Mental health and psychological adaptation on parents of children with neuromuscular diseases

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The diagnosis of a pediatric neuromuscular disease has a psychological impact on parents. The study aimed to describe mental health and psychological adaptation in parents of children with neuromuscular diseases. The evaluation of parents (n=35) through the Psychological Adaptation Scale and Adult Self-Report Questionnaire showed that (a) 82.4% of the participants had an adequate level of psychological adaptation; (b) parents presented high levels of mental health problems, (c) significant correlations were found between the mental health problems and psychological adaptation. This study highlights the need for psychological interventions which aim to promote psychological adaptation to NMD diagnoses and protect parental mental health.

Keywords: mental health; psychological adaptation; neuromuscular diseases; parents; caregivers

Introduction

Neuromuscular diseases (NMD) comprise about 150 diagnoses characterized by progressive muscle weakness. These conditions tend to be genetic and hereditary. Other signs and symptoms they present are muscle atrophy or hypertrophy, fatigue, myalgia, and degeneration of muscles and nerves (Aguerre et al., 2014; Caro et al., 2014). Worldwide it is estimated that between 7.1 and 26.5 in 100,000 people have a NMD, therefore they are considered Rare Diseases (RD) (Deenen et al.,2015).

Previous studies have shown that the diagnosis of conditions that compromise pediatric health and involve a genetic component in their etiology, generates a psychological impact on parents in terms of stress, anxiety, pain, sadness and

depression. These lead to feelings of worry, uncertainty, anger, shame, and guilt (Biesecker, 2016; Canbulat et al., 2014; Dinc & Terzioglu, 2006; McAllister et al., 2007; Ortega & Vázquez, 2018).

Parents of children with Duchenne Muscular Dystrophy (DMD), experiment a state of shock at the moment of diagnosis, followed for a modification of their personal projects (Gargiulo et al., 2013). Some authors suggest the diagnostic impact is usually more significant in mothers, due to the inheritance pattern of this disease. Impact can also be more significant in those families that do not have a family history with these diseases (Buchanan et al., 1979).

In addition to the initial psychological impact that accompanies the communication of these diagnoses, NMD generate loss of functional autonomy (Camacho et al., 2015). Muscle degeneration leads to the need of a caregiver to help cover daily activities and provide support in the progression of the disease (Achury et al., 2011). The caregiver role is usually carried out by a family member, which implies a reassignment of family responsibilities as well as a change in supportive relationships in the family (Cueto et al., 2013). It has been described that mothers tend to assume the role of the main caregiver, and fathers tend to take a more support role (Tomiak et al., 2007; De Albuquerque et al., 2010). Studies that examined caregiver and family burden in caregivers have found that half of caregivers report high levels of stress, substantial burden and lower quality of life (Kennenson & Bobo, 2010; Landfelt et al., 2016; Landfelt et al., 2018; Moura et al., 2015). Burden has been significantly associated with low income, low resiliency, and time caring for the child (Kennenson & Bobo, 2010; Moura et al., 2015). ().

Parents of children with NMD tend to have high levels of marital conflict or

problems and concerns related to family functioning (Buchanan et al., 1979; Baiocco et al., 2017; Thomas et al., 2014). Marital conflicts often revolve around children's care, discipline matters and fatigue related to the caregiver role (Lázaro Pérez, 2012). Many parents express not having time to enjoy family life, as well as the loss of friends, resignation of their careers and their own desires (Landfelt et al., 2018; Mah et al., 2008). Caregivers also report dividing their time between house chores and caregiving duties, leaving no time for expanding their social support networks (De Albuquerque et al., 2010).

The lack of social support, both within and outside the family, has been associated with higher levels of stress and psychological burden in parents of children with NMD (Frishman et al., 2017; Magliano et al., 2015; Kennenson & Bobo, 2010). Other factors that have been associated with psychological distress, higher burden and lower mental health in parents of children with NMD are low resiliency, low income, unemployment, years of caring, not living with a partner and feelings of guilt and pessimism (Landfelt et al., 2016; Moura et al., 2015; Magliano et al., 2015).

Although going through the diagnosis is a difficult process, most families manage to adapt psychologically to the situation (Biesecker & Erby, 2008). Regarding psychological adaptation, Peay et al. (2016) conducted a study with mothers of children with DMD in which they found that they manage to adapt to the disease. On the contrary, other study showed that most of the parents were not in terms with the diagnosis yet, and that those unresolved parents presented lower levels of family functioning and satisfaction (Baiocco et al., 2017).

Psychological adaptation over time does not seem to be predicted by caregiver overload or by the child's functional status, but rather seems to be associated with

resilience and the positive impact perceived by mothers (Peay et al., 2016). Often, parents of children with DMD experience a period of personal achievement that comes from focusing in the positive aspects of their lives and feeling able to successfully overcome the daily challenges of caring for the child (Mah et al., 2008; Samson et al., 2009). This sense of personal achievement and the coping mechanisms lead families to a feeling of hope (Samson et al., 2009). Other positive effects that have been described by parents are an increase in strength and courage to face problems and, appreciation of life (Magliano et al., 2014).

Regarding mental health, parents of children with NMD report significantly lower levels than the general population, having a higher risk of suffering an episode of major depression, anxiety problems, and even suicide risk (Chen et al., 2020; Chen & Clark, 2010; Daoud et al., 2004; De Alba Agredano et al., 2015; Landfelt et al., 2018; Mah et al., 2008; Nozoe et al., 2016). Likewise, these parents report sleep problems and difficulty performing their usual activities (Chen et al., 2020; Mah et al., 2008; Nozoe et al., 2016).

The scientific relevance of different contributions demonstrating the impact of NMD on quality of life is undeniable. Chronicity and loss of autonomous functioning due to progressive muscle weakness result in greater dependence on caregivers, which has a negative impact on family functioning; affecting the quality of life of people with NMD at all ages (children, adolescents and adults) and reduces the quality of life of their caregivers (Baiardini et al., 2011; Bendixen et al., 2012; Bhullar et al., 2019; Boyer et al., 2006; Gocheva et al., 2019; Graham et al., 2011; Landfelt et al., 2016; Ortega, 2020; Souares de Moura et al., 2015; Uzark et al., 2012; Zhi et al., 2019). However, there is a lack of research centered on the study of mental health and the level

of acceptance of the diagnosis of caregivers. These variables are considered key variables because of how they can affect the quality of care.

Most of the studies that have been mentioned come from Europe or the United States. In Latin America and particularly in Argentina, there is a lack of research that addresses the study of NMD from a health psychology perspective. These studies are needed to consider a possible influence of the health care contexts on quality of life. Despite the fact that Argentina has a Rare Disease Regulation, few programs on diagnosis and research in these conditions have been implemented to date (Dharssi et al., 2017). Unlike other countries, Argentina does not count with a formal Rare Disease Center, which contributes to patient association groups taking a more active role in supporting families and diagnosis (Dharssi et al., 2017). One of the first antecedents in the region that addresses genetic diseases from a psychological perspective concludes that mothers who achieve a good psychological adaptation to their children's diagnosis have better mental health, understand genetic information better and feel empowered (Vázquez et al., 2018). In order to contribute with specific knowledge in families with NMD, the present study aims to evaluate the association between psychological adaptation and mental health in parents of children with NMD in Argentina.

Methods

A quantitative, cross-sectional study with a non-experimental, correlational design was conducted to evaluate the association between psychological adaptation and mental health.

Participants

The sample selection was non-probabilistic. The sample consisted of parents of children (1 to 18 years old) with a confirmed diagnosis of neuromuscular disease who lived in

Argentina. Participants were contacted through the Argentinian Muscular Dystrophy Association (ADM) and other online groups of parents.

A total of 35 families participated in the study. The majority (85.3%) of the respondents were mothers of children with NMD. The mean age of mothers was 37.8 (SD=7.61), and the mean age for fathers was 42.09 (SD=7.20). For the children, ages ranged from 1 to 18, with a mean of 9.53 (SD=4.45). Table 1 shows in detail the sociodemographic characteristics of the children and adolescents, and the sociodemographic data of the caregivers who responded to the protocol.

Procedure

To be part of the study, participants were required to sign an informed consent form. Questionnaires were answered both online through the GoogleForms platform and in person. When it was in person, the questionnaires were answered by the parent in one session, and if they had difficulty reading or understanding a research assistant could help them. The online version was offered in those cases where a face-to-face meeting could not be scheduled or participants themselves had difficulty attending. Both types of data collection included the same set of items and instruments.

Instruments

The Psychological Adaptation Scale (PAS): This instrument measures psychological adaptation to a chronic condition or disease risk, and it was designed to see the extent or scope of adaptation at a given time (Biesecker et al., 2013). The original scale is composed of four sub-scales, each representing a domain of the concept of adaptation, these being: effective coping, self-esteem, spiritual or existential well-being, and social integration. It is composed of 15 items answered with a Likert scale (1-5). The PAS has a total score, that ranges from 15 to 75. The scale also can be scored

from 1 to 5, with a cut-off score of 3, from which the level of adaptation is considered adequate. Both scores are interpreted as the higher the score, the higher the level of adaptation. This scale was validated for the Argentine population. Construct validity was studied by a parallel analysis and an exploratory factor analysis. The significance of the Bartlett sphericity test ($\chi^2_{(105)} = 1928.4$; $p < 0.001$) was significant and the Kaiser-Meyer-Olkin (KMO) sample adequacy index indicated an adequacy of 0.87. A single factor was obtained, explaining 45% of the variance. Reliability was also reported for Argentine population, finding a high value of internal consistency $\alpha = 0.915$ (Vázquez et al., 2020). Cronbach's alpha coefficient was calculated for this sample, $\alpha = 0.93$, demonstrating high internal consistency.

The Adult Self Report (ASR:) This scale was designed to assess emotional, social, behavioral, and thinking problems, as well as personal strengths and adaptive functioning. The ASR consists of 126 items that assess behavioral and emotional problems and substance use, these items are evaluated as 0 (not true), 1 (somehow or sometimes true) and 2 (very true or generally true). Under these items, adaptive functioning (including friends, partner, family, work and education) and syndromes, which are problems that tend to occur together, are evaluated. The part of the scale that evaluates syndromes is composed of two broad scales: Externalizing and Internalizing problems. Externalizing problems include aggressive, criminal and intrusive behavior syndromes, and Internalizing problems include anxious-depressive, somatic complaints without known medical cause and withdrawal. Attention and thinking problems syndromes are not included in none of the broad scales but are part of the total score. This questionnaire has adequate levels of reliability, content validity and construct validity (Achenbach & Rescorla, 2003). This scale has also been adapted to Argentine

population, demonstrating validity criteria and high reliability with Cronbach's Alpha coefficients ranging from 0,63 to 0,94 (Ivannova et al., 2015; Samaniego & Vázquez, 2012). Internal consistency was also calculated for this sample, indicating a high realiabilty (α =0.89).

Ethics statement

It is important to mention that this research is part of a larger project entitled *Genetic Counseling: Analysis of its psychological impact on families with children suffering from genetic diseases*, which has been approved by the Ethics Committee and the Teaching and Research Committee of the Dr. Ricardo Gutierrez Children's Hospital (CEI N 16.09).

Data analysis

Data analysis was carried out using SPSS Statistics-25 (Statistical Package for the Social Sciences version 25, IBM SPSS Inc, Chicago, IL, USA). Percentages, means and standard deviations were calculated for the descriptive analysis of the PAS and ASR scales. Considering the cut-off score established by the author of the PAS instrument, a mean of the averaged score was calculated to determine the need for psychological intervention. Even though the PAS' validity analysis showed a unidimensional construct, means were calculated for the proposed dimensions in order to compare them with previous studies. The normality of the sample was checked using the Shapiro-Wilk method and since it did not have a normal distribution (ASR p=0.004, PAS p=0.001), it was decided to use Spearman's Rho non-parametric statistic to study the correlation between the variables. Correlations were calculated both for the total scores and dimensions of the instruments.

Results

Psychological Adaptation

Considering the PAS cut-off point (=>3), results of this study showed that 82.4% had an adequate level, while 17.6% were below the expected psychological adaptation. The total scale showed a mean of 4.03 (SD= 0.93). The scores for the dimensions considering the 1 to 5 range for the scale are shown in *Table 2*.

Mental Health and Adaptative Functioning

Regarding the adaptative functioning of the parents who participated in the study, specifically in the couple relationship perception, 75% out of the 70.6% of the sample of parents who were married or living together reported being satisfied with their partner. A 12.9% of the sample indicated that their partner's behavior frequently bothered them. Regarding friendships, it was found that 85.3% of the sample reported having 2 or more friends outside their family group, while the remaining 14.7% reported having 1 or no friends. Regarding the relationship with these friends, 61.8% of the parents reported getting along with their friends better than average, and 44.1% of them reported having contact with their friends more than 5 times a month. The mean scores for the dimensions of Adaptative Functioning are shown in *Table 3*. When comparing these mean scores to onmicultural means for the scale, a large effect was found on the Family subscale (*d*= 2,04), with higher means for the NMD families. Same as in the Personal Strengths scale, where a small effect size was found (*d*=0.23).

In terms ofmental health, parents presented high levels of mental health problems with a mean of 53.21 (SD = 31.17), a mean of 21.79 (SD = 13.81) for internalizing problems, and for externalizing problems a mean of 10.76 (SD = 9.21). These results are compared to omnicultural means in *Table 4.* Small and medium effect sizes were found for total problems and internalizing problems respectively, where

parents of children with NMD seem to have greater mental health problems than general population. Furthermore, the results obtained for the narrow scales can be seen in *Table 5*.

Correlations between mental health, psychological adaptation and adaptative functioning

First, the relationship between mental health problems and psychological adaptation was explored. Significant correlations were found between the total level of mental health problems and psychological adaptation (Rho= -0,42; *p*= 0.01; N=34). These correlations are moderate and inversely proportional, that is, those parents who have better levels of psychological adaptation, have fewer problems in their mental health. Correlations between psychological adaptation dimensions and mental health problems subscales are shown in *Table 6*.

Last, correlations were drawn between adaptative functioning and mental health, and adaptative functioning and psychological adaptation. Statistically significant correlations were found between the friends subdimension and internalizing problems (Rho= -0.394; *p*= 0.021; N = 34). Significant correlations were also found between the partner / husband subdimension and internalizing problems (Rho = -0.575; *p*= 0.001; N = 34), externalizing problems (Rho= -0.568; *p*= 0.001; N = 34) and total mental health problems (Rho= -0,605; *p*= 0.000; N= 34). All other significant and non-significant correlations examined in this study are presented in *Table 7*.

Discussion

This study investigated levels of mental health problems and psychological adaptation on parents of children with neuromuscular diseases in Argentina.

Regarding psychological adaptation, the results showed that most parents had adequate levels of psychological adaptation. These results reinforce the idea that most families that have children with NMD manage to adapt psychologically to these conditions (Magliano et al., 2015) and differ from the results of Baiocco et al. (2017). On the other hand, these levels are adequate, and similar to those found by Peay et al. (2016) in mothers of children with Duchenne Muscular Dystrophy and to those found in a previous study of Argentine families with children with different genetic diseases (Vázquez et al., 2018).

Another finding of this study shows that mental health seems to be impaired at a general level on parents of children with NMD. These parents presented higher levels of mental health problems compared to omnicultural means, and even families with different genetic diseases (Rescorla et al., 2016; Vázquez et al., 2018). Furthermore, the results obtained in this sample are similar to those found in the mental health clinical sample from Argentina, that is, adults who were starting a psychotherapeutic process (Samaniego & Vázquez, 2012).

Specifically, parents of children with NMD reported high levels of internalizing problems, which included symptoms of anxiety, depression, and withdrawal. These findings are in line with the revised antecedents of caregivers of patients with chronic and neuromuscular diseases (Chen & Clark, 2010; Daoud et al., 2004; De Alba Agredano et al., 2015; Landfelt et al., 2016; Landfelt et al., 2018). These results highlight the need to consider caregivers as a risk group of developing mental health problems and the need for psychological intervention that addresses these problems.

Contrary to what has been stated in the literature about couple´s problems in parents of children with NMD, most parents who participated in the research reported

being satisfied with their relationship (Buchanan et al., 1979; Lázaro Pérez, 2012; Mah et al., 2008). In addition, results on adaptative functioning, regarding friends, spouse, and family dimensions, were similar to or even greater than those reported by Rescorla et al. (2016) for general population. Furthermore, these resources were found to have a negative correlation with mental health, meaning those families with a greater perception of their personal strengths and family and friends support had lower mental health problems. These results may be related to the positive impact of the diagnosis described by Magliano et al., (2014), Mah et al., (2008) and Samson et al., (2009).

It is important to mention that 85.3% of the parents who participated in the study were mothers, however, this phenomenon is repeated various studies with NMD (Buchanan et al., 1979; Mah et al., 2008; De Alba Agredano et al., 2015). This result support the idea that it is the mothers who usually assume the role of main caregiver (De Albuquerque et al., 2010; Tomiak et al., 2007). It is also necessary to emphasize that the children with NMD were predominantly male. This is also repeated in the literature and could be related to the fact that the prevalence of some NMD is higher in males, as in the case of Duchenne Muscular Dystrophy (Nozoe et al., 2016).

This study is a contribution to the understanding of the situation that families with NMD go through on a local level. In Argentina, there are few intervention programs that address psychological and social problems that these families face. It is usually a chore left for patient associations to take care of. Health professionals should be aware of the conflict involved and be prepared to address it as part of health care.

One of the most relevant conclusions to point out is that this is the first study to explore and find a significant correlation between mental health and psychological adaptation on caregivers of children with NMD. In line with previous studies,

caregivers (mainly mothers) are at risk of presenting mental health problems, especially internalizing problems such as anxiety, depression and withdrawal. On the other hand, results indicate that most of the parents of children with NMD manage to adapt psychologically to the condition. These findings provide a clue as to where to target psychological interventions when working with these families.

Limitations

The results of the study should be considered in light of its limitations. First, even though neuromuscular disorders are considered rare diseases, a larger sample should be recruited. The size of this sample may have limited the power to identify significant correlations between variables.

In addition, significant variables, such as years since the diagnosis and level of disability, were not measured and should be considered for further studies. Moreover, it would be interesting to follow up on parent´s psychological variables through longitudinal studies. Likewise, it would be useful to correlate similar measures (psychological adaptation and mental health) between children and their parents. However, there are no instruments that measure the same constructs in both populations and that have been adapted to the characteristics of these diseases. Finally, it would be interesting to include other measures, such as resilience and social support, in future studies.

Implications for practice

These findings demonstrated the association between psychological adaptation and mental health. This association highlights the need for preventive strategies for those families who do not manage to adapt psychologically to the NMD. Health care professionals should consider this association when encountering families with

neuromuscular disorders. Psychological support should be available for the families at the time of diagnosis to manage its psychological impact on parental mental health. It should also accompany the progression of the disease, promoting family adaptation to this progression (Lorenza et al., 2017). Additional research is needed to identify psychological interventions that promote quality of life from a family-centered perspective in this population.

REFERENCES

Achenbach, T.M. & Rescorla, L.A. (2003). Manual for the ASEBA Adult Forms & Profiles. Burlington, VT: University of Vermont, Research Center for Children, Youth, & Families

Achury Saldaña, D., Castaño Riaño, H., Gómez Rubiano, L., & Guevara Rodríguez, N. (2011). Calidad de vida de los cuidadores de pacientes con enfermedades crónicas con parcial dependencia. Investigación En Enfermería: Imagen Y Desarrollo, 13(1), 27-46. https://revistas.javeriana.edu.co/index.php/imagenydesarrollo/article/view/1632

Aguerre, V. (2014). Consenso de cuidados respiratorios en enfermedades neuromusculares en niños. Resumen ejecutivo. Arch Argent Pediatr, 112(5), 476-477. http://dx.doi.org/10.5546/aap.2014.476

Baiardini, I., Minetti, C., Bonifacino, S., Porcu, A., Klersy, C., Petralia, P., ... & Braido, F. (2011). Quality of life in Duchenne muscular dystrophy: the subjective impact on children and parents. *Journal of child neurology, 26(*6), 707-713. https://doi.org/10.1177/0883073810389043

Baiocco, R., Gattinara, P. C., Cioccetti, G., & Ioverno, S. (2017). Parents' reactions to the diagnosis of Duchenne muscular dystrophy: associations between resolution,

family functioning, and child behavior problems. journal of nursing research, 25(6), 455-463. doi: 10.1097/JNR.0000000000000186

Bawa, K., Chen, E., Noone, J. M., Whitmire, S. M., Buchenberger, J. D., Arnold, W. D., ... & Dixon, S. (2020). Impact of Spinal Muscular Atrophy on Caregivers' Daily Activities and Health-related Quality of Life. Research Square. DOI: 10.21203/rs.3.rs-16569/v1.

Bendixen, R. M., Senesac, C., Lott, D. J., & Vandenborne, K. (2012). Participation and quality of life in children with Duchenne muscular dystrophy using the International Classification of Functioning, Disability, and Health. *Health and quality of life outcomes*, *10*(1), 43. https://doi.org/10.1186/1477-7525-10-43

Bhullar, G., Miller, M. R., Campbell, C., We, Y., & El-Aloul, B. (2019). P. 067 Quality of my life: perceptions of boys with Duchenne muscular dystrophy and their parents. Canadian Journal of Neurological Sciences, 46(s1), S32-S32. https://doi.org/10.1017/cjn.2019.167

Biesecker, B. B. (2016). Genetic Counselling: Psychological Issues. Encyclopedia of Life Sciences (eLS), 1–5. https://doi.org/10.1002/9780470015902.a0005616.pub3

Biesecker, B. B., & Erby, L. (2008). Adaptation to living with a genetic condition or risk: a mini-review. Clinical genetics, 74(5), 401–407. https://doi.org/10.1111/j.1399-0004.2008.01088.x

Biesecker, B. B., Erby, L. H., Woolford, S., Adcock, J. Y., Cohen, J. S., Lamb, A., . . . Reeve, B. B. (2013). Development and validation of the psychological adaptation scale (PAS): use in six studies of adaptation to a health condition or risk. Patient education and counseling, 93(2), 248-254. https://doi.org/10.1016/j.pec.2013.05.006

Boyer, F., Novella, J. L., Coulon, J. M., Delmer, F., Morrone, I., Lemoussu, N., Bombart, V., Calmus, A., Cornu, J.Y., Dulieu, V. & Etienne, J. C. (2006). Family caregivers and hereditary muscular disorders: association between burden, quality of life and

mental health. In *Annales de readaptation et de medecine physique: revue scientifique de la Societe francaise de reeducation fonctionnelle de readaptation et de medecine physique, Vol. 49* (1), 16-22. DOI: 10.1016/j.annrmp.2005.08.001

Buchanan, D. C., LaBarbera, C. J., Roelofs, R., & Olson, W. (1979). Reactions of families to children with Duchenne muscular dystrophy. General Hospital Psychiatry, 1(3), 262-269. https://doi.org/10.1016/0163-8343(79)90028-8

Camacho, A., Esteban, J., & Paradas, C. (2015). Informe de la Fundación Del Cerebro sobre el impacto social de la esclerosis lateral amiotrófica y las enfermedades neuromusculares. Neurología 33(1), 35-46. https://doi.org/10.1016/j.nrl.2015.02.003

Canbulat, N., Demirgöz Bal, M., & Çoplu, M. (2014). Emotional reactions of mothers who have babies who are diagnosed with Down syndrome. International journal of nursing knowledge, 25(3), 147-153. https://doi.org/10.1111/2047-3095.12026

Caro, I. A., Paz, J. F. L., & Pérez, E. L. (Eds.). (2014). *Enfermedades neuromusculares: Bases para la intervención* (Vol. 17). Universidad de Deusto.

Chen, J. Y., & Clark, M. J. (2010). Family resources and parental health in families of children with Duchenne muscular dystrophy. Journal of Nursing Research, 18(4), 239-248. doi: 10.1097/JNR.0b013e3181fbe37b

Cueto, H. B., Ortega, E. L., Bustillo, R. M., Suárez, K. P., Polo, D. R., & Prieto, A. M. (2013). Cuidadores familiares de niños con cáncer y su funcionalidad. Salud Uninorte, 29 (2), 249-259. https://www.redalyc.org/articulo.oa?id=817/81730430010

Daoud, M. S. A., Dooley, J. M., & Gordon, K. E. (2004). Depression in parents of children with Duchenne muscular dystrophy. Pediatric neurology, 31(1), 16-19. https://doi.org/10.1016/j.pediatrneurol.2004.01.011

de Alba Agredano, M., Valencia, A. C., & Loyo, L. M. S. (2015). Riesgo suicida y síntomas depresivos en padres de hijos con enfermedad neuromuscular. Acta de investigación psicológica, 5(1), 1872-1880. https://doi.org/10.1016/S2007-4719(15)30007-7

Deenen, J. C., Horlings, C. G., Verschuuren, J. J., Verbeek, A. L., & van Engelen, B. G. (2015). The epidemiology of neuromuscular disorders: a comprehensive overview of the literature. Journal of Neuromuscular Diseases, 2(1), 73-85. Doi: 10.3233/JND-140045

Dharssi, S., Wong-Rieger, D., Harold, M., & Terry, S. (2017). Review of 11 national policies for rare diseases in the context of key patient needs. *Orphanet journal of rare diseases*, *12*(1), 1-13. https://doi.org/10.1186/s13023-017-0618-0

Dinc, L., & Terzioglu, F. (2006). The psychological impact of genetic testing on parents. Journal of clinical nursing, 15(1), 45-51. https://doi.org/10.1111/j.1365-2702.2005.01228.x

Frishman, N., Conway, K. C., Andrews, J., Oleson, J., Mathews, K., Ciafaloni, E., ... & McKirgan, L. (2017). Perceived quality of life among caregivers of children with a childhood-onset dystrophinopathy: a double ABCX model of caregiver stressors and perceived resources. Health and quality of life outcomes, 15(1), 33. https://doi.org/10.1186/s12955-017-0612-1

Gargiulo, M., Angeard, N., Herson, A., Fosse, S., Noël, C. T., Jacquette, A., ... & Mazet, P. (2013). Impacto psicológico de la enfermedad de Duchenne sobre el niño y el adolescente, sus padres y sus familiares. Once años de experiencia en un equipo multidisciplinario. Rehabil. Integral, 8(2), 78-90. https://www.rehabilitacionintegral.cl/wp-content/files_mf/5gargiulo.pdf

Gocheva, V. Z. (2019). *Patient-reported outcomes in neuromuscular disorders–health-related quality of life and psychosocial adjustment in post-polio syndrome and Duchenne muscular dystrophy* (Doctoral dissertation, University_of_Basel).

Graham, C. D., Rose, M. R., Grunfeld, E. A., Kyle, S. D., & Weinman, J. (2011). A systematic review of quality of life in adults with muscle disease. *Journal of neurology*, *258*(9), 1581-1592. https://doi.org/10.1007/s00415-011-6062-5

Ivanova, M. Y., Achenbach, T. M., Rescorla, L. A., Turner, L. V., Ahmeti-Pronaj, A., Au, A., ... & Csemy, L. (2015). Syndromes of self-reported psychopathology for ages 18–59 in 29 societies. Journal of Psychopathology and Behavioral Assessment, 37(2), 171-183. https://doi.org/10.1007/s10862-014-9448-8

Kenneson, A., & Bobo, J. K. (2010). The effect of caregiving on women in families with Duchenne/Becker muscular dystrophy. Health & social care in the community, 18(5), 520-528. https://doi.org/10.1111/j.1365-2524.2010.00930.x

Landfeldt, E., Edström, J., Buccella, F., Kirschner, J., & Lochmüller, H. (2018). Duchenne muscular dystrophy and caregiver burden: a systematic review. Developmental Medicine & Child Neurology, 60(10), 987-996. https://doi.org/10.1111/dmcn.13934

Landfeldt, E., Lindgren, P., Bell, C. F., Guglieri, M., Straub, V., Lochmüller, H., & Bushby, K. (2016). Quantifying the burden of caregiving in Duchenne muscular dystrophy. Journal of neurology, 263(5), 906-915. DOI 10.1007/s00415-016-8080-9

Lázaro Pérez, E. (2012). Afrontamiento del estrés en padres y madres con hijos/as con enfermedades neuromusculares. [Tesis de Doctorado, Universidad de Deusto]. URL: https://dialnet.unirioja.es/servlet/tesis?codigo=158599

Lorenza, M., Marianna, S., Melania, P., Alessandra, S., Antonella, Z., Federica, C., ... & Luisa, P. (2017). Integrated care of muscular dystrophies in Italy. Part 2.

Psychological treatments, social and welfare support, and financial costs. *Acta Myologica*, *36*(2), 41. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5530600/

Magliano, L., Patalano, M., Sagliocchi, A., Scutifero, M., Zaccaro, A., D'angelo, M. G., ... & Messina, S. (2015). Burden, professional support, and social network in families of children and young adults with muscular dystrophies. Muscle & nerve, 52(1), 13-21. https://doi.org/10.1002/mus.24503

Magliano, L., Patalano, M., Sagliocchi, A., Scutifero, M., Zaccaro, A., D'Angelo, M. G.,... & Messina, S. (2014). "I have got something positive out of this situation": psychological benefits of caregiving in relatives of young people with muscular dystrophy. Journal of neurology, 261(1), 188-195. https://doi.org/10.1007/s00415-013-7176-8

Magliano, L., Scutifero, M., Patalano, M., Sagliocchi, A., Zaccaro, A., Civati, F., Brighina, E., Vita, G., Messina, S., Sframeli, M., Lombardo, M. E., Scalise, R., Colia, G., Catteruccia, M., Berardinelli, A., Motta, M. C., Gaiani, A., Semplicini, C., Bello, L., Astrea, G., … Politano, L. (2017). Integrated care of muscular dystrophies in Italy. Part 2. Psychological treatments, social and welfare support, and financial costs. *Acta myologica : myopathies and cardiomyopathies : official journal of the Mediterranean Society of Myology*, *36*(2), 41–45

Mah, J. K., Thannhauser, J. E., McNeil, D. A., & Dewey, D. (2008). Being the lifeline: The parent experience of caring for a child with neuromuscular disease on home mechanical ventilation. Neuromuscular Disorders, 18(12), 983–988. doi:10.1016/j.nmd.2008.09.001

McAllister, M., Davies, L., Payne, K., Nicholls, S., Donnai, D., & MacLeod, R. (2007). The emotional effects of genetic diseases: implications for clinical genetics.pea

American Journal of Medical Genetics Part A, 143(22), 2651-2661. https://doi.org/10.1002/ajmg.a.32013

Moura, M. C. D. S. D., Wutzki, H. C., Voos, M. C., Resende, M. B. D., Reed, U. C., & Hasue, R. H. (2015). Is functional dependence of Duchenne muscular dystrophy patients determinant of the quality of life and burden of their caregivers?. Arquivos de neuro-psiquiatria, 73(1), 52-57. http://dx.doi.org/10.1590/0004-282X20140194

Nozoe, K. T., Polesel, D. N., Moreira, G. A., Pires, G. N., Akamine, R. T., Tufik, S., & Andersen, M. L. (2016). Sleep quality of mother-caregivers of Duchenne muscular dystrophy patients. Sleep and Breathing, 20(1), 129-134. https://doi.org/10.1007/s11325-015-1196-9

Ortega, J. (2020). Resultados preliminares de la evaluación de la calidad de vida del niño con distrofia muscular de Duchenne y de su cuidador principal. *Memorias XII Congreso Internacional de Investigación y Práctica Profesional en Psicología.*

Ortega, J., & Vázquez, N. (2018). Diagnóstico de fisura labio palatina en niños pequeños de Nicaragua: impacto del diagnóstico a nivel familiar. Revista De La Facultad De Ciencias Médicas De Córdoba, 75(4), 270-278. http://dx.doi.org/10.31053/1853.0605.v75.n4.19931

Peay, H. L., Meiser, B., Kinnett, K., Furlong, P., Porter, K., & Tibben, A. (2016). Mothers' psychological adaptation to Duchenne/Becker muscular dystrophy. European Journal of Human Genetics, 24(5), 633. https://doi.org/10.1038/ejhg.2015.189

Rescorla, L. A., Achenbach, T. M., Ivanova, M. Y., Turner, L. V., Althoff, R. R., Árnadóttir, H. A., ... & Csemy, L. (2016). Problems and adaptive functioning reported by adults in 17 societies. International Perspectives in Psychology:

Research, Practice, Consultation, 5(2), 91. https://doi.org/10.1037/ipp0000046

Samaniego, V. C. & Vázquez, N. (2012). Adult psychopathology: Is there any agreement between self-reports and reports by other informants? Presented at the 30th International Congress of Psychology. South Africa: Cape Town.

Samson, A., Tomiak, E., Dimillo, J., Lavigne, R., Miles, S., Choquette, M., ... & Jacob, P. (2009). The lived experience of hope among parents of a child with Duchenne muscular dystrophy: perceiving the human being beyond the illness. Chronic illness, 5(2), 103-114. https://doi.org/10.1177/1742395309104343

Soares de Moura, M., Wutski, H., Voos, M., Resende, M., Reed, U., & Hasue, R. (2015). G.P.53 - Is functional dependence of Duchenne muscular dystrophy patients determinant of the quality of life and burden of their caregivers? *Neuromuscular Disorders, 25*, S202. http://dx.doi.org/10.1590/0004-282X20140194

Thomas, P. T., Rajaram, P., & Nalini, A. (2014). Psychosocial challenges in family caregiving with children suffering from Duchenne muscular dystrophy. Health & social work, 39(3), 144-152. https://doi.org/10.1093/hsw/hlu027

Tomiak, E. M., Samson, A., Miles, S. A., Choquette, M. C., Chakraborty, P. K., & Jacob, P. J. (2007). Gender-specific differences in the psychosocial adjustment of parents of a child with duchenne muscular dystrophy (DMD)-Two points of view for a shared experience. Qualitative Research Journal, 7(2), 2-21. doi: 10.3316/QRJ0702002.

Urzúa, A., & Caqueo-Urízar, A. (2012). Calidad de vida: Una revisión teórica del concepto. *Terapia psicológica*, *30*(1), 61-71. https://dx.doi.org/10.4067/S0718-48082012000100006

Vázquez, N., Ortega, J., Samaniego, V.C. & Arberas, C.L. (2018). Conferencia Magistral "Enfermedades genéticas en población pediátrica argentina: una mirada desde la

Psicología de la Salud". X Congreso Internacional de Psicología: Promoviendo el Bienestar Psicológico en Niños y Adolescentes. Octubre 2018.

Vázquez, N., Ortega, J., Scavone, K., Samaniego, V. C., & Arberas, C. (2020). Escala de Adaptación Psicológica al Asesoramiento Genético (EAP-AG): validación de una versión en español para padres. Revista Evaluar, 20(2), 20-34. https://doi.org/10.35670/1667-4545.v20.n2.30106

Zhi, H., Ho, H. T., Liang, R., Chan, H. S. S., Ip, Y. T., Zayts, O. A., ... & Fung, L. F. (2018). The Impact Of Paediatric Neuromuscular Disorders On Parents' health-Related Quality Of Life And Family Functioning. In *2nd Joint Annual Research and Scientific Meeting 2018*.

Tables

Table 1

Distribution of sample according to sociodemographic variables

Sociodemographic variables	**Sample**	
Children	N	%
Child's gender		
Masculine	28	82.40%
Femenine	6	17.60%
Education		
Does not attend school	5	1470%
Pre-school	3	8.80%
Special needs elementary	1	2.90%
Elementary	18	52.90%
Special needs high school	2	5.90%
High school	5	14.70%
Diagnosis		
Duchenne Muscular Dystrophy	21	61.80%
Congenital Muscular Dystrophy	5	14.70%
Leukodystrophies	4	11.80%
Mitochondrial myopathy	2	5.90%
Primary L-Carnitine Deficiency	1	2.90%
Mild spastic paraparesis	1	2.90%
Parents		
Gender		
Masculine	5	14.70%
Femenine	19	85.30%
Place of origin		
Buenos Aires City	12	35.30%
Buenos Aires Province	11	32.40%
Other province	11	32.40%
Marital status		
Married-living together	24	70.60%
Separated-divorced	9	26.40%
Single mother	1	2.90%
Educational level: mother		
High school completed or less	9	26.50%
Technical degree complete-incomplete	10	29.40%
University complete-incomplete	14	41.10%
No answer	1	2.90%
Educational level: parent		
High school completed or less	19	55.90%

Technical degree complete-incomplete	6	17.60%
University complete-incomplete	8	23.50%
No answer	1	2.90%
Main family income		
Person with greater income		
Father	11	40.70%
Mother	11	40.70%
Other	5	18.50%
Currently working		
Yes	31	88.6%
No	2	2.9%
Medical coverage		
Yes	31	88.6%
No	1	2.9%
No answer	3	8.5%

Table 2

Description of Psychological Adaptation Dimensions (PAS)

Psychological Adaptation Dimensions	Possible range for the dimensions	Mean	SD
Self-esteem	1-5	3.90	1.11
Coping Efficacy	1-5	4.03	0.99
Social Integration	1-5	4.24	1.09
Spiritual Well-being	1-5	4.04	1.21

Table 3

Description of Adaptative Functioning Scales (ASR)

Adaptative Functioning Scales	Scale Range	NMD		Omnicultural		Effect size	
		Mean	SD	Mean	SD	Cohen's d	r
Friends	0-12	8.05	2.58	8.2	0.7	-0.08	-0.03
Spouse/Partner	-8-8	3.61	4.05	4.3	0.8	-0.24	-0.12
Family	-4-4	2.25	0.51	1.5	0.1	2.04	0.71
Job	-10-4	2.36	1.71	-*	-*	-	-
Education	-4-6	4	1.96	-*	-*	-	-
Personal Strenghts	0-22	16.08	2.84	15.5	2.1	0.23	0.12

Note: Onmicultural means reported by Rescorla et al. (2016).

*Omnicultural means for Job and Education scales were not reported in the original study.

Table 4

Comparison of broad scales with omnicultural means (ASR)

ASR broad scales	NMD		Omnicultural		Effect size	
	Mean	SD	Mean	SD	Cohen's d	r
Total problems	53.21	31.17	42.7	6.1	0.47	0.23
Internalizing	21.79	13.81	14.6	2.1	0.73	0.34
Externalizing	10.76	9.21	10.6	1.8	0.02	0.01

Note: Onmicultural means reported by Rescorla et al. (2016).

Table 5

Description of Mental Health Narrow Scales (ASR)

ASR Narrow Scales	Scale Range	NMD		Omnicultural		Effect size	
		Mean	SD	Mean	SD	Cohen's d	*r*
Anxious/Depressed	0-36	11.88	7.26	8	1.1	0.75	0.35
Withdrawn	0-18	4.32	4.04	3.3	0.8	0.35	0.17
Somatic Complaints	0-24	5.59	4.72	3.3	0.7	0.68	0.32
Thought Problems	0-20	1.76	2.13	-*	-*	-	
Attention Problems	0-30	7.85	5.81	6.6	0.7	0.3	0.15
Aggresive Behaivor	0-30	6.26	5.96	5.3	1	0.22	0.11
Rule Breaking	0-28	2.24	2.75	-*	-*	-	
Intrusive Behavior	0-12	2.26	2.03	2.4	0.6	-0.09	-0.05

Note: Onmicultural means reported by Rescorla et al. (2016).

*Omnicultural means for Thought Problems and Rule Breaking scales were not reported in the original study.

Table 6

Correlations between psychological adaptation and mental health problems

		Internalizing problems-ASR	Externalizing problems-ASR	Total problems - ASR	Anxious/Depressed	Withdrawn	Somatic Complaints	Thought Problems	Attention Problems	Aggresive Behaivor	Rule Breaking	Intrusive Behavior
Psychological adaptation -PAS	Spearman's Rho	-0.38*	-0.33	-0.42*	-0.37*	-0.43*	-0.23	-0.26	-0.20	-0.23	-0.38*	-0.24
	p	0.03	0.06	0.01	0.03	0.01	0.19	0.14	0.25	0.19	0.03	0.18
	N	34	34	34	34	34	34	34	34	34	34	34
Coping efficacy - PAS	Spearman's Rho	-0.16	-0.14	-0.22	-0.17	-0.23	-0.13	-0.13	-0.04	-0.03	-0.23	-0.07
	p	0.35	0.41	0.2	0.35	0.19	0.45	0.48	0.83	0.85	0.19	0.68
	N	34	34	34	34	34	34	34	34	34	34	34
Self-esteem - PAS	Spearman's Rho	-0.38*	-0.35*	-0.42*	-0.35*	-0.43*	-0.24	-0.26	-0.25	-0-25	-0.41*	-0.29
	p	0.02	0.04	0.01	0.04	0.01	0.17	0.13	0.16	0.16	0.02	0.10
	N	34	34	34	34	34	34	34	34	34	34	34
Social Integration -PAS	Spearman's Rho	-0.37*	-0.33	-0.39*	-0.34*	-0.49*	-0.28	-0.11	-0.27	-0.26	-0.36*	-0.19
	p	0.03	0.06	0.02	0.05	0.00	0.10	0.55	0.13	0.14	0.03	0.27
	N	34	34	34	34	34	34	34	34	34	34	34
Spiritual Well-being -PAS	Spearman's Rho	-0.27	-0.24	-0.33	-0.29	-0.27	-0.11	-0.18	-0.28	-0.19	-0.18	-0.24
	p	0.12	0.18	0.05	0.09	0.12	0.55	0.30	0.11	0.28	0.31	0.17
	N	34	34	34	34	34	34	34	34	34	34	34

Note: *significant correlations <0.05

Table 7

Correlations between adaptative functioning, psychological adaptation and mental health problems

		Total Problems-ASR	Internalizing Problems - ASR	Externalizing Problems -ASR	Psychological adaptation -PAS	Coping efficacy - PAS	Self-esteem - PAS	Social Integration - PAS	Spiritual Well-being - PAS
Friends - ASR	Spearman's Rho	-0.28	-0.39*	-0.19	0.28	0.19	0.23	0.22	0.26
	p	0.09	0.02	0.28	0.11	0.27	0.18	0.21	0.14
	N	34	34	34	34	34	34	34	34
Spouse - ASR	Spearman's Rho	-0.61*	-0.57*	-0.57*	0.30	0.14	0.31	0.19	0.18
	p	0.00	0.00	0.00	0.10	0.46	0.08	0.32	0.32
	N	31	31	31	31	31	31	31	31
Family - ASR	Spearman's Rho	-0.06	-0.01	-0.14	0.14	0.01	0.19	0.28	0.25
	p	0.72	0.95	0.42	0.42	0.95	0.26	0.11	0.15
	N	34	34	34	34	34	34	34	34
Personal Strengths - ASR	Spearman's Rho	-0.33	-0.41*	-0.26	0.15	-0.02	0.31	0.12	0.31
	p	0.06	0.02	0.13	0.38	0.87	0.08	0.51	0.07
	N	34	34	34	34	34	34	34	34

Note: *significant correlations <0.05