

Patterns of decline in upper limb function of boys and men with DMD: an international survey

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Abstract With increasing life expectancy, upper extremity (UE) function becomes more and more important in boys with Duchenne muscular dystrophy (DMD). Knowledge of UE function in these children is, however, limited. The aim of this study was to gain insight into the changing patterns of UE function during the course of DMD. A Web-based questionnaire on UE function, covering all domains of the International Classification of Functioning Disability and Health, was distributed worldwide. Primary domains of the questionnaire were: participant characteristics, UE pain and stiffness, UE activities, and social participation. Data were described per disease stage and analyzed using descriptive analysis. A total of 213 boys/men with DMD (1–35 years) were included in this study. UE pain, stiffness, and activity limitations increased with disease stage. UE activity limitations already occurred in the early ambulatory stage. Compared to the healthy population, social participation was restricted in DMD patients and about 70 % of the respondents experienced UE limitations when performing social activities. Despite the existence of UE impairments, only 9 % of the respondents used supportive aids. Functional capacities and activities of the UE are limited already in the early ambulatory stage of patients with DMD affecting their social participation. Therefore, clinicians should pay attention to UE limitations before DMD patients lose their capacity to walk. Effective and adequate aids as well as attention for pain and stiffness in the therapeutic

management could help to reduce UE activity limitations and related restrictions in social participation.

Keywords Duchenne · Upper extremity function · Activities · Pain · Stiffness · Social participation

Introduction

Duchenne muscular dystrophy (DMD) is the most common form of muscular dystrophy in children, with an incidence of one in 6,000 male live births [22]. DMD is an X-linked recessive disorder characterized by progressive muscle wasting and weakness. Up to this point, there is no cure for DMD, and treatment is mainly aimed at delaying disease progression and preserving functional abilities. Due to these treatments (including nocturnal ventilation), life expectancy in boys with DMD has increased from 14 years of age in the 1960s to 25 years of age in the 1990s. Currently, the median survival of boys with DMD is estimated to be over 30 years [8, 16].

To improve care for DMD patients and to develop tailored training and new supportive aids, it is important to gain more insight into the course of the disease and the factors affecting its course. The current literature on disease progression is mostly aimed at the level of muscle or cell structures, and hardly at the level of function or activity [12, 23, 32]. The maintenance of function and activity, however, is highly related to the level of independence and quality of life [24]. Therefore, it is relevant to investigate disease progression from the levels of function and activity as well.

The little knowledge that there is on function and activity in boys with DMD is mainly focused on the lower extremity. Loss of lower extremity function can be

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compensated fairly well by using a wheelchair; in contrast, upper extremity (UE) function is much harder to support. There are only a few supporting devices for the arms available, and these devices do not cover the full range of function and activity [19]. With the current life expectancy, boys with DMD will live with impaired UE function for more than 15 years. If left unsupported, they may be seriously limited in UE activities and restricted in social participation for the same period of time.

In the literature, little is known about UE function in the course of DMD, especially regarding the execution of complex activities [30]. Understanding the execution of complex activities, e.g., during self-care and domestic life, is essential for the development of therapeutic interventions and supportive aids.

The International Classification of Function, Disability and Health (ICF) presents a framework to describe human functioning at three different levels: the level of body functions and structures, the level of activities, and the level of social participation [26]. The latter two are highly interrelated. The relation between these levels, however, is not linear. Therefore, it is necessary to study upper limb function in a broad perspective, taking all domains of the ICF into account.

The aim of this study was to gain insight into the changing patterns of UE function in the course of DMD by means of an internationally distributed Web-based questionnaire focusing on all levels of the ICF.

Methods

Procedures

A Web-based questionnaire, containing questions on all ICF domains (function, activity, participation), was translated into five languages (English, Dutch, German, Italian, Spanish). This questionnaire was subsequently distributed around the world by contacting Duchenne patients' organizations worldwide and asking them to send the Internet address of the questionnaire to their members. The full questionnaire can be found in "[Appendix](#)". This procedure was approved by the medical ethical committee in the Arnhem-Nijmegen region (the Netherlands) and has therefore been performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki and its later amendments.

Participants

The questionnaire could be filled in by patients with DMD or their parents or caregivers. Because an anonymous Web-based questionnaire was used in this study, the diagnosis of

DMD could not be confirmed by DNA diagnosis. However, to make sure that respondents fitted the clinical Duchenne phenotype, the diagnostic criteria of Emery were used [9]. Based on these criteria, respondents were excluded if the diagnosis was made after the age of 10 and if wheelchair confinement occurred after the age of 13 (when the respondents did not use corticosteroids). In addition, female DMD patients and respondents with the diagnosis of Becker muscular dystrophy (BMD) or any other muscular dystrophy were excluded. Respondents were also excluded if the stage of the disease could not be determined based on their answers.

Outcome measures

Outcomes were categorized in four different categories. Participant characteristics were shown to give insight in the population. Pain and stiffness give insight in the ICF function level, UE activity gives insight in the ICF activity level, and social participation gives insight in the ICF participation level.

Participant characteristics

To see if the population is comparable to the DMD population reported in literature, the following participant characteristics were assessed: age, age of diagnosis, age of wheelchair confinement, corticosteroid use, presence of scoliosis (based on the respondents knowledge, not confirmed by a physician) and the use of assistive devices for the arms.

Pain and stiffness

Questions concerning pain and stiffness were modified from the University of Michigan Upper Extremity Questionnaire [28]. Three different aspects of pain and stiffness were assessed: frequency (range, 0–6), severity (range, 0–10) and limitations due to pain and stiffness (range, 0–10). Pain and stiffness combination scores were calculated by taking the sum of the frequency, severity and limitation scores for pain and stiffness, respectively (range, 0–26). The percentage of respondents that experience pain was set at a combination score larger than 1.

UE activity

Items at the level of activities were chosen from existing measures used in clinical practice and were based on the study by van Beek et al. (submitted) [30]. They concluded that the Capabilities of Upper Extremity questionnaire (CUE) [18] and the ABILHAND questionnaire [31] are the most applicable self-report instruments to investigate the upper extremity activity level in teenage boys with DMD.

The CUE examines basic UE mobility activities. In addition, the ABILHAND examines complex UE activities. Next to the 22 ABILHAND items described by Vandervelde et al. [31], four more items were added (i.e., eat with a spoon, use fork and knife, drink a glass of water without straw, and use the keyboard of a computer) because these activities were indicated as very important by boys with DMD [30]. This adapted scale will be referred to as ABILHAND-plus. Furthermore, the Brooke scale [3] was selected as the gold standard for assessing basis UE activity in patients with DMD. Lastly, participants were asked for the three activities that cause the most problems due to UE impairments.

Social participation

Concerning social participation, participants were asked whether they went to school, had a job, practiced sports, had hobbies, performed activities with friends, and/or were involved in a romantic relationship. In addition, the respondents were asked if they experienced UE limitations while performing social activities (5-point scale).

Analysis

For all outcome measures, the total group score was determined as well as the score per disease stage. Four different disease stages were defined based on the guidelines of Bushby et al. [4]: in the early ambulatory stage, walking difficulties are experienced, however, the person is still able to climb stairs; in the late ambulatory

stage, the person is still able to walk, but not able to climb stairs; in the early non-ambulatory stage, persons are no longer able to walk, but their UE function is not very limited (Brooke scale 1–2 [3]); and in the late non-ambulatory stage, UE function is increasingly limited (Brooke ≥ 3).

Descriptive analysis of the data was performed: mean, median, standard deviation, and frequency tables were calculated if applicable. When the participants did not fully complete the questionnaire, all available items were included in the analysis. Wilcoxon rank-sum tests for independent groups were used to compare differences in pain and stiffness between the preferred and non-preferred side. All statistical analyses were done using IBM SPSS Statistics version 20 for Windows (IBM, Somers, NY, USA).

Results

Participant characteristics

In total, 344 participants from 14 different countries (Italy, the Netherlands, England, Spain, USA, Germany, Belgium, Switzerland, Canada, Ireland, Australia, Nepal, Peru, and India) answered the questionnaire, of which 131 were excluded based on the exclusion criteria. From the 213 remaining participants, 198 filled in the complete questionnaire, whereas 15 participants filled in the questionnaire only partially. Table 1 shows the participant characteristics.

Table 1 Participant characteristics

Participant characteristics	Total	Early ambulatory stage	Late ambulatory stage	Early non-ambulatory stage	Late non-ambulatory stage
Age (median range)	13.1 (1.5–35.2)	7.2 (1.5–16.7)	11.6 (7.1–21.7)	13.5 (8.4–19.5)	19.9 (9.2–35.2)
<i>N</i>	213	66	29	24	94
Age of diagnosis (median range)	4 (0–10)	3 (0–8)	4 (0–10)	4 (0–9)	4 (0–10)
<i>N</i>	213	66	29	24	94
Age wheelchair confined	10 (1–20)	–	–	12 (6–17)	10 (1–20)
<i>N</i>	112	–	–	23	89
Corticosteroid use (%) ^a	54.7/11.3/34.0	71.2/1.5/25.8	96.6/3.4/0.0	79.2/8.3/12.5	23.4/21.3/55.3
<i>N</i>	212	65	29	24	94
Scoliosis (%) ^b	17.8/31.0/51.2	3.0/13.6/83.3	0.0/37.9/62.1	4.2/33.3/62.5	37.2/40.4/22.3
<i>N</i>	213	66	29	24	94
Percentage wearing arm splints (%)	9.4	6.1	0.0	0.0	17.0
<i>N</i>	213	66	29	24	94
Percentage using arm support (%)	8.5	0.0	3.4	4.2	17.0
<i>N</i>	213	66	29	24	94

^a First number = currently uses corticosteroids, second number = did use corticosteroids in the past, third number = does not use corticosteroids

^b First number = severe scoliosis, second number = mild scoliosis, third number = no scoliosis

Pain and stiffness

The pain and stiffness combination scores ranged between 0 and 26 (26 = maximum possible score). No differences were found between the preferred and non-preferred side. Pain was most frequently present the shoulders, while stiffness was most frequently present in the fingers. Pain levels gradually increased with disease stage, while stiffness levels increased most in the late non-ambulatory stage (Fig. 1; Table 2). In Table 2, the pain and stiffness levels of the preferred and non-preferred side are combined.

UE activity

Forty-four percent of the respondents in the early ambulatory stage reported limitations while performing the basic activities of the CUE. In addition, 25 % of the respondents in the early ambulatory stage reported that it was difficult

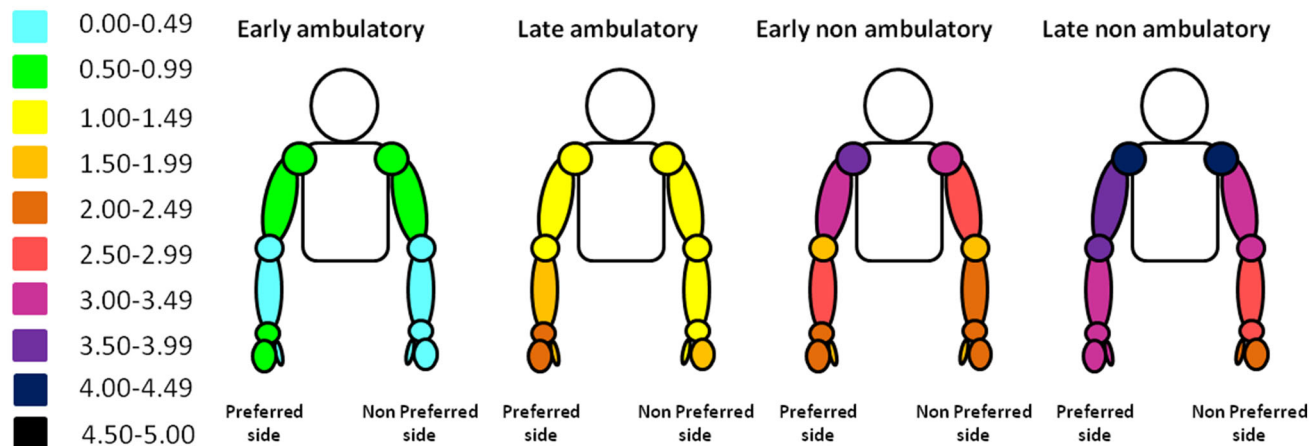
or impossible to perform some of the daily activities from the ABILHAND-plus. These percentages increase to 95 and 90 %, respectively, in the late non-ambulatory stage (Table 3).

Overall, the activity “eat and preparing food” was experienced most problematic. However, in the ambulatory stages the activities “get dressed”, “reach to objects/lift objects” and “write” were mentioned more often, while in the non-ambulatory stages “personal hygiene”, “drink”, and “use the computer” were mentioned most next to the activity “eat and preparing food” (Table 4).

Social participation

Restrictions in social participation increased with increased disease stage. The percentage of respondents that experience UE limitations when performing social activities increases with the stage of the disease (Table 5).

Pain Combination Score



Stiffness Combination Score

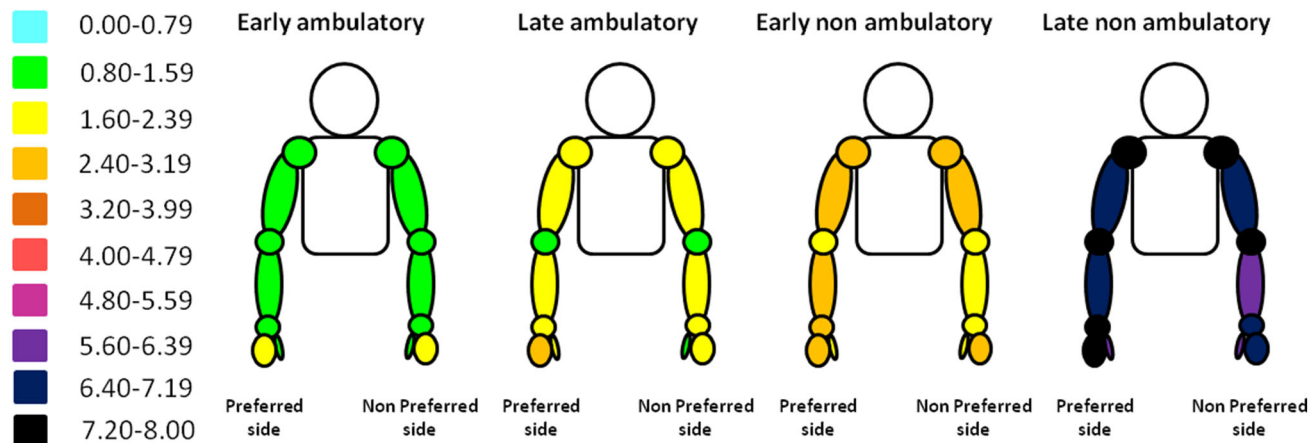


Fig. 1 Average pain and stiffness combination scores per body segment

Table 2 Pain and stiffness

	Total	Early ambulatory stage	Late ambulatory stage	Early non-ambulatory stage	Late non-ambulatory stage
Percentage of respondents that experienced pain (%) (pain combination score >1)					
Shoulders	39.4	13.6	24.1	45.8	60.6
Upper arms	35.0	17.4	24.1	43.8	48.4
Elbows	33.3	9.1	22.4	29.2	54.8
Forearms	32.9	13.6	27.6	39.6	46.3
Wrists	31.2	11.4	29.3	35.4	44.7
Thumbs	25.8	7.6	24.1	29.2	38.3
Fingers	29.3	11.4	27.6	33.3	41.5
<i>N</i>	213	66	29	24	94
Percentage of respondents that experienced stiffness (%) (stiffness combination score >1)					
Shoulders	42.7	25.0	32.8	39.1	59.0
Upper arms	39.6	25.8	32.8	34.8	52.7
Elbows	40.1	22.7	29.3	28.3	58.5
Forearms	38.9	24.2	31.0	32.6	53.2
Wrists	41.3	25.0	34.5	28.3	58.0
Thumbs	38.4	25.0	31.0	30.4	52.1
Fingers	45.3	30.3	39.7	34.8	60.1
<i>N</i>	213	66	29	24	94

Discussion

This study showed that activity limitations of the upper extremity in DMD already occur in the early ambulatory phase, and increase with more advanced stages of the disease. In addition, pain and stiffness increase with more advanced disease stages and restrictions in participation are more frequently present in more advanced disease stages.

Participant characteristics

The respondents in this study were between 1 and 37 years old and comprised DMD patients in all stages of the disease. Age of diagnosis, age of being wheelchair confined, prevalence of scoliosis, and corticosteroid use were comparable to the results reported in literature [5, 16, 20, 21].

The use of splints and supportive devices for the arms was around 9 %. However, the percentage of participants that reported having difficulties using their UE was much larger. A Brooke scale of 1 was reported by merely 34 % of the respondents, indicating that 66 % of the boys already experienced some activity limitations, even in an early stage of the disease. Only a small percentage of the participants that experienced upper extremity limitations used an arm support. This finding is in contrast with the lower extremity, where splints are highly recommended and used. The non-frequent use of arm supports could be caused by the fact that arm supports do not give natural support or

that the arm supports are too prominent. Both invisibility and the ability to give natural support are important for orthotics to be worn in daily life [27].

Pain and stiffness

In total, 35.6 % of the respondents experienced pain in their UE more than a few times a month; in adults this percentage was 55.4 %. These numbers are comparable with the literature, where percentages between 4.3 and 54 % have been reported [10, 29, 33].

Pain combination scores gradually increased in the more advanced disease stages. The average pain and stiffness combination scores are relatively low (Fig. 1). This is probably due to the large number of respondents that do not experience pain or stiffness. The pain combination scores of the respondents that did experience pain ranged from 1 to 21 and the stiffness combination scores ranged from 1 to 26. No other studies on the relation between pain and disease progression in DMD were found. Stiffness also appeared to increase with the stage of the disease, which is in correspondence with Cornu et al. [7]. They, however, measured the stiffness in the joint, whereas we assessed the subjective experience of joint stiffness.

Overall, pain was most severe in the shoulders. This is in accordance with the results of Engel et al. [10] and Tifferau et al. [29]. Stiffness was most severe in the fingers. One explanation for this could be that the participants were still

Table 3 Activity limitations (Brooke, CUE, ABILHAND-plus) per disease stage

Brooke scale	Total	Early ambulatory stage	Late ambulatory stage	Early non-ambulatory stage	Late non-ambulatory stage
Median (range)	2 (1–6)	1 (1–2)	2 (1–4)	2 (1–2)	5 (3–6)
<i>N</i>	213	66	29	24	94
CUE ^a (%/%)					
Reach forward, shoulder level	23/42	16/0	52/11	60/4	9/90
Arms over head	20/48	23/0	46/16	52/21	2/98
Reach to the floor	28/55	48/11	52/30	46/42	3/96
Raise a 5-pound object over the head	29/60	61/13	29/50	58/42	0/100
Slide a light object towards you	34/29	17/0	52/2	54/0	35/63
Slide a 10-pound object towards you	36/54	69/9	57/32	50/42	4/94
Slide a light object away from you	32/28	17/0	48/2	38/4	37/61
Slide a 10-pound object away from you	35/53	63/9	57/7	46/46	7/90
Push up in chair	25/68	53/23	36/64	29/71	1/99
Curl wrist upward	40/31	44/0	57/7	44/15	32/63
Supination	39/36	42/2	59/11	65/15	25/72
Hold a hammer					
Pick up a small object with thumb and first two fingers	33/20	20/0	46/0	21/4	42/43
Hold a small object between thumb and index finger	36/19	27/2	48/0	17/8	44/40
Hold/open a 2-pound object with the tips of the fingers	35/51	59/5	63/29	52/40	6/91
Manipulate a small object with the fingers	42/27	45/8	32/21	44/8	42/47
Push a button with tip of the index finger	34/20	23/0	36/0	33/0	40/44
Average	33/40	39/5	48/19	44/23	21/74
ABILHAND-plus ^b (%)					
Take the cap off a bottle ^c	74	49	48	63	98
Cut nails ^c	88	81	83	77	96
Button up a shirt	77	56	55	68	97
Fasten the zipper of a jacket	63	22	32	41	100
Turn a key in a keyhole	67	34	52	38	99
Fasten a snap, e.g., from jacket or bag	67	34	40	43	100
Open a pack of chips ^d	73	57	38	59	95
Open a pack of biscuits	69	49	38	52	93
Insert a key in keyhole ^c	54	11	23	33	94
Turn off a tap	52	16	19	18	90
Turn on a tap	52	18	19	18	90
Fill a glass with water	62	13	60	45	96
Sharpen a pencil ^d	52	25	27	25	82
Open a lunch box	56	21	27	32	91
Squeeze toothpaste onto a toothbrush	57	26	31	21	92
Spread butter on a slice of bread	60	30	28	35	92
Open a toothpaste tube	56	30	19	21	92
Count banknotes ^c	46	12	14	25	74
Deal cards ^d	54	16	33	24	87
Unwrap a chocolate bar ^d	45	7	15	21	82
Dry hands	43	5	0	13	87
Wash hands	40	2	8	4	84
Eat with a spoon	38	2	4	13	76
Use fork and knife	56	19	27	43	89
Drink a glass of water without straw	45	0	8	13	92

Table 3 continued

Brooke scale	Total	Early ambulatory stage	Late ambulatory stage	Early non-ambulatory stage	Late non-ambulatory stage
Use keyboard of a computer	31	4	4	4	63
Average	57	25	29	33	90

^a CUE: first number = percentage of respondents that answered the activity to be moderately, somewhat or a little limited; second number = percentage of respondents that answered the activity to be very, extremely, or totally limited. The rest percentage is the percentage of respondents that answered that the activity was not limited (percentage not shown in table)

^b ABILHAND-plus: percentage of respondents that answered the activity to be difficult or impossible. The rest percentage is the percentage of respondents that answered the activity to be easy (percentage not shown in table)

^c Items that were specific to adults

^d Items that were specific to children

Table 4 Activities that cause the most problems in daily life due to UE impairments

	Total	Early ambulatory stage	Late ambulatory stage	Early non-ambulatory stage	Late non-ambulatory stage
Eat and prepare food (%)	13	6	4	10	18
Get dressed (%)	12	19	20	16	7
Reach to objects/lift objects (%)	11	15	24	18	6
Write (%)	9	15	14	10	7
Personal hygiene (%)	9	6	6	4	12
Drink (%)	8	2	4	2	12
Using the computer (%)	7	0	0	6	11
Play/crafts (%)	4	11	6	2	2
Use the toilet (%)	4	2	6	4	4
Playing video games/control television/use telephone (%)	3	0	0	4	5
Touch/scratch the face (%)	3	0	0	0	5
Open packaging (%)	3	5	6	4	1
Open doors/unlock locks (%)	3	5	4	4	2
Control the wheelchair (%)	3	0	4	0	4
Use books/schoolbags (%)	2	2	0	6	2
Practice sports (%)	2	8	0	4	0
Interaction with other humans (%)	1	0	0	2	1
Other (%)	3	5	2	2	4
<i>N</i>	213	66	29	24	94

The numbers in this table represent the percentage of respondents that mentioned the activity when asking for the activities that cause the most problems in daily life due to UE impairments

able to use their fingers in a relatively late stage of the disease, while the shoulder and elbow could not be moved anymore, making patients probably less aware of the stiffness in their shoulders.

UE activity

The Brooke scale is the most commonly used instrument to evaluate the upper limb activity level in boys with DMD. The CUE has never been used in boys with DMD. The ABILHAND has been validated in boys with DMD [31]; and van Opstal et al. [25] used the ABILHAND to measure the capacity to manage daily activities that require the use

of the upper limb. They also divided the results for the different disease stages (“ambulant”, “nonambulant, relatively good arm abilities”, and “nonambulant, decreased arm abilities”). The scores per items were not shown in this study, however the total score indicated that arm function decreases with disease stage. This is comparable to the results of this study.

The ABILHAND has been validated in children older than 6 years, the CUE has only been used in adult subjects. Since 23 participants were under the age of 6 years and 175 participants were under the age of 18 years, it could be that some activities in the CUE and ABILHAND are not valid for the participants. The CUE, however, consists of basic

Table 5 Social participation per disease stage

	Total	Early ambulatory stage	Late ambulatory stage	Early non-ambulatory stage	Late non-ambulatory stage
Participants					
Going to school (%)	78	11	38	83	3
<i>N</i>	200	198	198	198	197
Working (%)	91	2	50	66	0
<i>N</i>	58	56	56	56	55
Playing sports (%)	96	4	30	89	0
<i>N</i>	27	27	27	27	27
Having a hobby	96	4	33	92	0
<i>N</i>	24	24	24	24	24
In a romantic relationship	59	20	35	89	7
<i>N</i>	91	91	91	91	91
Participation restrictions					
Experiencing limitations of the arms and/or hands during school activities (%) ^a	68/14	63/2	85/0	78/4	59/35
<i>N</i>	154	51	26	23	54
Experiencing limitations of the arms and/or hands during work activities (%) ^a	62/14	100/0	100/0	100/0	56/17
<i>N</i>	21	1	1	1	18
Experiencing limitations of the arms and/or hands playing sports (%) ^a	66/16	75/0	88/0	50/25	56/31
<i>N</i>	76	28	8	8	32
Experiencing limitations of the arms and/or hands during hobbies (%) ^a	60/7	46/0	58/0	45/0	70/15
<i>N</i>	164	37	24	22	81
Experiencing limitations of the arms and/or hands in a romantic relationship (%) ^a	33/33	–	–	–	33/33
<i>N</i>	6	0	0	0	6

^a Participation: first number = percentage of respondents that experienced mild participation restrictions due to UE limitations; second number = percentage of respondents that experienced severe participation restrictions due UE limitations. The rest percentage is the percentage of respondents that answered to experience no limitations in the arms and/or hands (percentage not shown in table)

activities that were considered as not very age-specific, therefore age was not expected to be a limiting factor to perform the CUE activities. The ABILHAND consists of more complex activities, which could be more difficult to perform by very young children. For example, the activities “cut nails”, “open a pack of chips” and “open a pack of biscuits” are pointed out more difficult in the early ambulatory stage compared to the late ambulatory stage. This is probably due to the fact that the children in the early ambulatory stage (median age, 7.2 years) are too young to be able to perform the item without difficulties.

Regarding the CUE, item 12 (holding an object like a hammer with your hand) was erroneously not included in the questionnaire. This, however, did not influence the remaining results of the CUE, since we looked at the separate items and not at the total score.

The scores on the reported Brooke scale increased with age. The median age at which UE activity level started to deteriorate (Brooke 2) was 11.4 years. This result is

comparable to the results of Jung et al. who found that the median age of Brooke scale 2 was around 10 years [15], and to Lord et al. [17] who found a median age around 11 years.

The Brooke scale gives a stepwise insight into UE activity, whereas CUE and ABILHAND-plus provide us with a more detailed description of the activity limitations related to the UE. Items from the CUE as well as the ABILHAND-plus are already difficult in the early ambulatory stage. This indicates that difficulties performing upper extremity tasks occur already long before boys with DMD lose the ability to walk. These early activity limitations related to UE impairments have not been reported before.

Social participation

Restricted social participation is a huge problem in boys with DMD [13]. This can result in reduced engagement in

social activities, social withdrawal, or even social isolation [4]. The results of this study show that 95.1 % of the respondents between 5 and 20 years went to school or attended other classes, which is comparable to the worldwide population in developed countries where 95.9 % of children attend school [1]. Of the respondents over 20 years of age, 34.8 % worked and 26.1 % of the respondents over 20 years of age still attended school. In the healthy population of the same age, over 80 % of the people are employed or have education [6].

Of all the boys with DMD, 37 % participated in sports and 7.4 % of the adults reported having a romantic relationship. These percentages are lower than in the healthy (adult) population worldwide [11, 14]. In comparison, Bendixen et al. [2] stated that boys with DMD showed less participation in the physical domain, but not in the recreational and social domains. The results of the current study, however, showed that participation in boys with DMD was also restricted in these other domains. This difference can be explained by the applied measurement instruments. Bendixen et al. [2] used the children's assessment of participation and enjoyment (CAPE), while we used open questions. In addition, the participants of the study by Bendixen et al. [2] were between 5 and 15 years of age, whereas the population also included boys older than 15 years. Older boys are expected to have more participation restrictions, since they have more UE impairments. This is confirmed by the results in Table 5, where about 70 % of the respondents report experiencing mild or severe UE limitations when performing social activities and these percentages tend to increase with disease stage.

Conclusions

Pain, stiffness, activity limitations, and social participation restrictions are higher in more advanced disease stages. However, they are already present in the early ambulatory stage. About 70 % of the respondents state that they experience UE limitations when performing social activities. Therefore, clinicians should already pay attention to upper limb activity limitation before the DMD patients lose their capacity to walk. Effective and adequate aids as well as attention for pain and stiffness in the therapeutic management could help to reduce UE activity limitations and related restrictions in social participation.

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Conflicts of interest On behalf of all authors, the corresponding author states that there are no conflicts of interest to report.

Appendix

Nr.	Questions	Answer options
1	Response ID	Given by the computer
2	What is your age?	Open question
3	Who filled out the questionnaire?	1 = Myself 2 = Mother 3 = Father 4 = Caregiver 5 = Other (open question)
4	In which country do you live?	Open question
5	Were you born in this country?	1 = Yes 2 = No
6	Since when have you lived in this country? (if question 5 = 2)	Open question
7	In which country were you born? (if question 5 = 2)	Open question
8	How tall are in cm?	Open question
9	What is your weight in kg?	Open question
10	What is your preferred hand?	1 = Right 2 = Left 3 = First right now left 4 = First left now right 5 = No preference

continued

Nr.	Questions	Answer options
11	When was the diagnosis Duchenne muscular dystrophy made for you?	Open question
12	Who made the diagnosis?	1 = General practitioner 2 = Pediatrician 3 = Pediatric neurologist 4 = Neurologist 5 = I do not know 6 = Other (open question)
13	Do you know which gene deviation you have?	1 = No 2 = Yes
14	Which gene deviation you have? (if question 13 = 2)	Open question
15	Do you have other chronic diseases?	1 = No 2 = Yes
16	What chronic disease do you have? (if question 15 = 2)	Open question
17	Have you ever seriously injured, e.g., a bone fracture one of your arms or hands?	1 = No 2 = Left arm 3 = Right arm 4 = Left hand 5 = Right hand (more than one answers possible)
18	Have you ever had surgery of one of your arms or hands?	1 = No 2 = Left arm 3 = Right arm 4 = Left hand 5 = Right hand (more than one answers possible)
19	Do you have spinal deformities, e.g., scoliosis?	1 = No 2 = Yes mild 3 = Yes severe
20	Was surgery performed to correct for spinal deformities? (if question 19 = 2 or 3)	1 = No 2 = Yes
21	In what year was surgery performed? (if question 20 = 2)	Open question
22	Do you use corticosteroids prednisone/prednisol or deflazacort at this moment?	1 = No 2 = Not anymore 3 = Yes
23	Which type of medication did you use? (if question 22 = 2)	1 = Prednisolon/prednisone 2 = Deflazacort
24	Did you use it continuously or with intervals? (if question 22 = 2)	1 = Continuously 2 = 10 days on 10 days off 3 = Other (open question)
25	Which dose did you use? (if question 22 = 2)	Open question
26	When did you start using this medication? (if question 22 = 2)	Open question
27	When did you stop using this medication? (if question 22 = 2)	Open question
28	Why did you stop using this medication? (if question 22 = 2)	Open question
29	When did you start using this medication? (if question 22 = 3)	Open question
30	Which type of medication do you use? (if question 22 = 3)	1 = Prednisolon/prednisone 2 = Deflazacort

continued

Nr.	Questions	Answer options
31	Do you use it continuously or with intervals? (if question 22 = 3)	1 = Continuously 2 = 10 days on 10 days off 3 = Other (open question)
32	Which dose did you use? (if question 22 = 3)	Open question
33	Do you use other medication that can possibly affect the course of Duchenne muscular dystrophy?	1 = No 2 = Yes
34	What medication that can possibly affect the course of Duchenne muscular dystrophy do you use? (if question 33 = 2)	Open question
35	Do you use supplements like vitamins or homeopathic remedies?	1 = No 2 = Yes
36	What supplements do you use? (if question 35 = 2)	Open question
37	Did you ever had physiotherapy?	1 = Never 2 = Yes, but not anymore 3 = Yes, with periods of no therapy 4 = Yes, continuously
38	How often do you have physiotherapy now? (if question 37 = 3 or 4)	Open question
39	For how long are your arms/hands treated by the physiotherapist each week? (if question 37 = 3 or 4)	1 = None 2 = ... minutes a week (open question)
40	What kind of physiotherapy do you receive for your arms/hands? (if question 39 = 2)	1 = Stretching 2 = Supported active movements 3 = Passive movements 4 = Other movements, namely (open question) (more than one answer possible)
41	Do you exercises your arms/hands yourself or with your parents/ caregivers?	1 = No 2 = Yes, on average once a week 3 = Yes, on average once a day 4 = Yes, more than once a day
42	What kind of exercises do you do by yourself or with your parents/ caregivers? (if question 41 = 2, 3 or 4)	1 = Stretching 2 = Supported active movements 3 = Passive movements 4 = Other movements, namely (open question) (more than one answer possible)
43	Do you swim or do you get hydrotherapy?	1 = No 2 = Yes
45	Did you ever receive occupational therapy, e.g., practicing daily activities or use of assistive devices?	1 = Never 2 = Yes, but not anymore 3 = Yes, with periods of no therapy 4 = Yes, continuously
46	How often do you receive occupational therapy currently? (if question 45 = 3 or 4)	Open question
47	For how long are your arms/hands treated by the occupational therapist each week? (if question 45 = 3 or 4)	1 = None 2 = ... minutes a week (open question)
48	What kind of occupational therapy do you receive for the arms/ hands? (if question 47 = 2)	1 = Practice use of devices 2 = Practice use of arm support 3 = Fitting of splints 4 = Different, namely... (open question)

continued

Nr.	Questions	Answer options
49	Which devices do you use for walking, transfers, or in therapy?	1 = Standing frame 2 = Long leg braces 3 = Wheelchair, pushed by somebody else 4 = Manual wheelchair (independent traveling) 5 = Manual wheelchair with electrical supported wheels 6 = Electrical wheelchair (independent traveling) 7 = Scooter 8 = Other motorized vehicle (e.g., Segway) 9 = 2-wheeled-bicycle 10 = 2-wheeled-bicycle with electrical support 11 = 3-wheeled-bicycle 12 = 3-wheeled-bicycle with electrical support
50	How often do you use these devices? (asked for the separate devices)	1 = Never 2 = Few times a year 3 = Few times a month 4 = Few times a week 5 = Almost every day 6 = Daily for a significant part of the day
50	Are you completely wheelchair confined?	1 = No 2 = Yes, since (open question)
51	Do you use splints for your arms and/or hands?	1 = No 2 = Yes, namely (open question)
52	How often do you wear these splints?	1 = Few times a year 2 = Few times a month 3 = Few times a week 4 = Almost every day 5 = Daily for a significant part of the day 6 = During the night
53	Do you use some kind of arm support other than splints?	1 = No 2 = Yes, namely (open question)
54	How often do you use this arm support?	1 = Few times a year 2 = Few times a month 3 = Few times a week 4 = Almost every day 5 = Daily for a significant part of the day
55	For which activities do you use the arm support?	Open question
56	How often do you have pain in your right shoulder?	0 = Never 1 = Few times a year 2 = Few times a month 3 = Few times a week 4 = Almost every day 5 = Daily for a significant part of the day 6 = Always
57	How often do you have pain in your right upper arm?	Same as question 56
58	How often do you have pain in your right elbow?	Same as question 56
59	How often do you have pain in your right forearm?	Same as question 56
60	How often do you have pain in your right wrist?	Same as question 56
61	How often do you have pain in your right thumb?	Same as question 56

continued

Nr.	Questions	Answer options
62	How often do you have pain in the fingers of your right hand?	Same as question 56
63	How often do you have pain in your left shoulder?	Same as question 56
64	How often do you have pain in your left upper arm?	Same as question 56
65	How often do you have pain in your left elbow?	Same as question 56
66	How often do you have pain in your left forearm?	Same as question 56
67	How often do you have pain in your left wrist?	Same as question 56
68	How often do you have pain in your left thumb?	Same as question 56
69	How often do you have pain in the fingers of your left hand?	Same as question 56
70	How severe is the pain in your right shoulder?	0 = No pain 10 = Worst pain imaginable
71	How severe is the pain in your right upper arm?	Same as question 70
72	How severe is the pain in your right elbow?	Same as question 70
73	How severe is the pain in your right forearm?	Same as question 70
74	How severe is the pain in your right wrist?	Same as question 70
75	How severe is the pain in your right thumb?	Same as question 70
76	How severe is the pain in the fingers of your right hand?	Same as question 70
77	How severe is the pain in your left shoulder?	Same as question 70
78	How severe is the pain in your left upper arm?	Same as question 70
79	How severe is the pain in your left elbow?	Same as question 70
80	How severe is the pain in your left forearm?	Same as question 70
81	How severe is the pain in your left wrist?	Same as question 70
82	How severe is the pain in your left thumb?	Same as question 70
83	How severe is the pain in the fingers of your left hand?	0 = No limitations 10 = Fully limited
84	How limited are you due to the pain in your right shoulder?	Same as question 83
85	How limited are you due to the pain in your right upper arm?	Same as question 83
86	How limited are you due to the pain in your right elbow?	Same as question 83
87	How limited are you due to the pain in your right forearm?	Same as question 83
88	How limited are you due to the pain in your right wrist?	Same as question 83
89	How limited are you due to the pain in your right thumb?	Same as question 83
90	How limited are you due to the pain in the fingers of your right hand?	Same as question 83
91	How limited are you due to the pain in your left shoulder?	Same as question 83
92	How limited are you due to the pain in your left upper arm?	Same as question 83
93	How limited are you due to the pain in your left elbow?	Same as question 83
94	How limited are you due to the pain in your left forearm?	Same as question 83
95	How limited are you due to the pain in your left wrist?	Same as question 83
96	How limited are you due to the pain in your left thumb?	Same as question 83
97	How limited are you due to the pain in the fingers of your left hand?	Same as question 83
98	How often do you experience stiffness in your right shoulder?	0 = Never 1 = Few times a year 2 = Few times a month 3 = Few times a week 4 = Almost every day 5 = Daily for a significant part of the day 6 = Always
99	How often do you experience stiffness in your right upper arm?	Same as question 98
100	How often do you experience stiffness in your right elbow?	Same as question 98
101	How often do you experience stiffness in your right forearm?	Same as question 98

continued

Nr.	Questions	Answer options
102	How often do you experience stiffness in your right wrist?	Same as question 98
103	How often do you experience stiffness in your right thumb?	Same as question 98
104	How often do you experience stiffness in the fingers of your right hand?	Same as question 98
105	How often do you experience stiffness in your left shoulder?	Same as question 98
106	How often do you experience stiffness in your left upper arm?	Same as question 98
107	How often do you experience stiffness in your left elbow?	Same as question 98
108	How often do you experience stiffness in your left forearm?	Same as question 98
109	How often do you experience stiffness in your left wrist?	Same as question 98
110	How often do you experience stiffness in your left thumb?	Same as question 98
111	How often do you experience stiffness in the fingers of your left hand?	Same as question 98
112	How severe is the stiffness in your right shoulder?	0 = No stiffness 10 = Worst stiffness imaginable
113	How severe is the stiffness in your right upper arm?	Same as question 112
114	How severe is the stiffness in your right elbow?	Same as question 112
115	How severe is the stiffness in your right forearm?	Same as question 112
116	How severe is the stiffness in your right wrist?	Same as question 112
117	How severe is the stiffness in your right thumb?	Same as question 112
118	How severe is the stiffness in the fingers of your right hand?	Same as question 112
119	How severe is the stiffness in your left shoulder?	Same as question 112
120	How severe is the stiffness in your left upper arm?	Same as question 112
121	How severe is the stiffness in your left elbow?	Same as question 112
122	How severe is the stiffness in your left forearm?	Same as question 112
123	How severe is the stiffness in your left wrist?	Same as question 112
124	How severe is the stiffness in your left thumb?	Same as question 112
125	How severe is the stiffness in the fingers of your left hand?	Same as question 112
126	How limited are you due to the stiffness in your right shoulder?	0 = No limitations 10 = Fully limited
127	How limited are you due to the stiffness in your right upper arm?	Same as question 126
128	How limited are you due to the stiffness in your right elbow?	Same as question 126
129	How limited are you due to the stiffness in your right forearm?	Same as question 126
130	How limited are you due to the stiffness in your right wrist?	Same as question 126
131	How limited are you due to the stiffness in your right thumb?	Same as question 126
132	How limited are you due to the stiffness in the fingers of your right hand?	Same as question 126
133	How limited are you due to the stiffness in your left shoulder?	Same as question 126
134	How limited are you due to the stiffness in your left upper arm?	Same as question 126
135	How limited are you due to the stiffness in your left elbow?	Same as question 126
136	How limited are you due to the stiffness in your left forearm?	Same as question 126
137	How limited are you due to the stiffness in your left wrist?	Same as question 126
138	How limited are you due to the stiffness in your left thumb?	Same as question 126
139	How limited are you due to the stiffness in the fingers of your left hand?	Same as question 126

continued

Nr.	Questions	Answer options
140	Which description is most suitable for you? (Brooke scale)	1 = Starting with my arms at my sides, I can lift both arms sideways in a full circle until they touch above my head 2 = I can raise both of my arms above my head only by flexing my elbow (i.e., shortening the circumference of the movement) or using trick movements 3 = I cannot raise my hands above my head but I can raise an 8-oz. (250 ml) glass of water to my mouth (by using one or both hands) 4 = I can raise my hands to my mouth (I can raise each hand separately) but I cannot raise an 8-oz. (250 ml) glass of water to my mouth 5 = I cannot raise my hand to my mouth but I can use my hands to hold a pen or pick up coins from the table 6 = I cannot raise my hands to my mouth and I have no useful function of my hands
141	Which description is most suitable for you? (Vignos scale)	1 = I walk and climb stairs without assistance 2 = I walk and climb stairs with aid of railing 3 = I walk and climb stairs slowly with aid of railing (over 12 s for 4 standard stairs) 4 = I walk unassisted and rise from chair but I cannot climb stairs 5 = I walk unassisted but I cannot arise from chair or climb stairs 6 = I walk only with assistance or I walk independently with long leg braces 7 = I walk in long leg braces but I require assistance for balance 8 = I stand in long leg braces but I am unable to walk even with assistance 9 = I am confined to a wheelchair 10 = I am confined to bed
142	Think about reaching out with your arm to touch something directly in front of you that is at shoulder level. How limited are you doing this using your right arm	1 = Totally limited 2 = Extremely limited 3 = Very limited 4 = Moderately limited 5 = Some limitation 6 = A little limited 7 = Not at all limited
143	Think about reaching out with your arm to touch something directly in front of you that is at shoulder level. How limited are you doing this using your left arm	Same as question 142
144	Think about raising your arm directly over your head, with your arm straight. How limited are you doing this using your right arm	Same as question 142
145	Think about raising your arm directly over your head, with your arm straight. How limited are you doing this using your left arm	Same as question 142
146	Think about reaching down to touch the floor and sitting back up straight, without hooking with your other arm or using it to pull yourself up. How limited are you doing this using your right arm?	Same as question 142
147	Think about reaching down to touch the floor and sitting back up straight, without hooking with your other arm or using it to pull yourself up. How limited are you doing this using your left arm?	Same as question 142
148	Think about raising a 5-pound object like a heavy blanket over your head using both arms. (Do not worry about whether you could grab it with your hands, just if you could raise something that heavy over your head). How limited are you doing this using both arms?	Same as question 142

continued

Nr.	Questions	Answer options
149	Think about pulling or sliding (without grasping) a light object such as a can of soda, that is on a table, towards you. How limited are you doing this using your right arm?	Same as question 142
150	Think about pulling or sliding (without grasping) a light object such as a can of soda, that is on a table, towards you. How limited are you doing this using your left arm?	Same as question 142
151	Think about pulling or sliding (without grasping) a heavy object (up to 10 pounds), that is on a table, towards you. How limited are you doing this using your right arm?	Same as question 142
152	Think about pulling or sliding (without grasping) a heavy object (up to 10 pounds), that is on a table, towards you. How limited are you doing this using your left arm?	Same as question 142
153	Think about pushing a light object such as a can of soda on a table, away from you. How limited are you doing this using your right arm?	Same as question 142
154	Think about pushing a light object such as a can of soda on a table, away from you. How limited are you doing this using your left arm?	Same as question 142
155	Think about pushing a heavy object (up to 10 pounds) on a table, away from you. How limited are you doing this using your right arm?	Same as question 142
156	Think about pushing a heavy object (up to 10 pounds) on a table, away from you. How limited are you doing this using your left arm?	Same as question 142
157	Think about pushing down with both arms into your chair enough to lift your buttocks (both sides) off the seat (do a push-up weight shift). How limited are you doing this?	Same as question 142
158	With your hand on your lap palm down, think about curling your wrist upwards, keeping your arm on your lap. How limited are you doing this using your right arm?	Same as question 142
159	With your hand on your lap palm down, think about curling your wrist upwards, keeping your arm on your lap. How limited are you doing this using your left arm?	Same as question 142
160	Think about turning your hand over, keeping your elbow bent at your side (like turning a doorknob or a dial). How limited are you doing this using your right arm?	Same as question 142
161	Think about turning your hand over, keeping your elbow bent at your side (like turning a doorknob or a dial). How limited are you doing this using your left arm?	Same as question 142
162	Think about picking up a small object such as a paper clip or the cap of a tube of toothpaste with the tips of your thumb and first two fingers. How limited are you doing this using your right arm?	Same as question 142
163	Think about picking up a small object such as a paper clip or the cap of a tube of toothpaste with the tips of your thumb and first two fingers. How limited are you doing this using your left arm?	Same as question 142
164	Think about pinching and holding an object between your thumb and the side of your index finger, such as holding a key. How limited are you doing this using your right arm?	Same as question 142
165	Think about pinching and holding an object between your thumb and the side of your index finger, such as holding a key. How limited are you doing this using your left arm?	Same as question 142
166	Think about grasping a large object like the lid of a 2-pound jar of mayonnaise with the tips of the fingers hard enough to pick the jar up or open the lid. How limited are you doing this using your right arm?	Same as question 142

continued

Nr.	Questions	Answer options
167	Think about grasping a large object like the lid of a 2-pound jar of mayonnaise with the tips of the fingers hard enough to pick the jar up or open the lid. How limited are you doing this using your left arm?	Same as question 142
168	Think about using your fingers to manipulate objects, such as holding a coin and turning it over and over with your fingers. How limited are you doing this using your right arm?	Same as question 142
169	Think about using your fingers to manipulate objects, such as holding a coin and turning it over and over with your fingers. How limited are you doing this using your left arm?	Same as question 142
170	Think about pressing something with the tip of your index finger (not knuckle) such as dialing a touch-tone phone or ringing a doorbell. How limited are you doing this using your right arm?	Same as question 142
171	Think about pressing something with the tip of your index finger (not knuckle) such as dialing a touch-tone phone or ringing a doorbell. How limited are you doing this using your left arm? (ABILHAND) Describe for the following activities how well you have been able to implement these in the past 3 months, WITHOUT support of other people or assistive devices	Same as question 142
172	Taking the cap off a bottle	1 = Impossible 2 = Difficult 3 = Easy 4 = I do not know
173	Cutting my nails	Same as question 174
174	Buttoning up a shirt	Same as question 174
175	Fastening the zipper of a jacket	Same as question 174
176	Turning a key in a keyhole	Same as question 174
177	Fastening a snap (e.g., from jacket or bag)	Same as question 174
178	Opening a pack of chips	Same as question 174
179	Opening a pack of biscuits	Same as question 174
180	Inserting a key in keyhole	Same as question 174
181	Turning off a tap	Same as question 174
182	Turning on a tap	Same as question 174
183	Filling a glass with water	Same as question 174
184	Sharpening a pencil	Same as question 174
185	Opening a lunch box	Same as question 174
186	Squeezing toothpaste onto a toothbrush	Same as question 174
187	Spreading butter on a slice of bread	Same as question 174
188	Opening a toothpaste tube	Same as question 174
189	Counting banknotes	Same as question 174
190	Dealing cards	Same as question 174
191	Unwrapping a chocolate bar	Same as question 174
192	Drying my hands	Same as question 174
193	Washing my hands	Same as question 174
194	Eat with a spoon	Same as question 174
195	Use fork and knife	Same as question 174
196	Drink a glass of water (without straw)	Same as question 174
197	Use keyboard of computer	Same as question 174
198	Which 5 ABILHAND items (question 174–199) are most important to you?	
199	What are the most important problems you encounter in daily life due to limitations in arms and or hands in order of importance?	Open question

continued

Nr.	Questions	Answer options
200	Do you go to school or attend other classes?	1 = No 2 = Yes
201	Do you encounter limitations in the arms and/or hands during school or study? (if question 202 = 2)	1 = No 2 = A little inconvenience 3 = Regular inconvenience 4 = Severe inconvenience 5 = Proper participation of the education is impossible due to limitations in arms/hands
202	Which activities at school are limited? (if question 203 = 2–6)	Open question
203	What is the highest education you have finished until now?	1 = Primary school 2 = Secondary school 3 = College 4 = University 5 = Special education
204	Do you work internships and volunteering work included?	1 = No 2 = Yes
205	What kind of work do you do more than one is possible? (if question 206 = 2)	Open question
206	Do you suffer from limitations in your arms and/or hands in carrying out your work? (if question 206 = 2)	1 = No 2 = A little inconvenience 3 = Regular inconvenience 4 = Severe inconvenience 5 = Proper participation of the education is impossible due to limitations in arms/hands
207	Which activities are limited? (if question 208 = 2–6)	Open question
208	Are you participating in sport?	1 = No 2 = Yes
209	What kind of sports? (if question 210 = 2)	Open question
210	Do you suffer from limitations in your arms and/or hands in doing your sport? (if question 210 = 2)	1 = No 2 = A little inconvenience 3 = Regular inconvenience 4 = Severe inconvenience 5 = Proper participation of the education is impossible due to limitations in arms/hands
211	Which activities are limited? (if question 212 = 2–6)	Open question
212	Do you have a hobby or leisure time activity?	1 = No 2 = Yes
213	What are your hobbies or leisure time activities? (if question 214 = 2)	Open question
214	Do you suffer from limitations in your arms and/or hands when performing these activities? (if question 214 = 2)	1 = No 2 = A little inconvenience 3 = Regular inconvenience 4 = Severe inconvenience 5 = Proper participation of the education is impossible due to limitations in arms/hands
215	Which activities are limited? (if question 216 = 2–6)	Open question
216	How many friends do you have?	1 = None 2 = 1–5 3 = 6–10 4 = More than 10

continued

Nr.	Questions	Answer options
217	What kind of activities do you do with them?	Open question
218	Do you suffer from limitations in your arms and/or hands during these activities?	1 = No 2 = A little inconvenience 3 = Regular inconvenience 4 = Severe inconvenience 5 = Proper participation of the education is impossible due to limitations in arms/hands
219	Which activities are limited? (if question 220 = 2–6)	Open question
220	Do you have a romantic relationship?	1 = No 2 = Yes
221	Which activities do you like to do together more than one is possible? (if question 222 = 2)	Open question
222	Do you suffer from limitations in your arms and/or hands during these activities? (if question 222 = 2)	1 = No 2 = A little inconvenience 3 = Regular inconvenience 4 = Severe inconvenience 5 = Proper participation of the education is impossible due to limitations in arms/hands
223	Which activities are limited? (if question 224 = 2–6)	Open question
224	Comments	Open question

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