

MUCOPOLYSACCHARIDOSES: PSYCHOSOCIAL VULNERABILITY OF FAMILY CAREGIVER

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ABSTRACT

Background: The majority of caregivers are family members and within the symptoms that the caregiver may present, there are manifestations of stress, psychological state of overload, stress or discomfort caused by the continuous provision of care for the family member, feelings and negative perceptions regarding their caring function.

Objective: The purpose of this research is to study the psychosocial vulnerability of family caregivers of patients with Mucopolysaccharidoses (MPS).

Methods: This study is based on a mixed method with a cross sectional and a descriptive approach. The first stage was quantitative and the second stage was qualitative, both of them to explore the psychosocial vulnerability of a group of family caregivers of patients (n=111) with MPS belonging to the Colombian association of patients with lysosomal storage diseases ACOPEL by its acronym in Spanish. This study collects quantitative data using the patient admission request format from ACOPEL, employed to obtain relevant sociodemographic variables: Gender, Age, Marital status, Education, Occupation, living area, type of housing, live with, socioeconomic stratum, monthly income and time spent as a caregiver. And this study collects qualitative data using a structured interview held to psychologists and healthcare professionals belonging to ACOPEL.

Results: The results show that most of them are women, relatives in a relationship or married; most of them belonging to a low socioeconomic stratum and a low professional education. The categories of analysis revealed were the attitude is towards illness, taking account diagnosis and health staff, support networks and psychoeducation. Secondly, the impact of care work on the physical and psychological health of female caregivers. Third, coping resources and finally the double mother role-caregiver and her emotional bond with the patient.

Conclusion: In conclusion, the attitude and knowledge towards the disease depend on the diagnosis and the prognosis according to the type of MPS. It is important more research the psychosocial aspects and vulnerability of family caregivers in patients with MPS to develop evidence-based knowledge, psychoeducation, and, appropriate interventions.

Keywords: Psychosocial vulnerability, psychosocial characterization, psychosocial aspects, family caregiver, mucopolysaccharidoses.

Introduction

The purpose of this research was to study the psychosocial vulnerability of a group of family caregivers of patients with MPS belonging to the Colombian association of patients with lysosomal storage diseases (ACOPEL) by its acronym in Spanish, to identifying coping strategies for caregivers of patients with MPS and their families. This institution is responsible for providing all possible psychosocial support to both patients and their families through joint work with healthcare professionals. The contribution from this research to ACOPEL would be related to the generation of new strategies for identification, prevention, promotion and intervention of the psychosocial vulnerability indicators of family caregivers especially women.

In Colombia, the caregiver usually is a relative who takes care and support patients with MPS.¹ Caregivers are mostly family members and predominantly for some chronic diseases or debilitating women, added to this, the role of caregiver is more often assumed by the mother.² The socioeconomic status,

social support networks and the characteristics of disability care in the country also influence the quality of life (QoL) of the caregiver.

Few studies have accurately outlined the disease characteristics of patients with MPS³ associated disorders^{4,5} and the psychological profile of their caregivers, so it has been found in different patient associations and organizations that work for these diseases, caregivers burned, poor QoL, psychological distress and stress, and insomnia due to most of them have to be alert at night keep watching patients do not have breathing difficulties while sleeping or sleep apnea. The inheritance pattern of mitochondrial diseases and the lack of information about the diseases can affect the QoL of mothers and can increase the anxiety and burden of the caregiver⁶, mainly related to functionality, vitality, and mental health.

The most frequent Mucopolysaccharidoses (MPS) with the highest case reports in Colombia are types I, II, IV and VI, being Morquio A syndrome the most frequent with 0.68 cases per 100,000 live births, suggesting that Colombia is one of the countries with the highest prevalence of this syndrome

worldwide, as well as the least frequent is type III with 0.17 cases and type VII.^{7,8,9} MPS are a set of lysosomal storage disorders associated with the accumulation of glycosaminoglycans (GAG`s) in body tissues and organs, due to an enzymatic deficiency required for the degradation of cellular GAG`s. Clinical signs shared by MPS disorders include skeletal abnormalities such as kyphosis, scoliosis, pectus carinatum, valgus alteration of the knees, carpal tunnel syndrome, joint abnormalities, spinal cord compression, reduced growth, gross facial features, involvement in vision and hearing; and cardiorespiratory manifestations.¹⁰ In addition, some subtypes of MPS (I, II, III and VII) present intellectual and neurological deterioration due to the collection of GAG`s in the encephalon.

Studies about neurodevelopmental assessment¹¹ and QoL¹² mention physical health domain scores lower for the caregivers of patients with MPS III B or Sanfilippo III-B syndrome and highest scores^{13,14} in the domain of physical health for caregivers of patients with MPS IV-A. Despite the mobility problems these patients face as a result of their phenotype, it seems that their caregivers do not experience the need for greater physical support.

In addition, patients with MPS-IV-A do not have mental retardation, they can be more independent and autonomous for activities of daily living^{8,15} therefore, require less maternal support. However, in all three studies, aspects of psychosocial vulnerability or demographic characteristics that may affect the QoL of these women are not considered. In the evaluation of the QoL of caregivers of patients with MPS-VI, low quality of life scores was identified in all domains³ finding also related to the characteristic phenotype of the disease, which means that mothers should provide more attention and support for activities of daily living. However, the authors report that in this study it was not possible to confirm this hypothesis, due to the study sample included only 1 caregiver of a child with MPS-VI.

This impact on caregivers can be divided into two aspects: an objective overload, which is due to the effort involved in the performance of care tasks, with the economic demands and social restrictions derived from them; and a subjective one, which refers to the dimension of the feelings and emotions caused by the care work.¹⁶

Medical social workers have a long history of providing psychosocial support for physical and mental health problems to patients with chronic or debilitating diseases.^{17,18} They are capable of building relationships and providing in multidisciplinary teams. Nevertheless, studies on caregivers mostly incorporate community practice and knowledge to solve problems and provide emotional support, identify psychosocial problems that can be a barrier to achieving optimal health¹⁹ and help address the relationship between diagnosis and environment, but very few studies report what are those factors of vulnerability or psychosocial risk presented by caregivers.

Caregivers of people with disabilities or limitations are prone to reduced QoL due to their complete dedication to the patient, understood as the perception about the position of a person in life, in a given context, based on a system of values or beliefs in relation to their goals²⁰, standards or expectations of life itself³ which leads them to neglect their own lives.²¹

Patients can be born healthy but the clinical manifestations worsen during the course of the disease³ and due to the extreme difference in symptom presentation²² and unpredictability of MPS, it may contribute to increased stress levels in caregivers. The impact of raising a patient with MPS can be devastating, and the first challenges facing caregivers are emotional distress, financial strains and serious behavior difficulties. Similarly, a study by Schadeewald, Kimball, & Ou²³ found that families with infants with MPS moved away from social events and limited visitors to their homes, which reduced levels of social support they could access.

The emotional and social spheres of caregivers' lives turn out to be the most affected and lead to high levels of overload. They are called hidden or unknown patients and are characterized by not attending medical services, delaying their health care.^{17,16} The majority of caregivers are family members and within the symptoms that the caregiver may present, there are manifestations of stress, psychological state of overload, stress or discomfort caused by the continuous provision of care for the family member, feelings and negative perceptions regarding their caring function.

This investigation contributes in Colombia to develop more efforts between academic and health institutions in order to create a baseline for neuropsychological assessment, diagnosis and intervention processes with orphan and lysosomal storage diseases, that improve the QoL of caregivers and their families, thus reducing the different impact factors of illness and caregiver work.

Methods

This study is based on a mixed method with a cross sectional and a descriptive approach, to explore the psychosocial vulnerability of a group of family caregivers of patients (n=111) with MPS belonging to the Colombian association of patients with lysosomal storage diseases ACOPEL by its acronym in Spanish. The first stage was quantitative and the second stage it was qualitative. This study collects quantitative data using the patient admission request format from ACOPEL, employed to obtain relevant sociodemographic variables: Gender, Age, Marital status, Education, Occupation, living area, type of housing, live with, socioeconomic stratum, monthly income and time spent as a caregiver. The SPSS statistics for Windows. Version 21.0. [24] program was used to analyze the information.

Additionally, the study collected qualitative data using a group structured interview held to psychologists and healthcare professionals belonging to ACOPEL, those were recorded, transcribed and uploaded into Atlas.ti 7 Windows version²⁵ to analyze the discourse. The interview included three main questions: What facilitates and what hinders the care activity? What is the impact on the psychological and physical health of caregivers? What are the coping resources they have developed?

Using a convenience sampling, a local Colombian psychology student contacted a population of caregivers of MPS patients (n=111), MPS I (n=10), MPS III (n=8), MPS IV – A (n=75), MPS VI (n=18), belonging to ACOPEL, from 2017 to 2018, and four professionals with experience in caring for caregivers, two from the social field and two psychologists from the clinical field (systemic and cognitive-behavioral).

ACOPEL was contacted to use the information collected in the patient admission request format to identify the sociodemographic variables necessary for the study. The information was updated and corroborated by caregivers, patients and family members in some cases, as well as the signing of informed consent to participate in the study. The SPSS program was used to get and analyze descriptive statistics, frequency and means.

Secondly, interviews with key informants were carried out. All of the group structured interviews were audio-recorded and transcribed by the authors. The questions cover three main points: the psychosocial elements that facilitate and hinder the care activity; the impact on the psychological and physical health of caregivers; and the coping resources they have developed. Participants were de-identified on transcription and initially, a reading of the de-identified data was conducted to gain an overview of the whole of the data. Atlas.ti 7 Windows version was used to facilitate the data analysis and all interviews were transcribed by one research and the quality of the transcription was double-checked by another research. In addition, the Discourse Analysis (DA) [26] was a strategy to understand language as a communicative event within a social context.

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional research committee (Corporación Universitaria Minuto de Dios – UNIMINUTO - SP) with the 1964 Helsinki declaration and its later amendments or comparable ethical standards and informed consent was obtained from all individual participants included in the study.

Results

The results of the investigation are presented in the following order; quantitative data and qualitative data. Table 1 summarizes the demographic characteristics of caregivers, the median age was 42,64 years, the minimum age was 20 years and the maximum 85. Most caregivers were female (96.4%) with a median age of 42,64 years and Mucopolysaccharidosis IV-A or Morquio Syndrome Type A was the most common MPS type (n=75).

The Marital status shows caregivers in a relationship or married (65%), followed by a 25,2% single. 10,1% of them has no Education (n=11), 38,5% finished secondary and 15,6% are professional, but 81,8% represents no formal Occupation, being the mother who is most often engaged in this labor (82.9%), followed by grandmothers, sisters and aunts. There is only one case where the caregiver is not a relative, is a woman and a friend.

Caregivers belong mostly to the low socioeconomic stratum (79%), followed by the middle stratum (18%) and the high socioeconomic stratum (3.7%). Generally, 68,5% live in a rural area and 31,5% in urban living areas with type of housing (36,9%) rented. Women spend full time as a caregiver (79,1%) more than halftime (10,9%) and part time (10%). The majority of them take care of one patient, which represents 92,8% of cases.

The analyses of the responses from the semi-structured interviews shows one of the elements that becomes a facilitator or obstacle to care is the attitude towards the disease. Regarding the impact of care work on the physical and psychological health of caregivers, it was found: exhaustion,

lack of sleep, guilt, rejection and hopelessness; and finally, among the coping resources identified are: spirituality, social leadership, not abandoning the life project, self-care practices, and linking to support networks.

Table 1. Demographic characteristics of caregivers

Characteristics	n	%
Gender		
Female	107	96.4
Male	4	3.6
Age		
18–33	30	27.5
34–49	48	44.0
50–64	20	18.3
65+	11	10.1
Marital status		
Single	28	25.2
Relationship	37	33.3
Married	35	31.5
Divorced	7	6.3
Widowed	4	3.6
Education		
None	11	10.1
Primary	33	30.3
Secondary	48	44.0
Professional	17	15.6
Occupation		
No formal	90	81.8
Formal	20	18.2
Living area		
Rural	35	31.5
Urban	76	68.5
Type of housing		
Rented	41	36.9
Own	48	43.2
Familiar	19	17.2
Other	3	2.7
Live with		
2-3	30	28.0
4-6	60	56.1
7-11	17	15.9
Socioeconomic stratum		
High	4	3.7
Middle	19	17.6
Low	85	78.7
Monthly income		
Less than 1 SMMLV	74	69.2
Between 1 y 2 SMMLV	31	29.0
More than 3 SMMLV	2	1.9
Time spent as caregiver		
Part time	11	10.0
Halftime	12	10.9
Full time	87	79.1

Age=According to WHO (World Health Organization); Live with=number of people; Monthly income=Salario Mínimo Mensual Legal Vigente (SMMLV) by its acronym in Spanish.

In the attitude towards illness, the professionals observe that there is a relationship between the attitude towards the disease with the diagnosis and the health staff; with the link to support networks; and, with the level of psychoeducation acquired (Figure 1).

Diagnosis and health staff: Differences between female caregivers who receive a diagnosis in time, explained by a doctor who is an expert in the disease, and who receive a late diagnosis with inaccurate information and prognosis of death. Although, professionals report for the mother to receive the

diagnosis of a pathology always generates stress, the way in which the health staff informs about the diagnosis and possible prognosis determines the positive or negative attitude towards the disease.

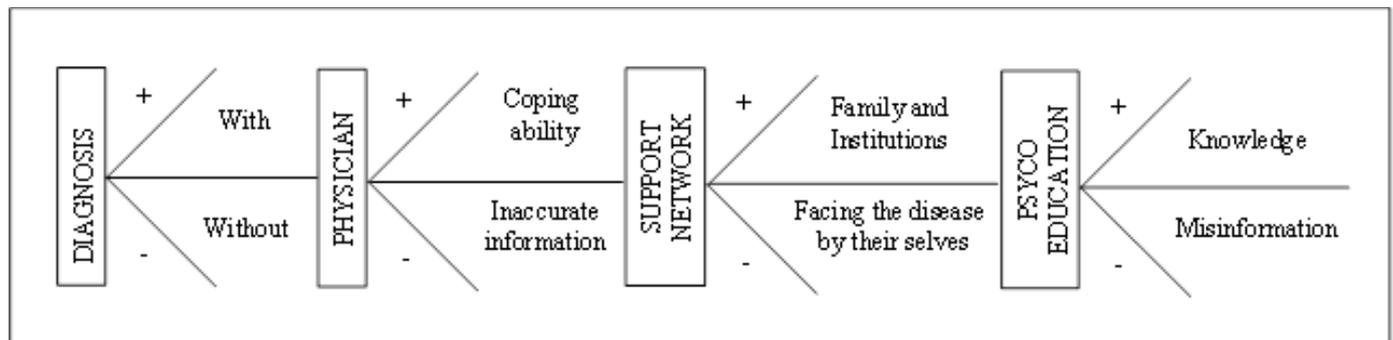


Figure 1. The attitude towards illness

Support networks: Differences between female caregivers who have a family and institutional support network, such as ACOPEL, compared to female caregivers who have long been facing the disease by their selves or with a weak family support network. These last women have a negative attitude that not only is directed towards the disease but towards life in general.

Psychoeducation: Differences between female caregivers who are acquiring knowledge about the disease, its effects and ways to get through, compared to female caregivers in whom there is a high lack of knowledge of the disease. In accompanied cases where female caregivers do not have scientific knowledge about the disease tend to establish links of overprotection with patients, there is a lot of fear, because the environment becomes threatening and they generalize that their relatives are not capable to be independent, preventing them from carrying out activities of their life cycle.

About the impact of care work on the physical and psychological health of female caregivers, in relation to physical health, greater physical symptoms emerge, especially in those women who have been facing the care of patients for themselves for a long time. Emotional support does not always include physical, instrumental or material support, there are patients with reduced mobility who must be hold, raised and carried. In these cases, physical fatigue of the female caregiver is evident, which is accompanied by an emotional component.

A recurring difficulty on a physical level is lack of sleep, the vast majority of patients have sleep apnea, and some must use a continuous positive airway pressure (CPAC) at night, which is why mothers-caregivers sleep with them, having their rest affected as well as your quality of life. Regarding the impact on an emotional level, female caregivers express constant feelings of guilt because of from their knowledge and the way they understand the disease; they believe they are to blame for the disease.

Another group of recurrent negative emotions in female caregivers is hopelessness accompanied by rejection of their sons and daughters, this set of emotions in some cases carries thoughts and desires of death themselves and their families.

The coping resources developed in female caregivers are characterized by a high sociocultural component, which has allowed them to properly manage the disease. First, give a spiritual sense to the disease; second, participate in women's empowerment processes as social leaders of patients' health rights, specifically on the issue of the requirement for compliance

with public policy that benefits the population in a condition of disability; Third, do not abandon the life project despite the diagnosis, both personal and that of their sons and daughters maintaining the normal activities of their daily lives.

Fourth, have self-care practices such as playing sports or reading. These women are characterized by having a high organizational capacity between patient care responsibilities and their personal space, specifically between transfers to the site where enzyme replacement therapy is carried out, and their self-care spaces.

The fifth and last coping resource is to link and participate in support networks. To know there are other people with the same pathology and similar life stories, makes them feel they are not alone and they are part of a group where they receive help and teach each other how to solve the problems presented in the daily care.

Regarding the double mother role - caregiver and her emotional bond with the patient, it is common for women to assume the care of the patient by the patriarchal culture of the country in which they are immersed, due to their biological condition it is assumed that the woman is the most indicated to carry out the care activities, situation that they reinforce by establishing that neither their sentimental partners nor another family member have the capacity to provide adequate care and it is reported that in the majority of cases accompanied, that the man's function is as a provider.

Fulfilling a double role as a mother and caregiver, women are disconnected from their educational and/or work activities because they must be accompanying all the moments in which the patient requires medical attention, which becomes a risk factor for their health. On the other hand, there is recognition of their work in the health field by medical staff recognizing advances in the patient due to care but their work in social and cultural terms is poorly valued.

The meaning of their life project tied to the work of care begins with the understanding of the diagnosis, and then with adherence to the treatment, accompanying the patient so that he does not abandon it. The progressive deterioration of the disease adding to the treatment causes loss of autonomy in mobility, feeding and hygiene in the patient. The caregiver becomes the physical and emotional support

of the patient, displacing their personal needs, losing their identity and subjectivity.

When the female caregiver and the patient are going through two different moments of the life cycle, adulthood and puberty-adolescence, the relationship begins to tense due to the transition to adolescence requires adult distancing as long as their sexual dimension begins as well as the question about their identity. The caregiver is not prepared for that separation and the patients are not open to express their desire for their own space because they feel responsible for making their caregiver feel bad. The stage of transition to puberty-adolescence is characterized by feelings of worry and emotional tension.

Discussion

The study primary aim was to explore the psychosocial vulnerability of a group of family caregivers of patients with MPS and to identify coping strategies for caregivers of patients with MPS and their families. As regards the most common type of MPS in Colombia, the results show Morquio Syndrome (n=75) according to the literature^{7,8,9, 3, 19} and the least common type Sanfilippo Syndrome (n=8).

The impact on caregivers divided into two aspects remains an objective overload, which is due to the effort involved in the performance of care tasks, with the economic demands and social restrictions derived from them; and the subjective one, which refers to the dimension of the feelings and emotions caused by the care work.^{6,16,21,17,3}

The woman stands out as a primary caregiver being the most prevalent female sex in the country (96.4%), which correlates with the literature² since women are the most dedicated to the work of caring for patients with lysosomal storage disorders such as MPS, rare, chronic or debilitating diseases. These female caregivers are characterized by belonging to a low socioeconomic stratum (79%), just 38,5% finished secondary and 15,6% being professionals located in rural areas (68.5%)¹⁹ with ages between 30 and 49 years. Moreover, most of them work full time as caregivers (79.1%) and in relation to economic solvency,^{16,27} they depend on their sentimental partner, mostly men, performing a double role, mothers and caregivers.

In terms of psychosocial vulnerability factors and impact on health, the attitude towards the disease depends not only on the diagnosis, it depends on the prognosis that varies according to the type of MPS, because the signs and symptoms are not the same in each of them. This means that the negative prognosis of the disease can influence more as a risk factor in the care work of the female caregiver than as a protective factor to cope with the disease. In addition, the most of them have no finished secondary education, increasing the risk of vulnerability to get a formal job representing no formal occupations and having no time to dedicate to understand medically and socially the disease.

The context, the patriarchy and the same socio-economic system of the country, do not allow women caregivers to carry out a stable life project in parallel with patient care. Socially, the role of care is assumed by women and in this population, particularly mothers. It involves changes in the physical and mental health on female caregivers with the appearance of symptoms such as stress, anxiety, lack of restful sleep.¹⁸ Also, symptoms like emotional lability.³ Caregivers are more receptive if they feel they have been listened to and their views and preferences can be considered.

Coping resources are protective factors for female caregivers, for instance, adequate adherence to treatment by patients and families, their beliefs such as religion and spirituality^{28,29} leadership understood as empowerment when you have the adequate knowledge and psychoeducation in the disease. In other words, these factors can allow women caregivers to lead processes of demand for health rights, be the voice of patients with different types of disabilities, help create laws that protect their care work and work hand in hand with associations of patients such as ACOPEL who seek not only physical well-being of the patient^{20,3} but psychological, neuropsychological and social, specifically in rare, orphan or low prevalence diseases such as MPS.

Conclusion

Psychoeducation is a vulnerability factor in this population of caregivers due to a lack of knowledge about the disease and its prognosis. If women caregivers can count on more learning and knowledge resources, they can strengthen their support networks and care strategies. Besides, the impact of their work as full-time caregivers can significantly influence their physical and mental health, as reported in the literature and health professionals thus presenting the syndrome of the burned caregiver.

Health prevention and promotion programs such as self-care, exercise, sports practices, meditation and life project aimed at mothers who are heads of households and at the same time caregivers, would be the main pillar to reduce the risk of the burned caregiver in relation to the diagnosis and prognosis of the disease in this type of population with MPS. Alliances with different organizations and associations, both national and international, could minimize the factors of vulnerability and psychosocial risk in relation to the Colombian context presented and the role of the female caregiver.

It is important from different medical and social organizations, such as patient associations, to provide information for the emotional support of patients in relation to changes in the life cycle. An emerging category relevant to the study of the psychosocial vulnerability of the female caregiver is the cultural sense of her work, and how this cultural sense to deconstruct and to turn into an emotional bond with the patient. Possibly, because the difference between the role of mother and the role of caregiver cognitive-behavioral.

Similarly, those kinds of studies allow strengthen the lines of research in the caregiver from interdisciplinary work, likewise to create baselines to subsequently build an instrument in the future that measures risk and protection factors taking into account specific psychosocial aspects in this population.

Limitations of the study especially are sample size and study design. The small sample size is a common critical issue within the field of rare disease including MPS. Evidently, the results could not be generalized. Furthermore, a longitudinal study design in this context would explore more the psychosocial vulnerability of that

group of female caregivers and to identify their coping strategies to looking for recommendations about psychological interventions, wellbeing, improvement of the QoL and decrease their feeling of burden and others psychosocial risk factors.

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