

BURDEN, PROFESSIONAL SUPPORT, AND SOCIAL NETWORK IN FAMILIES OF CHILDREN AND YOUNG ADULTS WITH MUSCULAR DYSTROPHIES

LORENZA MAGLIANO, MD, PhD,¹ MELANIA PATALANO, MS,¹ ALESSANDRA SAGLIOCCHI, MD,¹ MARIANNA SCUTIFERO, MD,² ANTONELLA ZACCARO, Psych, PhD,² MARIA GRAZIA D'ANGELO, MD, PhD,³ FEDERICA CIVATI, Psych,³ ERIKA BRIGHINA, MD,³ GIUSEPPE VITA, MD,⁴ GIAN LUCA VITA, MD, PhD,⁴ SONIA MESSINA, MD, PhD,⁴ MARIA SFRAMELI, MD,⁴ MARIKA PANE, MD, PhD,⁵ MARIA ELENA LOMBARDO, MD,⁵ ROBERTA SCALISE, MD,⁵ ADELE D'AMICO, MD, PhD,⁶ GIULIA COLIA, MD,⁶ MICHELA CATTERUCCIA, PT,⁶ UMBERTO BALOTTIN, MD,⁷ ANGELA BERARDINELLI, MD,⁸ MARIA CHIARA MOTTA, MD,⁸ CORRADO ANGELINI, MD,⁹ ALESSANDRA GAIANI, MD,¹⁰ CLAUDIO SEMPLICINI, MD,¹⁰ LUCA BELLO, MD,¹⁰ ROBERTA BATTINI, MD, PhD,¹¹ GUJA ASTREA, MD, PhD,¹¹ and LUISA POLITANO, MD²

¹Department of Psychology, Second University of Naples, Viale Ellittico, 31, I-81100 Caserta, Italy

²Cardiomyology and Medical Genetics, Department of Experimental Medicine, Second University of Naples (SUN), Italy

³NeuroMuscular Unit, Department of NeuroRehabilitation, IRCCS "E. Medea", Bosisio Parini (Lc), Italy

⁴Department of Neurosciences, University of Messina, Rome, Italy

⁵Department of Paediatric Neurology, Catholic University, Rome, Italy

⁶Unit of Neuromuscular and Neurodegenerative Diseases, Bambin Gesù Children's Hospital, Rome, Italy

⁷Department of Brain and Behavioural Sciences - Child Neuropsychiatry Unit, University of Pavia, Italy

⁸Child Neuropsychiatry, IRCCS "C. Mondino" Foundation, Pavia, Italy

⁹IRCSS San Camillo, Lido, Venice, Italy

¹⁰Department of Neurosciences, University of Padova, Italy

¹¹Developmental Neuroscience, IRCCS Stella Maris, Pisa, Italy

Accepted 27 October 2014

ABSTRACT: *Introduction:* This study explores burden and social and professional support in families of young patients with muscular dystrophies (MDs) in Italy. *Methods:* The study was carried out on 502 key relatives of 4- to 25-year-old patients suffering from Duchenne, Becker, or Limb-Girdle MD who were living with at least 1 adult relative. *Results:* A total of 77.1% of relatives reported feelings of loss, 74.0% had feelings of sadness, and 59.1% had constraints in leisure activities. Burden was higher among relatives of patients with higher disability and who spent more daily hours in caregiving. Practical difficulties were higher among relatives who perceived lower help in patient emergencies and less practical support by their social network. Psychological burden was higher in those relatives who were unemployed, those with poorer support in emergencies, and those with lower social contacts. *Conclusions:* Caring for patients with MDs may be demanding for relatives even in the early stages of these disorders, especially when social support is poor and the patient's disability increases.

Muscle Nerve 52: 13–21, 2015

Muscular dystrophies (MDs) are rare degenerative diseases that lead to muscle strength loss and pro-

gressive restriction of functional abilities. Although only symptomatic therapies are available for these diseases, improved standards of care have led to a considerable increase in life expectancy.^{1–3} Most patients with MD live at home and receive daily assistance from their relatives.^{4,5}

Research and clinical practice highlight that family involvement in the care of patients with long-term illnesses facilitates patients' adaptations to the disease,^{6,7} improves their participation in therapeutic programs,⁸ and has a positive effect on the clinical response to treatment.⁹ However, this long-term assistance may be very demanding for caregiving relatives, mainly if the availability of professional support is poor.¹⁰

Difficulties experienced by relatives of patients as a consequence of their caregiving role are commonly referred to as "family burden" and are divided into "practical" and "psychological" burdens.¹¹ Practical burden refers to problems such as disruption of family relationships; constraints in social, leisure, and work activities; and financial difficulties. Psychological burden describes the reactions that family members experience, e.g., feelings of loss, sadness, tension, and feeling unable to cope with the situation. A higher prevalence of minor psychiatric disorders has been found in caregivers as a consequence of their long-term stress exposure.^{12–14} However, several studies have outlined the protective effect on family burden of social network and professional support on which relatives believe they can rely. In particular, an Italian national study on 709 key relatives of

This is an open access article under the terms of the Creative Commons Attribution-NonCommercial-NoDerivs License, which permits use and distribution in any medium, provided the original work is properly cited, the use is non-commercial and no modifications or adaptations are made.

Abbreviations: BI, Barthel Index; BMD, Becker muscular dystrophy; DMD, Duchenne muscular dystrophy; FPQ, Family Problems Questionnaire; LGMDs, limb-girdle muscular dystrophies; MD, muscular dystrophy; MD-CS, Muscular Dystrophy Care Schedule; SNQ, Social Network Questionnaire

Key words: caregiving; family burden; muscular dystrophy; professional support; social network

This study was supported by the Telethon UILDM grant GUP10002. We are grateful to Prof. Gabriele Siciliano who facilitated the enrolment of the young adult patients at the Neurological Clinic of the University of Pisa, Italy.

Correspondence to: L. Magliano; e-mail: lorenza.magliano@unina2.it

© 2014 The Authors. *Muscle & Nerve* Published by Wiley Periodicals, Inc. Published online 31 October 2014 in Wiley Online Library (wileyonlinelibrary.com). DOI 10.1002/mus.24503

patients with schizophrenia¹⁵ found that practical burden was significantly higher among the relatives who received poorer social support from professionals, and it was lower among the relatives who relied on a larger and more supportive social network. Another Italian study¹⁶ tested the efficacy of a psycho-educational family intervention for schizophrenia and found that the intervention improved family burden at 6 months and was associated with increased perception of professional support among relatives. Furthermore, a systematic review of studies on interventions for caregivers of patients with a life threatening, incurable illness¹⁷ found that family supportive interventions had a positive impact on caregivers' ability to provide care and led to positive changes in psychological burden. Finally, a review of the efficacy of psychological interventions for families of patients in the terminal phase of their illness¹⁸ reported that supportive interventions may help to reduce short-term caregiver psychological distress.

Available data suggest that family burden in long-term diseases shares some common characteristics as those related to work difficulties and economic burden.¹⁹ However, different pathologies, due to their clinical characteristics and social reactions to them,¹⁰ may dictate specific needs for care and require different therapeutic strategies.²⁰ In Italy, a comparative national study of burden on relatives of adult patients with a mental vs. a physical disease¹⁰ revealed that the consequences of caregiving most frequently reported as always present in all examined groups were constraints in social activities, negative effects on family life, and a feeling of loss. In addition, family burden was found to be significantly higher among relatives of patients with neurological diseases than in other groups.

Unlike mental disorders,^{14,21} cancer,²² and some neurological diseases such as dementia²³ in which burden has been explored extensively, few studies are available on family difficulties in MDs.

The few data available in MDs reveal that financial difficulties^{4,24,25} and constraints of social and work activities are the *practical* consequences of caregiving that are most burdensome for relatives,²⁶ while high stress, depression, and guilt feelings are the *psychological* consequences reported most frequently.²⁶⁻²⁸ In a recent study of the psychological benefits²⁹ in a group of 502 Duchenne MD (DMD), Becker (BMD), or Limb-Girdle MDs (LGMDs) key relatives, our work group reported that, despite the difficulties associated with caregiving, relatives identify valuable benefits in their experience.

In MDs, family burden is influenced significantly by socio-demographic characteristics of the relatives, clinical severity of the MD, and by perso-

nal, social, and professional resources. Burden is higher among mothers,²⁹ unemployed relatives,²⁷ and families at low income levels.^{27,30} Family difficulties are greater among relatives of patients at lower levels of functional abilities,^{30,31} in wheelchairs,²⁷ and on ventilation³² and/or tracheotomy.²⁶ Burden is lower among relatives with adequate coping skills with the illness³¹ and high self-esteem²⁶ and among those with high social support.^{27,30}

However, almost all these studies have several weaknesses, such as small sample size and poor national level representation.^{26,32} Most studies have been carried out in North America,^{4,27} which limits the generalization of their findings, because the burden may vary in relation to health care policies and cultural variables.¹³ Furthermore, these studies have mainly addressed family burden in DMD,^{26,27,30} and scarcely explored it in other MDs.^{31,32} Family constraints likely vary in relation to the type and stage of MD,^{29,30} medical needs of care,^{24,33} and expectations for the future, in both patients and their relatives.²⁴ Finally, little is known about the amount and quality of psychological treatments provided to patients and their relatives^{4,5,24} in routine conditions, and about the impact of these treatments on family burden.

Family involvement in the care of long-term diseases is particularly relevant in Italy, where the national health policy is strongly community oriented.³⁴ In 2012, within the framework of the Telethon-UILDM National Program for Clinical Research in Muscular Diseases, we carried out a national survey on family burden and professional and social support in families of young patients with several forms of MD, including DMD, BMD, and LGMDs. A total of 502 key relatives of 4- to 25-year-old patients with MDs who were enrolled in 8 specialized Italian centers for MDs participated in the survey. Given the large data bank, we planned to report the results in several papers, each focusing on a specific aspect of the complex experience of caregiving in these disorders. In the previous paper based on the above-mentioned survey,²⁹ we tested whether relatives' perceptions of psychological benefits varied in relation to their view of the patient as a valued person, the degree of involvement in care, and the levels of support provided by their social network and professionals. We found that 88% of key relatives stated they had gotten something positive out of the situation, such as personal growth, resilience, and altruism. In particular, positive aspects of caregiving were more recognized by key relatives who were more convinced that the patient was sensitive and who were perceived to receive a higher level of professional help and psychological social support.

A positive correlation between caregivers' acknowledgment of psychological benefits and the mean level of practical difficulties they reported, was also found.

In this study, based on the data bank mentioned above, we investigate in detail the kind of practical and psychological difficulties experienced by the 502 participating caregivers and explore whether their burden differs in relation to socio-demographic and clinical characteristics, and to perceived professional and social network support.

MATERIALS AND METHODS

Design of the Study. The study was carried out in 8 specialized centers for MDs located in Northern (3 centers), Central (3 centers), and Southern Italy (2 centers). In the period January-December 2012, the key relatives (i.e., the relative spending more daily time in contact with the patient and being more involved in his/her care) of patients who met the selection criteria listed below, were contacted consecutively and asked to give informed consent to participate in the study. Because the study protocol did not involve the patients directly, only those patients aged 18–25 were asked for their permission to contact key relatives.

Patient selection criteria were the following: diagnosis of DMD, BMD, or LGMD, confirmed by molecular analysis or muscle biopsy; age between 4 and 25 years; in charge of the participating center for at least 6 months; living with at least 1 adult relative; not suffering from diseases other than those MD-related.

Key relative selection criteria were the following: age between 18 and 80 years; not suffering from illness requiring long-term intensive care.

During a patient's scheduled clinical examination, each enrolled relative was interviewed face-to-face by a trained researcher with regards to: (a) the patient's functional abilities in the previous month, according to Barthel Index (BI)³⁵; (b) the family socio-demographic characteristics and patient's clinical variables by means of schedules designed *ad hoc*; and (c) the treatments and support received by the patient and his/her family by means of the Muscular Dystrophy Care Schedule (MD-CS). In the same interview, the key relative was asked to provide information on his/her caregiver's condition by filling the pen-and-pencil Family Problems Questionnaire (FPQ)¹³ and Social Network Questionnaire (SNQ).¹³ Relatives were not compensated for completion of the assessment.

The study protocol was approved by the Ethics Committee of the Second University of Naples (coordinating center), and by the Local Ethics Committee of each participating Center.

Instrument Descriptions. **Barthel Index (BI).** The BI³⁵ is a scale to assess a patient's degree of independence in daily activities. It provides a global 0–100 score (from 0 “totally dependent” to 100 “totally independent”) based on 10 items which explore the patient's performance in several activities and mobility. The inter-rater reliability in BI scoring of the researchers was tested preliminarily (Cohen kappa coefficient ranging from 1 to 0.90 for 9 BI items and equal to 0.67 for the lasting BI item).

Family Problems Questionnaire (FPQ). The FPQ¹³ is a self-reported tool containing 34 items that explore the caregiver's: (a–b) practical and psychological burdens (items rated on a 4-level scale from 1 “never” to 4 “always”); (c–d) perceived support provided to the family in emergencies concerning the patient by professionals and social network (items rated on a 4-level scale from 1 “not at all” to 4 “completely”); (e) attitude toward the patient (items on a 4-level scale from 1 “not at all” to 4 “completely”). An average subscale score is derived across the valid scores of the items included in each subscale. Furthermore, the FPQ contains additional sections on economic costs and an additional open-ended question that asks key relatives what they suggest to improve care-givers' quality of life. The answers to the open-ended question were grouped by 2 researchers into the following discrete categories: (a) welfare policies; (b) psychological support to users and families; (c) quality of health care; (d) investment in research; (e) information on MD illness; (f) school support; and (g) promotion of awareness campaigns about MDs for the public. FPQ was administered in its entirety, but, as this study is based on the data derived from sections a–d and the open-ended question, the remaining sections of FPQ were not analyzed and reported here. Inter-rater reliability in the use of the above-mentioned categories was measured using 50 randomly selected cases (Cohen kappa value ranging between .96 and 1). The psychometric properties of the FPQ were tested previously.¹³ Cronbach alphas of FPQ subscales a–e in this study sample were found to be consistent with those of previous measurements (alpha values ranging from 0.63 to 0.86).

Social Network Questionnaire (SNQ). The SNQ¹³ is a self-reported tool which includes 15 items that explore the caregiver's: (a) quality and frequency of social contacts; (b–d) perception of practical and psychological support received from social network and the partner. The sections contain items rated on a 4 level scale, from 1 “Never” to 4 “Always”. The psychometric properties of the SNQ have been tested previously.¹³ Cronbach alphas of the SNQ subscales, as measured in this study

Table 1. Characteristics of the 502 patients and their key relatives.

Socio-demographic and clinical variables	Patients (N = 502)	Key relatives (N = 502)
Gender, N (%)		
Males	484 (96.4)	74 (14.7)
Females	18 (3.6)	428 (85.2)
Age, mean (SD) years	12.8 (5.6)	43.4 (7.4)
Marital status, N (%)		
Single	502 (100)	61 (12.1)
Cohabiting/married	0	441 (87.8)
Education, N (%) yes	430 (85.6)*	502 (100)†
Pre-school	50 (11.6)	-
Primary school	90 (20.9)	35 (6.9)
Secondary school	90 (20.9)	184 (36.6)
High school	127 (29.1)	219 (43.6)
University	17 (4.0)	64 (12.7)
Currently employed (adults), N (%) yes	7 (7.4)	264 (52.6)
Relationship with the patient, %		
Mother	-	424 (84.6)
Father	-	70 (14.0)
Other	-	7 (1.4)
Duration of symptoms, mean (SD) years	8.9 (5.5)	-

*School attendance.

†School degree.

sample, were found to be consistent with those of previous measurements (alpha value ranging from 0.68 and 0.79).

Muscular Dystrophy Care Schedule (MD-CS). The MD-CS derives from a similar care schedule customized for mental disorders.¹⁰ It collects information on: (a) the patient's pharmacological therapies in the previous 2 months; (b) the patient's rehabilitative treatments; (c) the patient's and relatives' psycho-educational interventions; and (d) the patient's and relatives' social/welfare support in the previous 6 months. Data collected by the MD-CS were analyzed as "yes/no" variables.

Statistical Analysis. Percentages were calculated for categorical and ordinal variables describing socio-demographic characteristics of patients (gender, marital status, level of education, current employment) and of their key relatives (gender, marital status, level of education, current employment, relationship to the patient). Furthermore, percentages were calculated for patients' clinical features (type of MD, use of wheelchair), relatives' burden (FPQ *a-b* subscale items), and perceived professional and social support (FPQ *c-d* subscale items, and SNQ *a-d* subscale items). Means and standard deviations (SD) were computed for continuous variables describing patient (age) and key relatives (age, daily hours dedicated to patient's care) socio-demographic characteristics, patient

clinical features (BI, length of illness) and relatives' burden and support (FPQ and SNQ *a-d* subscales).

Differences in family burden (FPQ *a-b* subscale mean scores) in relation to the patient and key relative socio-demographic and clinical variables listed above, were explored by analysis of variance. Correlations of family burden (FPQ *a-b* subscale mean scores) with patient and relative socio-demographic characteristics, patient levels of functional abilities (BI global score) and professional and social support (SNQ *a-d* subscale mean scores and FPQ *c-d* subscale mean scores) were explored by Spearman *r* coefficients. Hierarchical multiple regression analyses were performed to explore the simultaneous effects of patients'/relatives' socio-demographic characteristics and of patient clinical variables (Block 1), and of the perception of professional and social support received by families (Block 2) on the practical and psychological burden. Only variables related statistically significantly to family burden in univariate analyses were included in the regressions. Multi-collinearity among the variables included in the regression was checked by variance inflation factors (VFI). In the case of a strong correlation between 2 variables, only 1 was included in the model. Statistical significance was set at $P < 0.01$ for univariate analyses and at $P < 0.05$ for multivariate analyses. Analyses were performed with SPSS 19.0.

RESULTS

Descriptive Results. Of the 504 eligible patients and their key relatives who were contacted consecutively, 502 agreed to participate and were assessed. Most patients were male and were attending school (Table 1). Three hundred thirty-three (66.3%) suffered from DMD, 192 (25.7%) from BMD, and 40 (8.0%) from LGMDs. The mean level of independence in daily activities, measured by the BI, was 68.3 (31.3 SD). One hundred ninety-four patients (38.6%) were in a wheelchair. Three hundred sixty-nine (73.5%) patients took prescribed medications (corticosteroids: 227 [61.5%]; cardiac drugs: 193 [52.3%]; drugs for bone metabolism: 147 [39.8%]; dietary supplements: 118 [31.9%]; gastric drugs: 75 [20.3%]; pulmonary and/or neurological drugs: 9 [2.4%]), and 351 (69.9%) attended rehabilitation programs.

Seventy-two patients (14.3%) received a psycho-educational intervention (52.8% psychological support and 38.9% information on MD treatment) in the 6 months preceding the interview. Furthermore, 331 patients (65.9%) received social/welfare support, mainly (96.4%) consisting of economic benefits, while 52 (15.7%) had a support teacher at school.

The majority of the 502 key relatives were mothers and lived with a partner (Table 1). Almost

Table 2. Practical and psychological burden experienced by relatives of patients with MDs (*N* = 502).

Practical burden	Variables			
	Always <i>N</i> (%)	Often <i>N</i> (%)	Sometimes <i>N</i> (%)	Never <i>N</i> (%)
I have had to wake up during the night	66 (13.2)	40 (8.0)	123 (24.6)	271 (54.2)
I have had to neglect my hobbies and things I like doing in my free time	74 (14.8)	75 (15.0)	146 (29.2)	204 (40.9)
I have had difficulty in going on Sunday outings	31 (7.5)	39 (9.5)	81 (19.7)	261 (63.4)
I found it difficult to have friends at home	10 (2.0)	20 (4.0)	41 (8.2)	429 (85.8)
I found it difficult to meet friends and people I like to spend my leisure time	17 (3.4)	35 (7.0)	91 (18.2)	356 (71.3)
I found it difficult to carry out my usual work or household activities or I had to stay at home from work or school	17 (3.4)	50 (10.0)	157 (31.5)	275 (55.1)
I had to neglect other family members	6 (1.2)	39 (8.1)	136 (28.2)	301 (62.4)
I had difficulty in going on holiday	40 (9.4)	38 (8.9)	60 (14.1)	287 (67.5)
I had economic difficulties	18 (3.6)	29 (5.8)	127 (25.4)	326 (65.2)
<i>Subscale mean score (SD)</i>	1.6 (0.6)			
Psychological burden				
I felt that I would not be able to bear this situation much longer	17 (3.4)	52 (10.5)	180 (36.2)	248 (49.9)
I cried or felt depressed	16 (3.2)	96 (19.2)	259 (51.7)	130 (25.9)
I worry for the future of other family members	36 (7.2)	85 (17.1)	238 (47.8)	139 (27.9)
When I went to a public place with my ill relative, I felt that everyone was watching us	31 (6.3)	38 (7.7)	119 (24.1)	306 (62.0)
I feel guilty because I believe that I or my spouse may have passed on the illness to our relative	51 (10.3)	64 (13.0)	164 (33.3)	214 (43.4)
When I think of how our ill relative was beforehand and how he/she is now, I feel disappointed	140 (28.3)	87 (17.6)	154 (31.2)	113 (22.9)
<i>Subscale mean score (SD)</i>	1.9 (0.6)			

half of the key relatives had received higher education and were employed. They spent on average 5.7 (4.6 SD) daily hours in patient care-giving in the previous 2 months. One hundred fifty-six relatives received educational interventions, including 107 (68.5%) education on clinical and rehabilitative procedures and 84 (53.8%) information on MD treatments. Thirty-four relatives (21.7%) received psychological support, while 46 (9.1%) received support by Family/Patient Associations.

The practical consequences most frequently mentioned by the caregivers concerned neglect of their hobbies (59.1% of relatives) and night-time awakening due to the patient's condition (45.8%, Table 2). In addition, 34.3% of the caregivers stated that, in the previous year, they had had economic difficulties due to the patient's illness.

As far as psychological difficulties, 77.1% of relatives reported feelings of loss, 74.0% stated they cried or felt depressed, and 72.1% worried about the future of other family members (Table 2). Furthermore, 56.6% of relatives felt guilty for having transmitted the illness to their children.

Eighty-two percent of relatives reported feeling completely confident of receiving professional help in a crisis situation, and 63.2% believed they had received adequate information from clinicians on how to cope with the patient's medical emergencies. With regard to social network support, 77.1%

of relatives stated they felt sure they have somebody who would take care of them in case of their own physical illness, and 76.8% reported 2 or more trustworthy friends on whom they could rely. Furthermore, 75.7% believed their friends would definitely help them in the case of the patient's emergencies (Table 3).

Recommendations on how to improve their condition were given by 291 (57.9%) responders. In particular, 42.9% of caregivers suggested improvements in the quality of care ("implementing health centers covering all range of clinical and social aspects of MDs"), and 33.3% suggested improvements in welfare policies ("simplify bureaucratic procedures," "removal of architectural obstacles," and "economic facilities in transfer"), while 30.2% recommended provision of psychological support to families and patients ("to not abandon the families" and "meeting with other families who are dealing with MDs in the presence of psychologists"), and 18.9% suggested increased investment in research on rare diseases. Other recommendations included providing information on MD to families (8.6%), school support to patients (5.8%), and promotion of sensitization initiatives about MDs for the public (5.5%).

Relationships of Family Burden with Socio-demographic and Clinical Variables. Both practical and psychological burden were higher among unemployed relatives

Table 3. Perceived support received from social network and professionals by relatives of patients with MDs (N = 502).

	Variables			
	N (%) More than 3 persons	N (%) 2 or 3 persons	N (%) 1 person	N (%) None
Social contacts				
How many trustworthy friends do you have outside the family?	102 (20.5)	249 (50.0)	76 (15.3)	71 (14.3)
How many relatives do you consider as trustworthy friends?	154 (30.7)	231 (46.1)	68 (13.6)	48 (9.6)
I called up or met friends or relatives not living with me	Always 56 (11.2)	Often 198 (39.4)	Sometimes 212 (42.2)	Never 36 (7.2)
Friends and relatives not living with me called me up or came to see me	56 (11.2)	210 (41.8)	216 (43.0)	26 (5.2)
<i>Subscale mean score (SD)</i>	2.7 (0.6)			
Perceived practical support	Always	Often	Sometimes	Never
Had I been ill, there would have been somebody to take care of me	273 (54.4)	114 (22.7)	87 (17.3)	28 (5.6)
When I needed help for my housework, I easily found somebody who gave me a hand	139 (32.8)	89 (21.0)	119 (28.1)	77 (18.2)
I easily found somebody who went on demands for me when I could not go myself	124 (27.1)	101 (22.1)	168 (36.8)	64 (14.0)
<i>Subscale mean score (SD)</i>	2.9 (0.8)			
Perceived psychological support	Always	Often	Sometimes	Never
When I felt anxious or worried, I had somebody who reassured me	110 (23.1)	116 (24.3)	194 (40.7)	57 (11.9)
When pleasurable things happened to me, there was somebody I could share them with	157 (32.1)	155 (31.7)	152 (31.1)	25 (5.1)
I had a trustworthy friend or relative to talk to about my own problems	139 (27.7)	161 (32.1)	151 (30.1)	50 (10.0)
Friends and relatives not living with me talked with me about their own problems	23 (4.6)	167 (33.3)	264 (52.6)	48 (9.6)
<i>Subscale mean score (SD)</i>	2.6 (0.6)			
Perceived support by partner	Always	Often	Sometimes	Never
My partner understands me	135 (29.5)	198 (43.3)	109 (23.8)	15 (3.3)
My partner helps me	195 (42.7)	149 (32.6)	76 (15.3)	71 (14.3)
<i>Subscale mean score (SD)</i>	3.1 (0.7)			
Perceived social support in emergencies	Completely	Quite confident	A little	Not at all
When things are going particularly badly, I can ask relatives or friends to help or support our family	123 (24.6)	160 (32.1)	111 (22.2)	105 (21.1)
*, I am confident that help will be provided by my relatives or friends	254 (50.6)	126 (25.1)	83(16.5)	39 (7.8)
<i>Subscale mean score (SD)</i>	2.9 (0.8)			
Perceived professional help in emergencies	Completely	Quite confident	A little	Not at all
I have received information from professionals on what to do *	236 (48.5)	72 (14.8)	45 (9.2)	134 (27.5)
*, I am confident that help will be provided immediately by professionals	193 (38.8)	216 (43.4)	73 (14.7)	16 (3.2)
Doctors who are taking care of my ill relative are helping him/her	151 (30.4)	252 (50.8)	82 (16.5)	11 (2.2)
<i>Subscale mean score (SD)</i>	3.0 (0.7)			

*In an emergency concerning my ill relative.

vs. employed ones (practical burden, mean [SD]: 1.7 [0.6] vs. 1.5 [0.6], $F = 12.0$, df 1,500, $P < 0.001$; psychological burden, mean [SD]: 2.1 [0.6] vs. 1.8 [0.5], $F = 21.0$, df 1,500, $P < 0.0001$), and among relatives who did not live with a partner compared with those who did ([practical burden, mean [SD] 1.5 [0.6] vs. 1.9 [0.8], $F = 23.8$, df 1,500, $P < 0.0001$; psychological burden, mean [SD]: 1.9 [0.6] vs. 2.0 [0.6], $F = 5.0$, df 1,500, $P < 0.05$). Furthermore,

practical burden correlated positively with the daily time spent in taking care of the patient ($r = 0.43$, $P < 0.0001$).

Family burden was higher in relatives of patients who were not attending school compared with those who were (practical burden, mean [SD] 1.8 [0.7] vs. 1.5 [0.6], $F = 10.8$, df 1,500, $P < 0.001$; psychological burden, mean [SD]: 2.1 [0.6] vs. 1.9 [0.6], $F = 9.0$, df 1,500, $P < 0.005$). Moreover,

burden increased in relation to the patient's level of functional disability (practical burden: Spearman $r = -0.65$, $P < 0.0001$; psychological burden: $r = -0.35$, $P < 0.0001$), and it was higher among relatives of patients who were receiving drug treatments (practical burden, mean [SD]: 1.7 [0.6] vs. 1.3 [0.5], $F = 31.8$, df 1,500, $P < 0.0001$; psychological burden: 2.0 [0.6] vs. 1.7 [0.5], $F = 25.1$, df 1,500, $P < 0.0001$), attended rehabilitative interventions (practical burden, mean [SD]: 1.7 [0.6] vs. 1.2 [0.4], $F = 92.3$, df 1,500, $P < 0.0001$; psychological burden: 2.0 [0.6] vs. 1.7 [0.5], $F = 26.1$, df 1,500, $P < 0.0001$), and/or received welfare support (practical burden, mean [SD]: 1.7 [0.6] vs. 1.3 [0.4], $F = 67.1$, df 1,500, $P < 0.0001$; psychological burden: 2.0 [0.6] vs. 1.8 [0.5], $F = 16.7$, df 1,500, $P < 0.0001$). Furthermore, burden was higher in relatives of patients with DMD compared with those of patients with LGMD and BMD, respectively (practical burden, mean [SD]: 1.7 [0.6] vs. 1.2 [0.4] vs. 1.4 [0.5], $F = 39.8$, df 2,499, $P < 0.0001$; psychological burden: 2.0 [0.6] vs. 1.9 [0.5], vs. 1.7 [0.5], $F = 19.2$, df 2,499, $P < 0.0001$). Finally, practical burden correlated positively with patient age (Spearman $r = 0.23$; $P < 0.0001$) and years of illness ($r = 0.28$; $P < 0.0001$).

Relationships of Family Burden with Perceived Professional and Social Support. Practical and psychological burden were higher in relatives who were perceived to receive lower levels of practical and psychological support by their social network (practical burden: $r = -0.28$, $r = -0.17$, $P < 0.0001$; psychological burden: $r = -0.18$, $r = -.23$, $P < 0.0001$) and had fewer social contacts (practical burden: $r = -0.19$, $P < 0.0001$; psychological burden: $r = -0.26$, $P < 0.0001$). Furthermore, burden was higher among relatives who perceived that they had lower levels of support for the patient's emergencies from their social network (practical burden: $r = -0.47$, $P < 0.0001$; psychological burden: $r = -0.42$, $P < 0.0001$) and/or professionals (practical burden, and psychological burden: $r = -0.15$, $P < 0.001$).

Multiple Regression Analyses. Socio-demographic and clinical variables accounted for 39% of the variance in practical burden. As shown by the standardized beta weights, practical burden was higher among relatives of patients with higher disability (beta = -0.44 ; $P < 0.05$), and in those without a cohabitating partner (beta = $-.11$; $P < 0.001$). Relatives' perception of available professional and social support accounted for a further 10% of variance. In particular, practical burden was higher among relatives who perceived lower help in the patient's emergencies (beta = -0.24 ; $P < 0.0001$) and less practical support by their social network

(beta = -0.5 ; $P < 0.0001$; model $F = 39.7$; df 11, 448; $P < 0.0001$).

Sixteen percent of variance observed in psychological burden was given by families' socio-demographic characteristics and patients' clinical status. In particular, psychological burden was higher among relatives of patients with more severe disabilities (beta = -0.15 ; $P < 0.001$), who suffered from DMD (beta = 0.09 ; $P < 0.05$), and in unemployed relatives (beta = -0.10 ; $P < 0.01$). The inclusion of the second block of variables, explaining a further 10% of variance, revealed that burden was higher among relatives who perceived that they received lower levels of support in emergencies from their social network (beta = -0.26 ; $P < 0.0001$) and had lower social contacts (beta = -0.012 ; model $F = 16.6$, df 10, 489; $P < 0.0001$).

DISCUSSION

This Italian study has systematically explored the difficulties experienced by the families of young patients with MDs and the professional and social resources on which they may rely. Although the results are as expected, this study attempted to quantify the burden, which will be helpful for evaluating potential interventions in MDs. The large sample size and the selection of 8 participating centers located in different geographical areas make these data representative of the national situation and useful for comparisons with burden in other pathologies and in other countries. Furthermore, replication of the study in international contexts could be facilitated by the use of questionnaires already validated in different languages.^{13,35}

Despite the strengths mentioned above, the study has some limitations that should be taken into account in the interpretation of the results. In particular, the cross-sectional design of the study does not allow us to examine the relationship between burden and resources from a causal perspective. Moreover, as the BI was assessed by interviewing the caregivers, the assessment could be biased by the relatives' burden and attitude to MD. Furthermore, the results of this study cannot be generalized to families of older patients³⁰ or with other types of MD whose impact on the relatives' and patients' quality of life might be significantly different. Finally, the study did not explore the burden experienced by other family members who are often co-involved in a patient's daily care. Most of these limitations will be addressed in further studies now in the planning stage.

In line with previous studies on family caregiving in other long-term illnesses,^{10,26,36-40} relatives stated that caring for a patient with MD mainly results in feelings of sadness and loss and worry

for the future. Feelings of sadness may lead to depression and other minor psychiatric disorders.^{27,28,31} These conditions, in turn, may increase relatives' perception of daily assistance as being very burdensome.²⁶ These results indicate that psychological consequences of caregiving in MD may be high from the early stages of illness, though the patient's functional abilities are still relatively adequate.^{1,37} Psychological burden is greatly influenced by relatives' opportunities to maintain social relationships, which result in lower burden among relatives who are employed,²⁵ less involved in the patient's daily care, and who maintain social contacts. These findings confirm the protective effects of social resources on burden^{10,13,30} and outline the importance for families to share their experience with other families through participation in associations and self-help groups.

The results of this study also indicate that psycho-social interventions are poorly available for patients with MD and their relatives. The scarce number of patients (38) and caregivers (34) who report receiving actual psychological support could be related to poor availability of professionals trained in the psychological care of children with rare diseases and their relatives, and to patient/family reticence to receive support.^{33,41}

Providing relatives with information on MDs and their treatment may strengthen their ability to deal with practical aspects of daily assistance.³⁴ However, medical education may not address relatives' needs for psychological support, which remain largely unmet.^{33,39} The need for psychological support in all phases of MDs is also outlined by the high percentage of relatives who recommended provision of psychological support to families among the strategies to improve their condition.

Higher levels of burden were found among relatives of patients with DMD. However, when the diagnosis was analyzed within the multivariate model, this difference is confirmed only for psychological burden. This finding, likely related to the well-known severe course of DMD, should be taken into account by clinicians when they provide information to families in the early stages of the disease.⁴⁰

These findings confirm that home management of patients with MDs may be demanding for patients' relatives, especially when social and professional resources are poor and patients' functional abilities decrease. Efforts are needed, both at the level of health policies and professional training, to help caregivers and patients face the difficulties of MDs and to value such complex family experience.²⁹

COMPETING INTERESTS

The authors declare that they have no competing interests.

REFERENCES

- Emery AEH. The muscular dystrophies. *Lancet* 2002;359:687–695.
- Passamano L, Taglia A, Palladino A, Viggiano E, D'Ambrosio P, Scutifero M, et al. Improvement of survival in Duchenne Muscular Dystrophy: retrospective analysis of 835 patients. *Acta Myol* 2012;31:121–125.
- Vanasse M, Paré H, Zeller R. Medical and psychosocial considerations in rehabilitation care of childhood neuromuscular diseases. *Handb Clin Neurol* 2013;113:1491–1495.
- Ouyang L, Grosse SD, Fox MH, Bolen J. A national profile of health care and family impacts of children with muscular dystrophy and special health care needs in the United States. *J Child Neurol* 2012;27:569–576.
- Anderson M, Elliott EJ, Zurynski YA. Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support. *Orphanet* 2013;8:22.
- Hatchett L, Friend R, Symister P, Wadhwa N. Interpersonal expectations, social support, and adjustment to chronic illness. *J Pers Soc Psychol* 1997;73:560–573.
- Trief P, Himes C, Orendorff R, Weinstock RS. The marital relationship and psychosocial adaptation and glycemic control of individuals with diabetes. *Diabetes Care* 2001;24:1384–1389.
- Sellwood W, Tarrier N, Quinn J, Barrowclough C. The family and compliance in schizophrenia: the influence of clinical variables, relatives' knowledge and expressed emotion. *Psychol Med* 2003;33:91–96.
- Hauser S, Jacobson A, Lavori P, Wolfsdorf J, Herskovitz R, Milley J, et al. Adherence among children and adolescents with insulin-dependent diabetes mellitus over a four-year longitudinal follow-up. II. Immediate and long-term linkages with the family milieu. *J Pediatr Psychol* 1990;15:527–542.
- Magliano L, Fiorillo A, De Rosa C, Malangone C, Maj M. Family burden in long-term diseases: a comparative study in schizophrenia vs. physical disorders. *Soc Sci Med* 2005;61:313–322.
- Hoening J, Hamilton M. The schizophrenic patient in the community and his effect on the household. *Int J Soc Psychiatry* 1966;12:165–176.
- Rossi Ferrario S, Zotti AM, Baroni A. Emotional reactions and practical problems of the caregivers of hemodialysed patients. *J Nephrol* 2002;15:54–60.
- Magliano L, Fadden G, Madianos M, Caldas de Almeida JM, Held T, Guarneri M, et al. Burden on the families of patients with schizophrenia: results of the BIOMED I study. *Soc Psychiatry Psychiatr Epidemiol* 1998;33:405–412.
- Awad AG, Voruganti LN. The burden of schizophrenia on caregivers. *Pharmacoeconomics* 2008;26:149–162.
- Magliano L, Marasco C, Fiorillo A, Malangone C, Guarneri M, Maj M. The impact of professional and social network support on the burden of families of patients with schizophrenia in Italy. *Acta Psychiatr Scand* 2002;106:291–298.
- Magliano L, Fiorillo A, Malangone C, De Rosa C, Maj M. Patient functioning and family burden in a controlled, real-world trial of family psychoeducation for schizophrenia. *Psychiatr Serv* 2006;57:1784–1791.
- Applebaum AJ, Breitbart W. Care for the cancer caregiver: a systematic review. *Palliat Support Care* 2013;11:231–252.
- Candy B, Jones L, Drake R, Leurent B, King M. Interventions for supporting informal caregivers of patients in the terminal phase of a disease. *Cochrane Database Syst Rev* 2011;CD007617.
- López-Bastida J, Perestelo-Pérez L, Montón-Alvarez F, Serrano-Aguilar P, Alfonso-Sánchez JL. Social economic costs and health-related quality of life in patients with amyotrophic lateral sclerosis in Spain. *Amyotroph Lateral Scler* 2009;10:237–243.
- Rossi Ferrario S, Zotti AM, Ippoliti M, Zotti P. Caregiving-related needs analysis: a proposed model reflecting current research and socio-political developments. *Health Soc Care Community* 2003;11:103–110.
- Schulze B, Rössler W. Caregiver burden in mental illness: review of measurement, findings and interventions in 2004–2005. *Curr Opin Psychiatry* 2005;18:684–691.
- Kim Y, Given BA. Quality of life of family caregivers of cancer survivors: across the trajectory of the illness. *Cancer* 2008;112:2556–2568.
- Etters L, Goodall D, Harrison BE. Caregiver burden among dementia patient caregivers: a review of the literature. *J Am Acad Nurse Pract* 2008;20:423–428.
- Bothwell JE, Dooley JM, Gordon KE, MacAuley A, Camfield PR, MacSween J. Duchenne muscular dystrophy—parental perceptions. *Clin Pediatr (Phila)* 2002;41:105–109.
- Larkindale J, Yang W, Hogan PF, Simon CJ, Zhang Y, Jain A, et al. Cost of illness for neuromuscular diseases in the United States. *Muscle Nerve* 2014;49:431–438.

26. Pangalila RF, van den Bos GAM, Stam HJ, van Exel NJA, Brouwer WBF, Roebroek ME. Subjective caregiver burden of parents of adults with Duchenne muscular dystrophy. *Disabil Rehabil* 2012;34:988–996.
27. Kenneson A, Bobo JK. The effect of caregiving on women in families with Duchenne/Becker muscular dystrophy. *Health Soc Care Community* 2010;18:520–528.
28. Abi Daoud MS, Dooley JM, Gordon KE. Depression in parents of children with Duchenne muscular dystrophy. *Pediatr Neurol* 2004;31:16–19.
29. Magliano L, Patalano M, Saggiocchi A, Scutifero M, Zaccaro A, D'Angelo MG, et al. "I have got something positive out of this situation": psychological benefits of caregiving in relatives of young people with muscular dystrophy. *J Neurol* 2014;261:188–195.
30. Chen JY, Clark MJ. Family function in families of children with Duchenne muscular dystrophy. *Fam Community Health* 2007;30:296–304.
31. Im SH, Lee SC, Moon JH, Park ES, Park YG. Quality of life for primary caregivers of muscular dystrophy Subject description. *Chin Med J (Engl)* 2010;123:452–457.
32. Boström K, Ahlström G, Sunvisson H. Being the next of kin of an adult person with muscular dystrophy. *Clin Nurs Res* 2006;15:86–106.
33. Farmer JE, Marien WE, Clark MJ, Sherman A, Selva TJ. Primary care supports for children with chronic health conditions: identifying and predicting unmet family needs. *J Pediatr Psychol* 2004;29:355–367.
34. Shaw SE, Meads G. Extending primary care: potential learning from Italy. *Prim Health Care Res Dev* 2012;13:289–293.
35. Mahoney F, Barthel D. Functional evaluation: the Barthel Index. *Md State Med J* 1965;14:61–65.
36. Akkuş Y. Multiple sclerosis patient caregivers: the relationship between their psychological and social needs and burden levels. *Disabil Rehabil* 2011;33:326–333.
37. Bushby K, Finkel R, Birnkrant DJ, Case LE, Clemens PR, Cripe L, et al. Diagnosis and management of Duchenne muscular dystrophy. 1. Diagnosis, and pharmacological and psychosocial management. *Lancet Neurol* 2010;9:77–93.
38. Green SE. "We're tired, not sad": benefits and burdens of mothering a child with a disability. *Soc Sci Med* 2007;64:150–163.
39. Miller AR, Condin CJ, McKellin WH, Shaw N, Klassen AF, Sheps S. Continuity of care for children with complex chronic health conditions: parents' perspectives. *BMC Health Serv Res* 2009;9:242.
40. Eccleston C, Palermo TM, Fisher E, Law E. Psychological interventions for parents of children and adolescents with chronic illness. *Cochrane Database Syst Rev* 2012:CD009660.
41. Green JM, Murton FE. Diagnosis of Duchenne muscular dystrophy: parents' experiences and satisfaction. *Child Care Health Dev* 1996;22:113–128.