

BACKGROUND

At least 10 percent of the women carrying the dystrophin gene develop symptoms of muscular dystrophy and become manifesting carriers. The symptoms are often invisible to other people and present with varying degrees of reduced muscle strength, reduced endurance, pain and fatigue. Pain and fatigue affect everyday life, functional ability and working capacity but knowledge about this is scarce.

This study investigates the influence of pain and fatigue in Danish manifesting carriers of Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD).

CONCLUSION

Pain and fatigue are very common symptoms in manifesting carriers of dystrophinopathy and have a high impact on general activities, physical activity, concentration, motivation and everyday life. The impact is emphasized by the fact that the majority (78%) of participants in this study were retired or not able to manage a fulltime job. We recommend that pain and fatigue should be included as a factor in the rehabilitation of these women.

FIGURE 1. FAMILILY RELATIONSHIPS

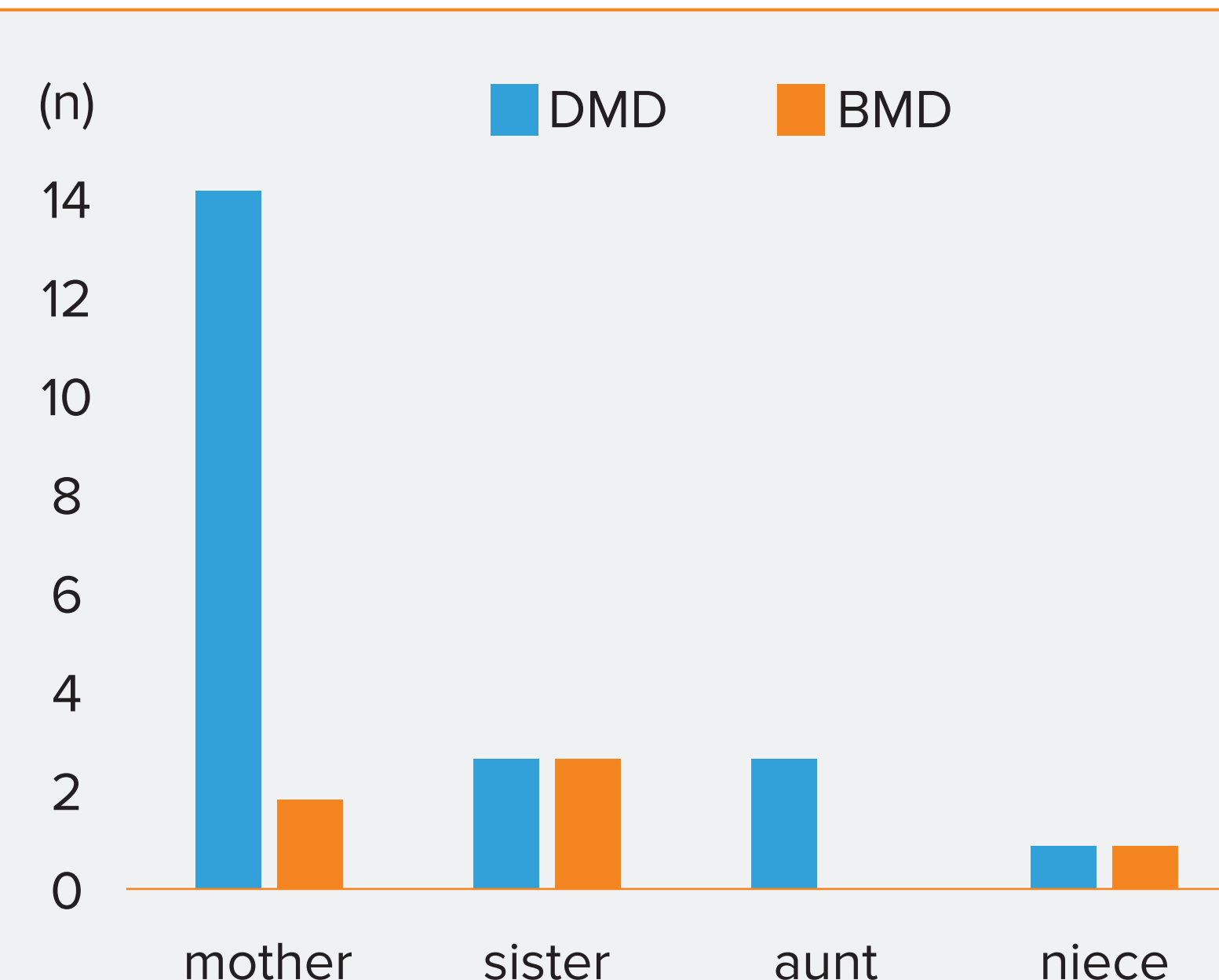
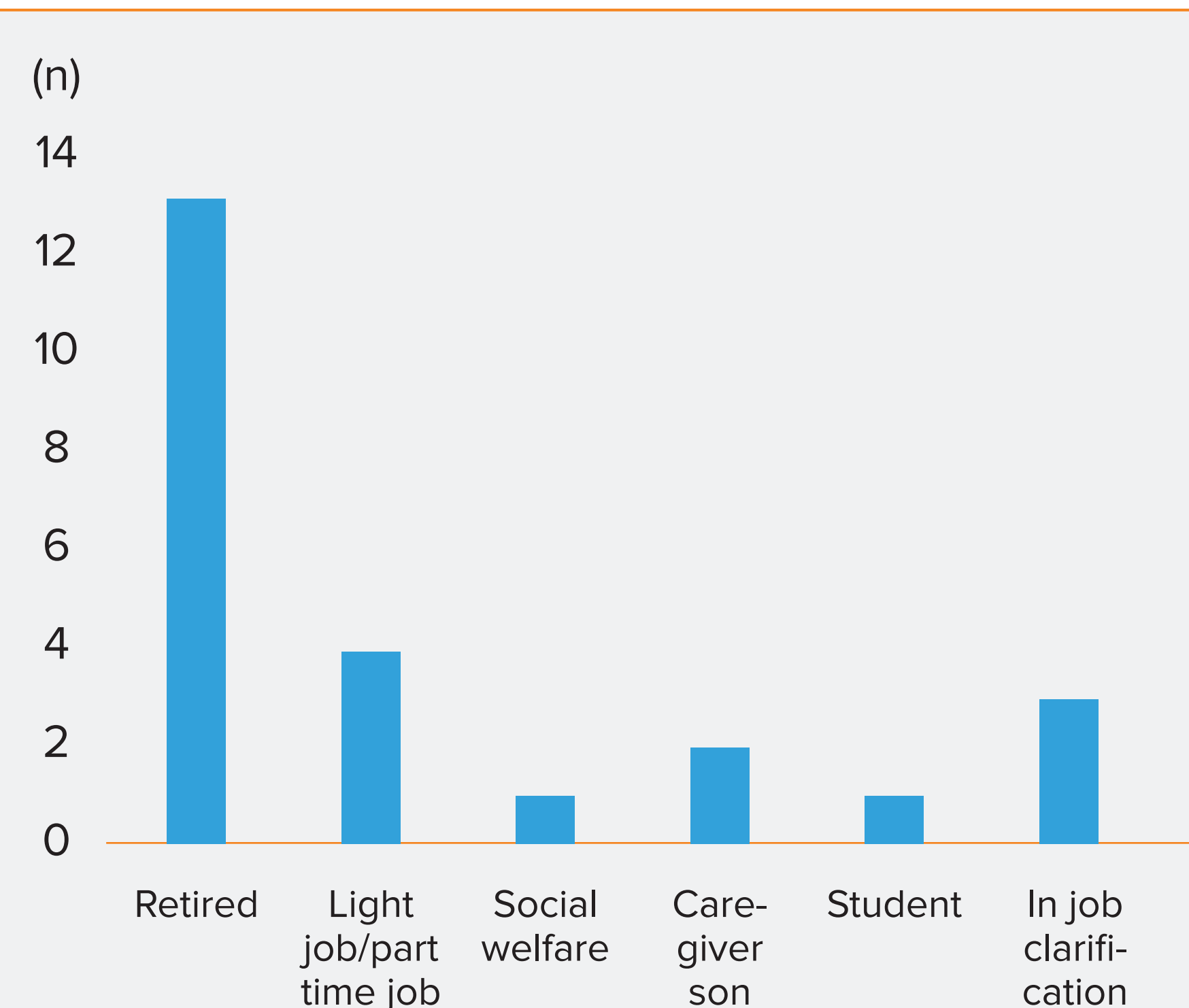


FIGURE 2. WORK



METHODS

The total population of manifesting carriers of DMD and BMD registered with the Danish National Rehabilitation Centre for Neuromuscular Diseases (n=34) were invited to participate in the study. Persons who agreed to participate were asked about their family relation to a patient with DMD/BMD and their association to the labour market. The participants filled in questionnaires on fatigue (Fatigue severity scale (FSS); VAS score; Checklist for Individual Strength (CIS20-R) and pain (Brief Pain inventory). The Questionnaires are presented in a fact box. Descriptive statistics were used to illustrate distribution of data.

FIGURE 3. PRIMARY LOCATION OF PAIN

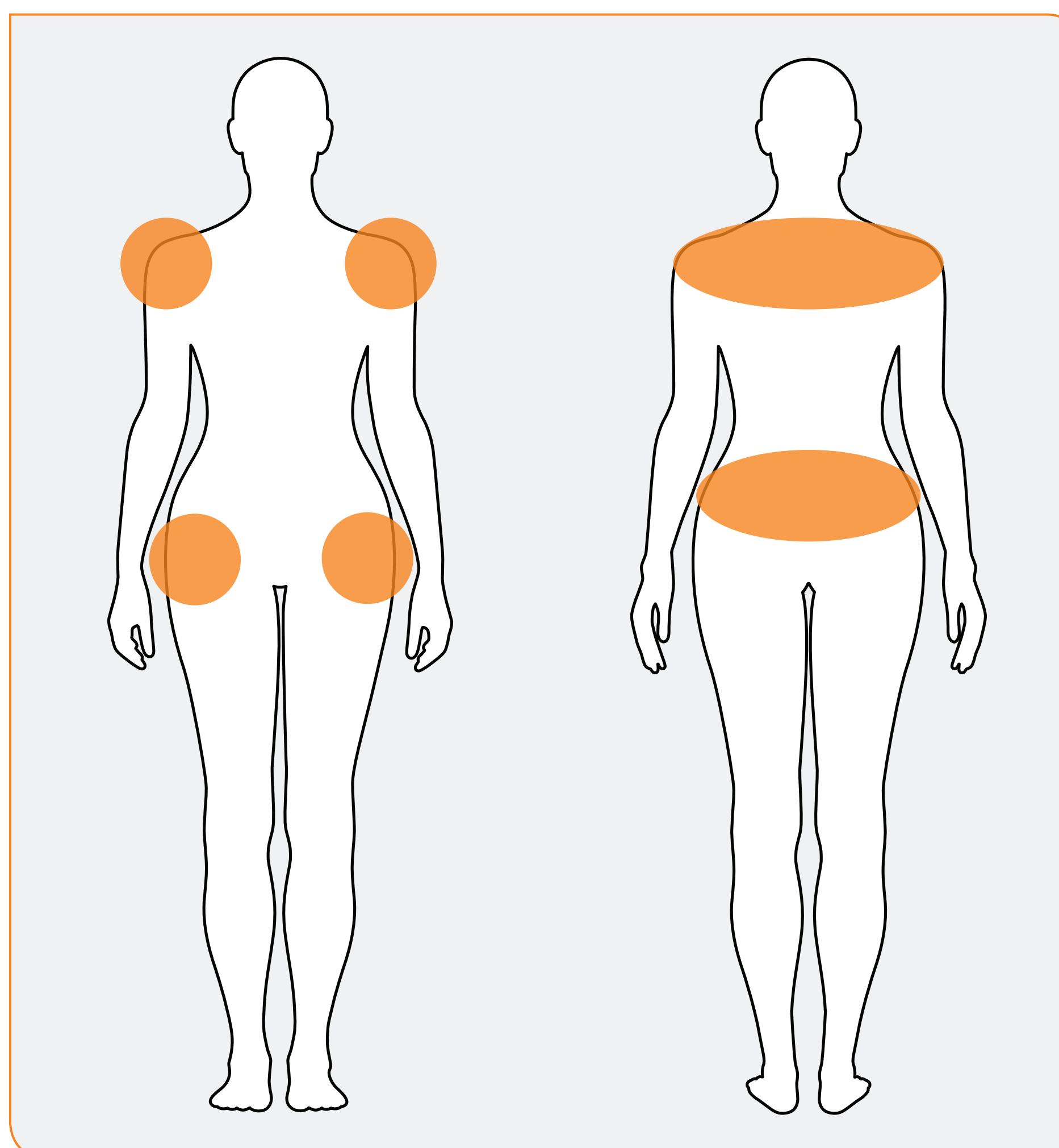
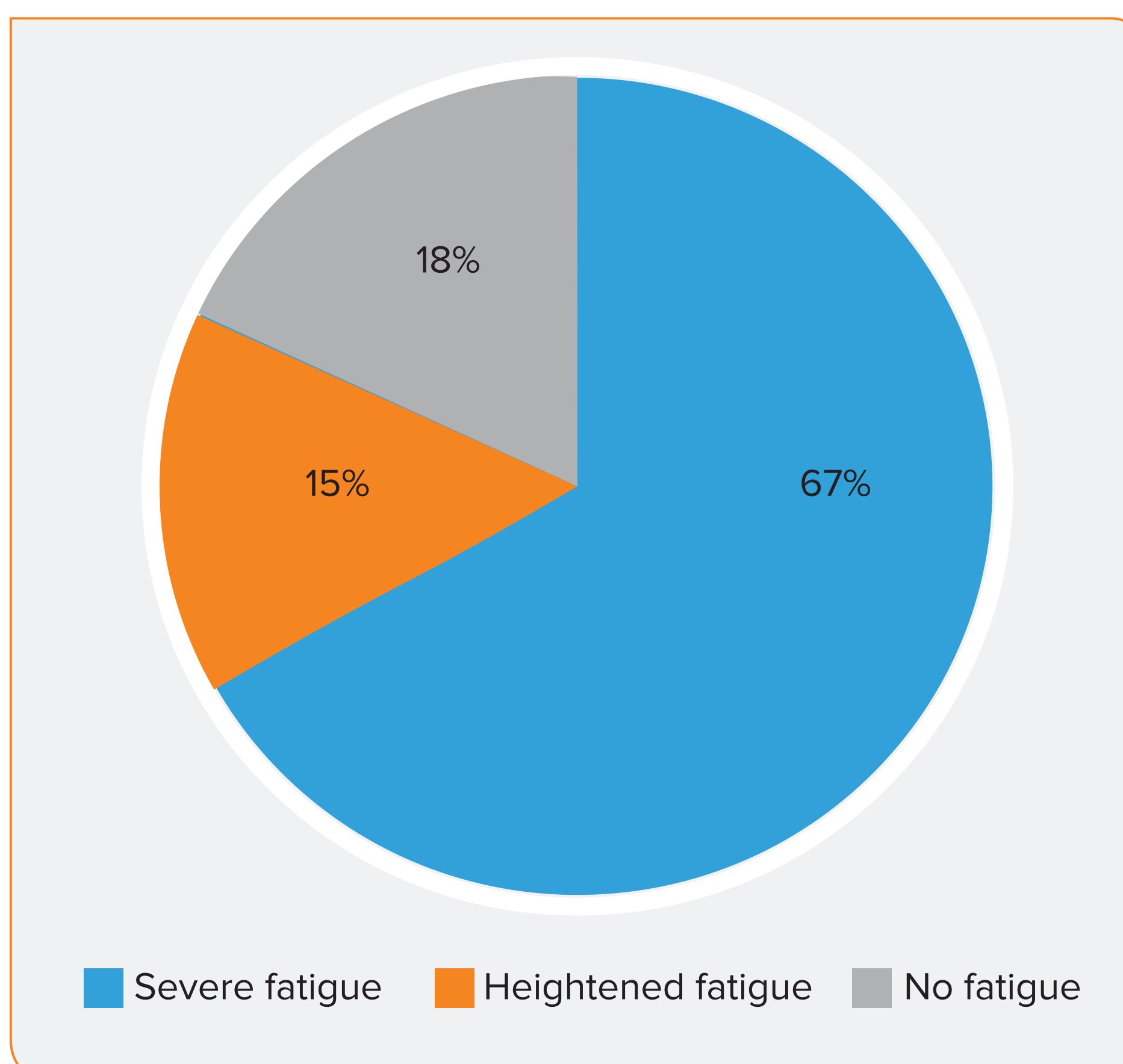


FIGURE 4. CIS-FATIGUE (CIS8R)



RESULTS

Twenty-seven out of 34 persons accepted the invitation. Mean age was 55 years (range 24-74 years).

17/27 (63%) had a genetic diagnosis, the rest was diagnosed based on increased CK or muscle biopsy.

The family relationship to patients with DMD/BMD were mothers (n=16), sisters (n=6), maternal aunts (n=3) and nieces (n=2), (Fig 1). Twenty-one out of the 27 participants were retired or not able to manage a fulltime job (Fig 2).

Pain was very common. Four participants out of 27 did not report any pain whereas the remaining 23 participants all reported daily pain. Mean score for "pain severity" was 4 (range 0-7) and 57% of the participants had a pain severity score of ≥ 5 . Primary location of pain was muscles in shoulder girdle, hips, upper and lower back (Fig 3). Sixteen participants used regular analgesics and/or physiotherapy to ease their pain. Mean score for "pain interference" was 4 (range 0.5-7); the most affected items were "general activity" and "work", illustrated in Table 1.

Fatigue was common and pronounced. The FSS mean score was 5 (range 1-7), and 14/27 participants had a mean score ≥ 6 indicating severe fatigue. The mean score for intensity of fatigues as scored on VAS was 6.3 (range 0-10).

CIS-Fatigue mean score was 81 (range 27-119), Table 2. The results from the subscale "subjective experience of fatigue" (CIS8R) showed that 82% of the participants suffered from heightened or severe fatigue. (Fig 4)

TABLE 1. BPI – PAIN INTERFERENCE – 7 ITEMS. SCORES REPORTED AS MEAN (RANGE)

Pain interference	Mean (range)
General activities	5 (0-10)
Mood	4 (0-9)
Walking	3 (0-10)
Work	6 (1-10)
Relations with others	3 (0-8)
Sleep	4 (0-10)
Enjoyment of life	3 (0-9)
Total	4 (0.5-7)

TABLE 2. CIS – ASPECTS OF FATIGUE – 4 DIMENSIONS

Subscale	Mean score (range)
CIS fatigue (CIS8R)	39 (10-56)
CIS concentration	18 (5-35)
CIS motivation	13 (4-24)
CIS activity	12 (3-21)
Total CIS20R	81 (27-119)

THE QUESTIONNAIRES

BRIEF PAIN INVENTORY (BPI) (CLEELAND ©2009)

- Is a self-reported assessment that rates pain severity and pain interference.
- Includes two dimensions:
 - pain severity (sensory dimension) with four items (worst, least, average and actual pain) rated on a 0-10 scale from "no pain" to "pain as bad as you can imagine"
 - pain interference in daily activities (reactive dimension) with seven items (general activity, walking, work, mood, enjoyment of life, relations with others, and sleep) rated on a 0-10 scale with 0 =does not interfere and 10 = completely interferes)
- The pain severity score illustrates pain severity on a 0-10 scale.
- The pain interference score illustrates pain interference on a 0-10 scale.

FATIGUE SEVERITY SCALE (FSS) (KRUPP 1989)

- Assesses the self-reported impact of fatigue on daily functioning
- Has nine items, each rated on a 7-point scale, ranking from 1= "strongly disagree" to 7= "strongly agree"
- The FSS score is calculated as the mean of all item scores.
- An FSS score ≥ 4 indicates that fatigues is a problem in daily life, and a score ≥ 5 indicates severe fatigue.
- The scoring of FSS involves a Visual Analogue Scale (VAS) where fatigue is rated on a 100 mm horizontal line with 0 = no fatigue and 100 = fatigue as bad as can be".

CHECKLIST INDIVIDUAL STRENGTH (CIS20-R)

(VERCOULEN ET AL, 1994)

- Quantifies subjective fatigue and related behavioural consequences of fatigue.
- Has 20 statements divided into four subscales (subjective experience of fatigue (CIS8R), concentration, motivation, physical activity).
- Each statement is rated on a -point scale, ranking from 1= "Yes, that is true" to 7 = "No, that is not true".
- CIS20R overall score is calculated as the sum of all items; a higher score reflects greater severity. A cut-point of >76 indicates increased risk of consequences of fatigue.
- CIS-fatigue (CIS8R) is calculated as the sum of eight items; sum scores between 27-35 indicate heightened fatigue, ≥ 35 indicate the presence of severe fatigue.