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Research Report

Recognition of emotions conveyed by facial expression and body postures in myotonic dystrophy (DM)

Sabrina Lenzoni [a,*], Virginia Bozzoni [a], Francesca Burgio [b], Beatrice de Gelder [c], Alexandra Wennberg [a], Annalisa Botta [d], Elena Pegoraro [a] and Carlo Semenza [a,b]

[a] Department of Neurosciences (Padova Neuroscience Center), Università degli Studi di Padova, Italy
[b] IRCCS San Camillo Hospital, Venice, Italy
[c] Brain and Emotion Laboratory, Maastricht University, the Netherlands
[d] Department of Biomedicine and Prevention, Medical Genetics Section, University of Rome Tor Vergata, Rome, Italy

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ABSTRACT

Introduction: Neuromuscular diseases may be of neuropsychological interest insofar as they may affect representations based on embodied cognition theories. Previous studies have shown impaired ability to recognize facial emotions and an association between facial emotion recognition and visuospatial abilities in myotonic dystrophy type 1 (DM1) patients. Here we examined the ability of both DM1 and DM2 patients to recognize emotions expressed by body postures and its relation, and their association with cognitive performance.

Methods: Participants included 34 DM1 patients, 8 DM2 patients, and 24 healthy control subjects. Emotional recognition ability was assessed through two computerized matching tasks (face and bodies). A neuropsychological battery was used to measure cognition in three domains and global cognition. We used univariate and adjusted linear regression models to investigate the association between cognition and emotion recognition performance.

Results: DM patients (combined DM1 and DM2) performed worse on emotional facial expression ($p = .006$) and body posture ($p = .004$) accuracy measures than healthy controls. In linear regression models, DM patients' facial expression accuracy was associated with executive function ($p = .013$) and visuospatial ($p < .001$) z-scores. Body posture accuracy was associated with visuospatial ($p = .001$) and memory ($p = .012$) z-scores. There were no associations among controls or between cognition and reaction time.

Discussion: These findings suggest that impaired emotional recognition among DM patients is also extended to emotions conveyed by body postures. Consistent with embodied

* *Corresponding author*. Department of Neurosciences (Padova Neuroscience Center), Università degli Studi di Padova, Via Giustiniani, 5, 35128, Padova, Italy.
E-mail addresses: sabrinalenzoni@gmail.com (S. Lenzoni), virginia.bozzoni@unipd.it (V. Bozzoni), francescaburgio@hotmail.com (F. Burgio), b.degelder@maastrichtuniversity.nl (B. de Gelder), amvwennberg.unipd@gmail.com (A. Wennberg), botta@uniroma2.it (A. Botta), elena.pegoraro@unipd.it (E. Pegoraro), carlo.semenza@unipd.it (C. Semenza).
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cognition theories, people affected in their body and its movement may have impaired sensorimotor representation in ways that have yet to be fully understood.

1. Introduction

Recent studies have highlighted the relationship between emotion and motor systems in the brain, however, many aspects of this remain elusive. Patients with motor disorders provide a window for better understanding the emotion-motor system interface. Myotonic dystrophy type I (DM1, or Steinert's Disease), is the most frequently inherited type of muscular dystrophy in adults, with a prevalence of 13.5/100,000. DM1 is caused by a $[CTG]_n$ triplet expansion of more than 50 repeats on chromosome 19q13.3 in the untranslated 3′ region of *DMPK* gene; it is a dominant autosomal inheritance. Pathological CTG_n expansion size is classified into three categories: E1 (50–150 CTG), E2 (150–1000 CTG), and E3 (>1000 CTG), which correlate with clinical severity and age of onset (Udd & Krahe, 2012).

Progressive muscular degeneration involves distal limbs, and facial and neck muscles. Rhinolalia, dysarthria, dysphagia, and myotonia are the main neuromuscular symptoms of the disease. DM1, however, is a multisystem condition, with cognitive, cardiac, respiratory, ocular, gastrointestinal, and endocrine involvement (Udd & Krahe, 2012). Central nervous system (CNS) dysfunction in DM1 patients has been widely reported in the literature. MRI studies show white matter abnormalities and global cortical and subcortical atrophy, and hypometabolism of frontal and temporal lobes has been shown with PET imaging (Meola & Sansone, 2007). Previous studies investigating the neuropsychological profile in DM1 have reported attention and executive function deficits, suggesting frontal lobe function impairment, but also memory, language, and visuospatial deficits have been shown (Gaul et al., 2006; Meola et al., 2003; Modoni et al., 2008; Sisitiaga et al., 2010; Okkersen et al., 2017; Sansone et al., 2007; Zalonis et al., 2010; Winblad, Samuelsson, Lindberg, & Meola, 2016; Peric et al., 2015).

The prevalence of myotonic Dystrophy type II (DM2, or Proximal Myotonic Myopathy) prevalence is unknown, because it is frequently underdiagnosed. It is caused by $(CCTG)_n$ expansion in intron 1 of *CNBP* gene (also known as *ZNF9*) in chromosome 3q21.3, also with autosomal dominant inheritance. The threshold size of CCTG repeats for the disease-causing mutation is still unclear, although normal alleles contain fewer than 30 repeats (Udd & Krahe, 2012). Progressive involvement of proximal limb muscles, and, less frequently, facial muscles, and myalgias and myotonia, with cardiac, ocular, and endocrinological involvement are key clinical features of DM2 (Wenninger, Montagnese, & Schoser, 2018). Poorer executive function and memory performance and MRI abnormalities, though usually milder than in DM1 patients, have been described and suggest frontal and temporal lobe dysfunction (Peric et al., 2015).

The focus of the present study is the recognition of emotions conveyed by faces and, critically, bodies in DM (throughout "DM" refers to both DM1 and DM2). Few studies have examined emotion recognition in DM. In 2006, Winblad and colleagues published the first study on emotion recognition in DM1. Their results showed a facial emotion recognition impairment in DM1 patients, that correlated with visuospatial abilities and CTG repeat expansion size. Additional studies also found facial emotion recognition deficits in DM1 patients (Labayru et al., 2018; Takeda, Kobayakawa, Suzuki, Tsuruya, & Kawamura, 2009; Kobayakawa, Tsuruya, Takeda, Suzuki, & Kawamura, 2010). Notably, Labayru et al. (2018) confirmed that DM1 patients have difficulty with facial emotion recognition that correlated with age, while no correlation was found with genetic load. In contrast with previous studies showing Theory of Mind impairments (Kobayakawa, Tsuruya, & Kawamura, 2012; Serra et al., 2016), no difference was found between DM and controls in Theory of Mind and empathy measures.

This limited body of work has only begun to scratch the surface on cognition and emotional recognition among DM patients. Neither emotional functioning in DM2 patients nor the ability to recognize emotions conveyed by body postures in DM has ever been investigated. The present study investigated the facial and body emotion recognition abilities of DM1 and DM2 patients, and the association between emotional processing and cognitive performance.

2. Materials and methods

2.1. Participants

34 DM1 patients, 8 DM2 patients and 24 healthy controls were recruited and, for each participant, written informed consent was obtained before testing. All procedures were approved by the IRCSS San Camillo Ethical Review Committee. All DM patients underwent regular multidisciplinary follow-up, with yearly neurological and cardiological evaluation, pulmonary function assessment, and blood tests.

DM diagnosis was confirmed by molecular analysis performed through PCR amplification of, respectively, CTG (*DMPK* gene, chromosome 19q13.32) and CCTG (ZNF9 gene, chromosome 3q21.3) repeats and subsequent hybridization with radioactively labeled oligonucleotides (CTG_{10} or $CCTG_5$).

Participants with a mini-mental state examination (MMSE; Folstein, Folstein, & McHugh, 1975) score of $\leq$24 were excluded. Participants were also excluded if they had history of psychiatric or somatic illness, other neurological conditions (e.g., autism spectrum disorder), major brain injury, substance abuse disorder, or depression, all assessed for at clinical evaluations.

Neuromuscular evaluation was performed with muscle strength assessment and the Muscular Impairment Rating Scale (MIRS; Mathieu, Boivin, Meunier, Gaudreault, & Begin, 2001). The neuropsychological assessment and the administration of FEAST-N (Facial Expression Action Stimulus Test – Neuropsychological population; FEAST-N; de Gelder, Huis in 't Veld, & Van den Stock, 2015) and BEAST-N (Bodily Expression Action Stimulus Test – Neuropsychological population; BEAST-N; de Gelder & Van den Stock, 2011) computerized tests were completed by a neuropsychologist. BEAST-N and FEAST-N stimuli can be found at www.beatricedegelder.com.

No part of the study procedures and analyses was preregistered prior to the research being conducted. We reported how we determined our sample size, all data exclusions (if any), all inclusion/exclusion criteria, whether inclusion/exclusion criteria were established prior to data analysis, all manipulations, and all measures in the study.

2.2. *Neuropsychological battery*

The neuropsychological battery included eleven tests in three cognition domains. Domains were determined with *a priori* knowledge and confirmed with principal component analysis. This approach was modeled on previous studies that have used similar approaches (e.g., Roberts et al., 2008). The domains were assessed with the following tests:

A) Executive function and attention: Raven Progressive Matrices (Raven, 1958; Carlesimo et al., 1996), Phonetic fluency (Novelli et al., 1986), Digit span backward (Orsini et al., 1987), Stroop Color Word Test (Caffarra, Vezzadini, Dieci, Zonato, & Venneri, 2002b), and Trail Making Test (TMT)-A and B (Tombaugh, 2004; Mondini et al., 2003).
B) Visuospatial abilities: Rey–Osterrieth Complex Figure Test – immediate copy and delayed recall (Caffarra, Vezzadini, Dieci, Zonato, & Venneri, 2002a; Osterrieth, 1944)
C) Memory: Digit Span Forward (Orsini et al., 1987) and Rey Auditory Verbal Learning Test – immediate and delayed recall (Rey, 1958; Novelli et al., 1986)

2.3. *Recognition of emotions*

To measure recognition of emotions expressed by the face and the body we used the FEAST-N and BEAST-N, respectively; the shortened versions were utilized to minimize participant burden.

FEAST-N, the subtest of the Facial Emotion Matching test, was used to measure facial recognition ability. The task is composed of 60 trials, each presenting 3 faces, one at the top (sample stimulus) and two at the bottom (target and distractor) of the screen. Subjects were asked to observe the pictures and to discriminate which one between the faces at the bottom expressed the same emotion as the one expressed by the face on the top. Faces expressed six basic emotions: happiness, sadness, fear, anger, surprise, and disgust. For each emotion, there were 10 randomly-ordered trials.

BEAST-N, the subtest Body Emotion Matching, was used to measure body emotion recognition ability using the same format as FEAST-N, but showing body posture instead of faces. The task is composed by 48 trials, each presenting three bodies with a covered face. Bodies express four basic emotions: happiness, sadness, fear, and anger. For each emotion, there were 12 randomly-order trials.

Both FEAST-N and BEAST-N were preceded by 3 practice trials, each one followed by a positive or negative feedback that were not presented in the experiment. Reaction times (RTs) and accuracy were measured as indices of performance.

2.4. *Statistical analysis*

We compared participant characteristics using Kruskal–Wallis or Fisher's exact tests for continuous and dichotomous variables, respectively. We chose nonparametric methods, because data were not normally distributed. For each cognitive domain (attention/executive function, memory, visuospatial ability), we calculated a z-score for each participant and a global cognitive z-score. FEAST-N and BEAST-N mean accuracy and reaction time scores were natural log transformed to create a more normal distribution. Moreover, we z-scored FEAST-N and BEAST-N reaction time scores to create a more normal distribution and aid in interpretation for linear regression models.

We used linear regression models adjusted for age and sex to examine the association between z-scored cognitive domains (independent variable) and FEAST-N and BEAST-N score (dependent variables). We additionally used linear regression models to investigate the association between diagnosis (independent) and FEAST-N and BEAST-N score (dependent variables), and further used the Fisher transformation to determine whether the differences between controls and DM patients was significantly greater in bodies as compared to faces accuracy measures.

Analyses were completed for all participants and, then, due to our *a priori* hypothesis that DM patients fundamentally differ from controls, we stratified analyses by participant group. We additionally utilized multivariable models specifying disease parameters (i.e., onset, duration, MIRS score, and expansion) as the independent variable and FEAST-N or BEAST-N scores as the dependent variable, to evaluate these potential associations among DM1 patients. All analyses were completed using Stata version 13.0 (StataCorp, College Station, TX).

3. Results

Patient characteristics, cognitive test scores, and FEAST-N and BEAST-N scores are presented in Table 1. DM1 patients were more likely to be male and had lower MMSE scores. Additionally, DM patients had lower FEAST-N and BEAST-N accuracy mean scores. DM1 patients had a median disease duration of 21 years (IQR 12.5, 30). Approximately two-thirds of DM1 participants (61.8%) had adult onset, 14.7% (N = 5) had congenital disease, 20.6% (N = 7) had childhood onset, and 2.9% (N = 1) had late/asymptomatic onset.

Table 1 – Participant characteristics, median (IQR) or N (%).

	All	Controls	DM1	DM2	*p*
	N = 66	N = 24	N = 34	N = 8	
Age	45 (33, 56)	42.3 (33, 56)	44.5 (33, 54)	62.5 (45.5, 64)	.094
Male	33 (50)	11 (46)	21 (62)	1 (13)	.039
Education	13 (11, 14)	13 (10.5, 15)	13 (11, 15)	11 (9.5, 12.5)	.118
MMSE	29 (28, 30)	30 (29, 30)	29 (28, 30)	30 (29.5, 30)	.004
Z Executive Function	.04 (−.22, .30)	.13 (−.11, .30)	.02 (−.24, .26)	−.04 (−.50, .45)	.474
Z Visuospatial	.18 (−.30, .60)	.37 (.01, .78)	−.06 (−.40, .44)	.38 (−.50, .70)	.054
Z Memory	−.43 (−.98, .05)	−.22 (−.74, .17)	−.54 (−1.18, −.22)	−.71 (−1.08, −.16)	.214
Z Global	−.03 (−.36, .16)	.04 (−.21, .39)	−.09 (−.53, .08)	−.10 (−.61, .26)	.206
FEAST-N Accuracy, mean	.88 (.85, .92)	.92 (.88, .93)	.85 (.82, .90)	.88 (.83, .90)	.006
Anger	.9 (.9, 1)	1 (.9, 1)	.9 (.8, 1)	.9 (.85, .95)	.067
Disgust	.9 (.9, 1)	.95 (.9, 1)	.9 (.8, 1)	.9 (.85, .95)	.203
Fear	.6 (.6, .7)	.7 (.6, .7)	.6 (.6, .7)	.7 (.55, .75)	.166
Happiness	1 (.9, 1)	1 (1, 1)	1 (.9, 1)	1 (.9, 1)	.751
Sadness	.9 (.8, .9)	.9 (.8, .9)	.9 (.8, .9)	.9 (.75, .95)	.830
Surprise	1 (.9, 1)	1 (1, 1)	.9 (.8, 1)	.9 (.9, 1)	.004
FEAST-N reaction time (msec), mean	4562 (3432, 6061)	4605 (3497, 5373)	4410 (3015, 6430)	4644 (4138, 5022)	.991
Anger	4553 (3163, 6366)	4458 (3124, 5553)	4513 (3139, 6943)	4873 (4013, 5384)	.867
Disgust	4381 (2925, 5513)	4037 (2978, 5027)	4689 (2925, 5730)	4799 (3546, 5211)	.794
Fear	5312 (3910, 6607)	5423 (4480, 6252)	5265 (3558, 8309)	5009 (4310, 5753)	.717
Happiness	3312 (2279, 4066)	3125 (2376, 3852)	3460 (2257, 4157)	3447 (2778, 4265)	.925
Sadness	4643 (3475, 6223)	5252 (3718, 6232)	3991 (3308, 6323)	5349 (4227, 5645)	.660
Surprise	4494 (3371, 5896)	3979 (3257, 5663)	4906 (3371, 6238)	4191 (3999, 5028)	.590
BEAST-N Accuracy, mean	.93 (.90, .96)	.96 (.92, .98)	.92 (.85, .94)	.94 (.92, .98)	.004
Anger	.92 (.83, 1)	1 (.92, 1)	.92 (.75, .92)	.92 (.83, 1)	.002
Fear	.83 (.75, .92)	.92 (.75, 1)	.83 (.75, .92)	.92 (.83, .92)	.051
Happiness	1 (.92, 1)	1 (.92, 1)	.92 (.92, 1)	1 (.92, 1)	.186
Sadness	1 (1, 1)	1 (1, 1)	1 (1, 1)	1 (1, 1)	.369
BEAST-N reaction time (msec), mean	3826 (2988, 4681)	3911.5 (3107.0, 4515.5)	3832.1 (2677.4, 5014.9)	3351.6 (2123.9, 5108.8)	.783
Anger	4413 (3250, 5556)	4731 (3681, 5551)	4299 (3001, 5674)	4060 (2395, 5562)	.544
Fear	4709 (3398, 5625)	4256 (3600, 5544)	4847 (3134, 6166)	4515 (2641, 6162)	.758
Happiness	3184 (2195, 4285)	3587 (2449, 4445)	3042 (2078, 4153)	2964 (2006, 3704)	.746
Sadness	2780 (2210, 3582)	2628 (2224, 3023)	3104 (2302, 3882)	3041 (1451, 3619)	.313

DM1 patient median CTG_n expansion size was 399 (IQR 145, 715; range 74–2290). Additionally, they had a median MIRS score of 4 (IQR 4, 4). Four was by far the most common score in this sample (obtained in 27/34 cases), thus there was limited variation. These data were available only for the DM1 group.

Although DM was associated with poorer performance on measures of both bodies [beta (B) = -.08, 95% confidence interval (CI) −.14, −.02, $p = .011$] and faces (B = −.05 95% CI -.09, −.02, $p = .007$), the difference in BEAST-N accuracy was not significantly greater than the difference in FEAST-N accuracy between controls and DM patients ($p = .897$). For individual emotions, DM patients had lower FEAST-N scores for surprise and lower BEAST-N anger scores.

In linear regression models adjusted for age and sex, better performance in the z-scored cognitive domains of executive function (B = .10, 95% CI .03, .17), visuospatial ability (B = .07, 95% CI .04, .09), and memory (B = .05, 95% CI .01, .08) were associated better FEAST-N accuracy scores among all participants (Table 2). Additionally, global cognitive z-score was associated with better FEAST-N accuracy (B = .09, 95% CI .04, .14). In stratified analyses, these results were significant only among DM patients, among whom better performance in the executive function (B = .12, 95% CI .03, .22) and visuospatial domains (B = .07, 95% CI .03, .10) was significantly associated with better FEAST-N accuracy score.

Additionally, higher z-scores in executive function (B = .13, 95% CI .04, .22), visuospatial ability (B = .11, 95% 95% CI .07, .15), memory (B = .09, 95% CI .04, .15), and global cognition (B = .13, 95% CI .07, .20) were associated with better BEAST-N accuracy scores among all participants (Table 2). Again, these results were driven by DM patients, among whom visuospatial (B = .11, 95% CI .05, .16), memory (B = .10, 95% CI .02, .18), and global (B = .13, 95% CI .02, .13) z-scores were significantly associated with better BEAST-N accuracy scores. There were no observed associations in the controls. Finally, we did not observe any association between cognitive test performance and FEAST-N and BEAST-N reaction time measures (Table 3).

Among DM1 patients, it was found that disease onset was associated with better FEAST-N accuracy score (B = .07, 95% CI .02, .13) in models adjust for age and sex (Table 4). However, we did not observe any other associations between disease parameters (i.e., onset, duration, MIRS score, and expansion) and FEAST-N and BEAST-N scores. Finally, in sensitivity analyses, we investigated whether excluding DM2 patients from all models changed the observed associations, but found it did not (results not shown).

Table 2 – Association between cognitive domains and FEAST-N AND BEAST-N accuracy scores.

	All		Control		DM	
	B (95% CI)	*p*	B (95% CI)	*p*	B (95% CI)	*p*
FEAST-N						
Z Executive Function	.10 (.03, .17)	.005	−.04 (−.10, .02)	.223	.12 (.03, .22)	.013
Z Visuospatial	.07 (.04, .09)	<.001	−.03 (−.07, .008)	.120	.07 (.03, .10)	<.001
Z Memory	.05 (.01, .08)	.011	.008 (−.02, .04)	.625	.05 (−.0002, .10)	.051
Z Global	.09 (.04, .14)	.001	−.02 (−.08, .03)	.423	.10 (.03, .18)	.009
BEAST-N						
Z Executive Function	.13 (.04, .22)	.007	.03 (−.04, .10)	.418	.11 (−.03, .26)	.114
Z Visuospatial	.11 (.07, .15)	<.001	.02 (−.03, .07)	.420	.11 (.05, .16)	.001
Z Memory	.09 (.04, .15)	.001	.03 (−.009, .06)	.130	.10 (.02, .18)	.012
Z Global	.13 (.07, .20)	<.001	.05 (−.02, .11)	.127	.13 (.02, .23)	.022

Models adjusted for age and sex.
B, beta coefficient; CI, confidence interval.

Table 3 – Association between cognitive domains and z-scored FEAST-N and BEAST-N reaction time.

	All		Control		DM	
	B (95% CI)	*p*	B (95% CI)	*p*	B (95% CI)	*p*
FEAST-N RTs						
Z Executive Function	.28 (−.02, .58)	.069	.37 (−.13, .87)	.138	.32 (−.09, .73)	.125
Z Visuospatial	−.06 (−.18, .07)	.346	−.06 (−.42, .31)	.753	−.05 (−.21, .11)	.540
Z Memory	.02 (−.13, .18)	.749	.14 (−.14, .43)	.302	−.02 (−.23, .18)	.814
Z Global	.06 (−.18 .31)	.606	.21 (−.27, .69)	.366	.05 (−.29, .39)	.748
BEAST-N RTs						
Z Executive Function	.05 (−.27, .36)	.768	−.10 (−.57, .38)	.673	.13 (−.35, .62)	.583
Z Visuospatial	−.06 (−.19, .06)	.301	−.16 (−.46, .14)	.290	−.07 (−.24, .10)	.420
Z Memory	.01 (−.14, .16)	.883	.14 (−.10, .39)	.236	−.04 (−.25, .17)	.709
Z Global	−.08 (−.22, .16)	.494	.01 (−.42, .45)	.951	−.14 (−.52, .23)	.438

Models adjusted for age and sex.
B, beta coefficient; CI, confidence interval.

Table 4 – Association between disease parameters and emotion recognition scores.

	Onset		Duration		MIRS		Expansion	
	B (95% CI)	*p*	B (95% CI)	*p*	B (95% CI)	*p*	B (95% CI)	*p*
FEAST-N Accuracy Mean	.07 (.02, .13)	.015	−.003 (−.007, .001)	.130	.05 (−.01, .11)	.128	−.003 (−.10, .10)	.946
BEAST-N Accuracy Mean	.08 (−.02, .18)	.112	−.001 (−.008, .006)	.694	.003 (−.10, .11)	.950	.01 (−.15, .17)	.897
FEAST-N RT Mean	−.19 (−.43, .04)	.106	.01 (−.004, .03)	.148	.03 (−.22, .28)	.821	.11 (−.27, .49)	.569
BEAST-N RT Mean	−.05 (−.30, .19)	.665	.0009 (−.02, .02)	.912	.02 (−.23, .27)	.867	.20 (−.17, .58)	.274

Models adjusted for age and sex.
B, beta coefficient; CI, confidence interval.

The conditions of our ethics approval do not permit public archiving of anonymized study data. Readers seeking access to the data should contact the lead author Sabrina Lenzoni or the local ethics committee. Access will be granted to named individuals in accordance with ethical procedures governing the reuse of sensitive data. Specifically, requestors must meet the following conditions to obtain the data: clearance by IRCSS San Camillo Ethical Review Committee.

4. Discussion

Measures of emotion recognition provide crucial information about social cognitive functioning (Labayru et al., 2018), and may indeed be in helping DM patients adjust to demands of everyday life as their disease progresses. Previous research has highlighted poor social engagement (Gagnon et al., 2008)

and emotional dysfunction in DM patients, who may be less cooperative and empathetic (Winbald et al., 2016), and may suffer from apathy, marked anxiety, irritability, and mood disorders (Gallais et al., 2015; Antonini et al., 2006; Meola et al., 2003).

Similar to previous studies (Kobayawaka et al., 2010; Takeda et al., 2009; Winblad et al., 2006; Labayru et al., 2018), our results confirm that DM (both DM1 and DM2) patients have impaired facial emotion recognition, and, for the first time, show that the ability to recognize emotions expressed by body postures is also impaired. Additionally, we found that DM2 patients' performance in both tasks does not differ from DM1 scores. No significant difference between FEAST-N and BEAST-N accuracy score was observed, thus suggesting that DM patients' performance was not affected by the type of stimuli expressing different emotions.

Analyzing the differences in detecting single emotions, we found lower scores for faces expressing surprise and bodies expressing anger among DM patients. Previous studies investigating facial emotion recognition reported lower scores not only for anger but also for disgust and fear (Kobayawaka et al., 2010, Takeda et al., 2009; Winblad et al., 2006; Labaryu et al., 2018), Importantly, we found evidence that cognitive test performance is associated with FEAST-N and BEAST-N accuracy scores among all participants. In partial contrast to previous literature (Winblad et al., 2006), facial emotion recognition accuracy was associated with executive function and visuospatial ability. Body emotion recognition accuracy was instead associated with visuospatial ability and memory. However, in stratified analyses, these findings were significant only among DM patients, and not controls. These results remained significant in sensitivity analyses excluding DM2 patients, suggesting that cognition is associated with emotional processing in DM, regardless of type.

Accuracy in recognition for both facial and bodily emotions was not associated with the genetic defect, type of onset and MIRS score among DM1 patients. The fact that CTG repeat expansion size is not associated with facial emotion accuracy is at odds with the findings from Winblad et al. (2006), but consistent with more recent results reported by Labayru et al. (2018). These factors, taken together, seem to indicate that emotion recognition impairment is not associated with disease severity. This negative finding may be because of the limited variability of the MIRS score in our sample.

The impaired ability to recognize emotions of DM patients is also extended, in equal measure, to emotions conveyed by body postures. Indeed, a question of theoretical order could also be addressed in this context. How does a neuromuscular disease influence social cognition? The hypothesis that the core deficit of social dysfunction in this pathology is the emotion recognition impairment was previously introduced by Labayru et al. (2018). Our results show an association between emotion recognition and cognition across multiple cognitive domains, suggesting that the relationship between cognition and emotion recognition is not limited to certain processes.

According to embodied cognition theories emotion recognition may be mediated by the activation of sensorimotor representations and selective muscular recruitment (Damasio, 1999; Barsalou, 2008; Winkielman et al., 2015; Winkielman, Coulson, & Niedenthal, 2018). Previous research has shown the importance of muscular activation for emotion recognition processing, by preventing participants from engaging expression-relevant facial muscles (Niedenthal, Brauer, Halberstadt, & Innes-Ker, 2001; Oberman, Winkielman, & Ramachandran, 2007) or through the temporary inactivation of face sensory areas (Pitcher, Garrido, Walsh, & Duchaine, 2008). Because the core symptom of DM is muscular degeneration affecting facial and limb muscles, facial and bodily emotion recognition impairment in these patients may be explained by a dysfunctional embodied simulation process. This is an alternative hypothesis that has not previously discussed in relation to DM.

Physical body changes caused by neuromuscular disease may lead to reduced mimicry and limited movements, and influence the corresponding sensorimotor representations. The general idea is that emotion expression and recognition networks overlap. The impairment of emotional expressions through non-efficient muscular engagement could lead to a defect in the *reenactment* or *embodied simulation* process (Niedenthal et al., 2007; Winkielman et al., 2015; Winkielman et al., 2018). Previous research has shown an association between emotion recognition and motor impairments in Huntington's disease (Trinkler et al., 2017). In these patients, impaired emotion recognition was associated with electromyography (EMG) impairments and with brain volume differences in pSTS, posterior parietal and somatosensory cortices, as compared to healthy controls.

Overall, these studies support the idea that sensorimotor representations play a crucial role in emotional processing of facial expressions, and that muscular activation can mediate emotion recognition through the non-conscious internal triggering of the corresponding expression. Therefore, muscular impairment, the core deficit of DM pathology, may crucially contribute to the impairment of emotion recognition abilities of these patients. The *reenactment* of emotional body postures was not previously investigated. However, de Gelder (2006) proposed a functional model describing three interconnected networks involved in *Emotional Body Language*. One of these systems is represented by the emotional *body awareness*, subserved by somatosensory cortex, insula, anterior cingulate cortex, and ventromedial prefrontal cortex, responding to EBL stimuli. Additionally, a recent study combining continuous theta burst stimulation and fMRI showed the involvement of inferior parietal lobule and ventral premotor cortex in perception of affective bodies (Engelen et al., 2018). In this framework, sensorimotor representations including body postures may be involved in emotional stimuli processing.

Nevertheless, emotional processing deficits may depend on a more global cognitive dysfunction, considering that we found associations between neuropsychological measures of different cognitive domains and emotion recognition ability. The neuropsychological profile of DM patients is not well defined and we found no associations between emotion recognition and disease parameters, though this may be due to lack of power. Poorer cognitive performance has been shown to be associated with age of onset, longer disease duration, and CTG expansion among DM patients (Perini et al., 1999; Sisitaga et al., 2010; Winblad et al., 2016). This suggests that emotional processing may be independent of disease

severity and, instead may rely on more factors such as muscle weakness and muscular dystrophy, which show less variability across patients.

Moreover, as reported in Winkielman et al., 2018, emotions may involve a cascade of events with somatosensory and motor resources recruited at multiple time points in perception, understanding, experience and production; the somatic engagements would reflect not only reflexive, associative connections, but also strategic re-enactments intended to support specific conceptual operations. One can only speculate at this point about what happens in our DM participants. In a recognition task, face- and body-conveyed emotion stimuli may trigger distinct embodied simulations based on distinct cognitive abilities. Our results may thus reflect a non-conscious attempt to strategic re-enactment, that may not lead to a successful completion of the task.

Our study has multiple strengths, including measurement of emotions expressed by faces and body postures, assessment in multiple cognitive domains, and inclusion of both DM1 and DM2 patients. However, the limitations must also be considered. We did not find any difference when comparing DM1 and DM2 FEAST-N and BEAST-N accuracy scores, but sample characteristics should be considered. DM2 group size is small ($n = 8$) and DM2 patients were older and slightly less educated than DM1 patients. However, the inclusion of DM2 group is novel, and future research should further examine emotion recognition and cognitive abilities in these patients. Although we found evidence for an association between cognition and FEAST-N and BEAST-N accuracy scores, these associations failed to reach significance for the association between memory and faces and executive function and bodies among DM patients. However, the magnitude of the association remained, suggesting that lack of power may have led to the lack of statistically significant findings for these particular associations.

In conclusion, recognition of emotions may entail the use of sensorimotor representations related to body representations in ways that still need to be fully understood. Neuromuscular diseases, such as DM, but, potentially other neuromuscular pathologies, may provide a good model to investigate this issue. These findings can crucially enrich our understanding of emotional processing in this patient population, and can provide interesting insights for neuropsychological rehabilitation strategies in the field of social cognition.

Author contributions

Contributions: SL had full access to all of the data in the study and takes responsibility for the integrity of the data and the accuracy of the data analysis.

Conceptualization: SL and CS.
Data curation: SL, AW, and CS.
Formal analysis: SL and AW.
Investigation: SL, EP, VB, AB and CS.
Methodology: SL, EP, BdG and CS.
Project administration: EP and CS.
Resources: EP and CS.
Software: AW.
Supervision: EP and CS.
Validation: EP and CS.
Visualization: All authors.
Roles/Writing, original draft: SL, AW, VB.
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Open practices

The study in this article earned an Open Materials badge for transparent practices.

Declaration of Competing Interest

Nothing to declare.

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