibility, then adjustments and corrections were made by the central team in conjunction with local teams. This ensured consistency in the number of questions and their possible answers, to support the merging of all data into one English database for analysis. The final English edition (supplemental figure 1) was delivered to individual country partners for reconciliation with the local language version before full distribution. Each participating patient or family also received a set of age-appropriate, validated quality of life questionnaires, the results of which will be published separately.

Supplemental table

Supplemental table 1. Outcome[1] and process indicators[2] for different domains covered in the questionnaire.

Domain	Outcome and process indicators
Diagnosis	▪ Age at diagnosis[1] ▪ Time from first symptoms to definite diagnosis[1] ▪ Genetic testing[2] ▪ Information about genetic counselling[2]
Neuromuscular management	▪ Stage of disease in relation to age[1] ▪ Sitting ability[1] ▪ Age at loss of ambulation[1] ▪ Attendance at neuromuscular centre[2] ▪ Information about disease course[2] ▪ Information/treatment with corticosteroids[2] ▪ Information about and receipt of physiotherapy[2]
Orthopaedic management	▪ Assessment of spine/scoliosis[2]
Cardiac management	▪ Information about cardiac problems in the course of the disease[2] ▪ Frequency of cardiac check-ups[2] ▪ Medical treatment for cardiomyopathy[2]
Pulmonary management	▪ Information about pulmonary problems in the course of the disease[2] ▪ Frequency of pulmonary check-ups[2] ▪ Mechanical ventilation[2]
Overall	▪ Unplanned hospitalisations during the past two years[1] ▪ Overall satisfaction with medical treatment[1] ▪ Quality of life[1]

Reference List

(1) Bushby K, Finkel R, Birnkrant DJ, Case LE, Clemens PR, Cripe L, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. Lancet Neurol 2010 Jan;9(1):77-93.

(2) Bushby K, Finkel R, Birnkrant DJ, Case LE, Clemens PR, Cripe L, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. Lancet Neurol 2010 Feb;9(2):177-89.