FACULTY OF NURSING

FINAL DEGREE PROJECT

INTERVENTIONS FOR FAMILIES OF CHILDREN WITH DRAVET SYNDROME TO IMPROVE THEIR QUALITY OF LIFE

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ABSTRACT

Title: Interventions for families of children with Dravet Syndrome to improve their quality of life.

Introduction: Dravet Syndrome is a developmental epileptic encephalopathy of genetic origin, in which 80% of affected patients have a mutation in the SCN1A gene. The literature indicates that families with DS have a poor quality of life due to the multiple manifestations of the disease.

Aim: To explore the interventions that can improve the quality of life of the families that have children with Dravet Syndrome.

Methodology: This narrative review used three different databases (Pubmed, CINAHL and PsycInfo). The researched articles were selected after having applied the limits and the criteria. A total of 10 articles were selected published between 2011 and 2022.

Results: Three main thematic areas were identified after reviewing the literature: early diagnosis, the new treatments for the management of DS throughout life and the person-centered health system.

Conclusion: This investigation has exposed the impact on the quality of life of the family with a child with Dravet Syndrome. In addition, derived from this, the importance of carrying out different interventions in order to improve the quality of their lives.

Key words: DS (Dravet Syndrome), quality of life, interventions, family.

RESUMEN

Título: Intervenciones para las familias de niños con Síndrome de Dravet para mejorar su calidad de vida.

Introducción: El síndrome de Dravet es una encefalopatía epiléptica del desarrollo de origen genético, en la que el 80% de los pacientes afectados presentan una mutación en el gen SCN1A. La literatura indica que las familias con SD tienen una mala calidad de vida debido a las múltiples manifestaciones de la enfermedad.

Objetivo: Presentar las intervenciones para mejorar la calidad de vida de las familias que tienen hijos con Síndrome de Dravet.

Metodología: Esta revisión utilizó tres bases de datos (Pubmed, CINAHL y PsycInfo) y tras haber aplicado los límites y los criterios, se seleccionaron un total de 10 artículos publicados entre 2011 y 2022.

Resultados: Se identificaron tres áreas temáticas tras el desarrollo de la investigación: el diagnóstico precoz, los nuevos tratamientos para el manejo del SD a lo largo de la vida y el sistema de salud centrado en la persona.

Conclusión: Analizando esta revisión, se ha expuesto el impacto sobre la calidad de vida que tiene la familia con un hijo con Síndrome de Dravet. Además, derivado de ello, la importancia de la realización de distintas intervenciones para poder mejorar la calidad de sus vidas.

Palabras clave: Síndrome de Dravet (DS), calidad de vida, intervenciones, familia.

1. Introduction

Dravet Syndrome, also known as Severe Myoclonic Epilepsy of Infancy (SMEI), was described in 1978 by the psychiatrist and epileptologist Charlotte Dravet. Dravet Syndrome is a developmental epileptic encephalopathy of genetic origin, with 80% of affected patients having a mutation in the SCN1A gene. Although the main manifestation is epilepsy, it is accompanied by various comorbidities, such as language and attention disorders, learning disabilities, behavioral disorders or gait ataxia (Aledo-Serrano and Mingorance, 2020).

The disease begins at 4 to 12 months of age and is characterized by generalized or unilateral clonic or tonic-clonic seizures of prolonged duration, both in a febrile context and other times not. Current remedy alternatives are limited, and the regular care required by a person with Dravet Syndrome considerably affects the quality of life of the affected person and family. For patients with Dravet Syndrome, the mortality rate due to SUDEP (sudden unexpected death in epilepsy) is approximately 15%. Therapeutic research provides patients and their families with hope for a better quality of life for their loved ones through a variety of programs and strategies (Dravet´s Foundation, 2021).

Due to its many symptoms, epilepsy has a significant impact on the quality of life of both DS affected people and their caregivers. Despite this, the literature shows that the DS family has a poor quality of life due to the multiple symptoms of the disease. Some of the families call it "living on alert so that their child does not die". This could be related to the fact that emotional, family and informational needs are not covered by such programs (EFE Salud, R., 2012).

2. Aim

To further explore this gap in the literature, a narrative review was conducted to examine the actual needs of families with DS in order to improve their quality of life through programs and strategies that address said needs. The main goal of this literature review is to generate interventions with the knowledge about the aspects and needs of families with DS to improve the quality of life that families yearn for.

3. Methodology

In order to conduct this narrative review, a research question was previously developed in order to outline the research terms. It was a PIS research question: Which interventions exist for families of children with Dravet Syndrome so as to improve their quality of life?

The scientific search was conducted in three different databases: Pubmed, CINAHL and PsycInfo. The searchers combined different terms, including “Dravet Syndrome”, “family” and “interventions” with Mesh Terms such as: “siblings”, “caregivers” and “quality of life” with their various synonyms. They were all combined with AND and OR as Boolean operators, as shown in Table 1. The limits applied in the three databases were articles from the last 10 years and articles in both English and Spanish. The inclusion and exclusion criteria are shown in Table 2.

The process of article selection is shown in Figure 1. Reviewing the literature considering the language boundaries, 46 articles were identified. After removing duplicates, and articles that did not meet the inclusion criteria, the number of articles was reduced to 5. A manual search of the Dravet Syndrome Foundation website reduced the final number of selected articles to 10.

4. Results of the Review

The research was finally done when the ten articles were selected. The countries in which the articles were written were: Spain, United Kingdom, France, Milan, USA, Canada, Belgium, Portugal and Paris. Of the twelve articles, there were two cohort studies, one transversal study, a formal study, a comprehensive research study, a report and and the last one was a large multinomial online survey.

Once the studies were reviewed, the data was collected and classified into three different thematic areas. The areas were divided according to the different possible interventions to improve the quality of life of families of children with Dravet Syndrome. The first one is early diagnosis, the second one is the new treatments for the management of DS throughout life and finally is the person-centered health system.

The different areas are described below with their different subcategories:

4.1. Early diagnosis

The time of diagnosis is defined as the number of months or years between the onset of the first symptoms and the diagnosis of DS. The number of months or years from the onset of the first symptoms to the diagnosis of DS was higher in older patients.

On average, 20% of physicians recognized DS at the first visit, and another disease was misdiagnosed in 80% of the cases. In 24% of cases it was the caregiver who suggested that it might be DS. This occurred more frequently in younger children. Diagnostic delays often lead to the use of medications that contradict DS in which 42% of patients with diagnostic delays greater than four years took three or more of these contraindicated drugs.

These data reflect the need for improvements in early diagnosis, coinciding with the opinion of DS specialists. The homogenization of the use of genetic testing between different regions and centers (public-private) facilitates this objective. Other barriers identified by the specialists include: lack of specific training for health professionals, lack of standardized clinical guidelines for DS, and lack of reference centers in the National Health Service (Aledo-Serrano and Mingorance, 2020).

Early detection allows families to organize their lives based on the knowledge that their children have ongoing problems (Camfield et al., 2016). Moreover, Sanchez Carpintero (2011) corroborates that information by saying that the consequence of an earlier diagnosis could improve the cognitive development of these children and opens up the prospects for improving prognosis.

4.2. The new treatments for the management of DS throughout life

There is a need for new treatments confirmed in Aledo-Serrano's study (2020) to improve the control of DS during the patient's lifetime. The research, the development and the approval of new drugs is essential as effective treatment would reduce the need for hospitalizations, which, as our results show, is associated with numerous problems that negatively impact families.

The need for effective treatment shown in the study, is one of the primary concerns of caregivers. Consequently, fenfluramine and cannabidiol have been developed and researched in order to ease this problem. For example, Cannabidiol has recently obtained a favorable opinion from the EMA Committee for Medicinal Products for Human Use and market approval by the European Commission, while fenfluramine is under evaluation by the EMA, with a decision expected in early 2020 (Aledo-Serrano and Mingorance, 2020).

4.3. Person-Centered Health System

In addition, it is important that the generation of health structures facilitate access to therapies that contribute to the comprehensive care of these patients, taking into account the different dimensions, aspects and comorbidities of the disease (Aledo-Serrano and Mingorance, 2020).

4.3.1. Disease conscious health care structures

It is suggested by a large body of evidence and experts that caregivers of patients with DS would benefit from medical attention when they are under stress (Campbell et al., 2018). Therefore, for providing a comprehensive patient-centered care is necessary physician-family cooperation. This will contribute to achieving the best possible quality of life and help the family "integrate the original identity and dreams with the adapted identity and dreams" (Granata, 2011).

Another intervention was to talk to the members of the clinical team about comorbidity and mortality and see the feasible and comprehensive information about the effects of DS (Gonçalves et al., 2021).

Furthermore, for effective support and follow-up, healthcare professionals should be knowledgeable about the impact of DS on caregivers. For example, psychological support or DS support teams should be offered, and providing them with helpline contacts as a result of the considerations of caregivers about their child's development and future with periodic screenings of these caregivers searching for signs of depression and anxiety (Gonçalves et al., 2021).

4.3.2. Different types of support

Caregivers felt that the indirect effects (i.e effects not directly attributable to the disease) were as important as the direct effects of seizures, and that Dravet Syndrome affected multiple areas of their lives. These findings underscore that a biopsychosocial approach to the treatment of DS should be developed, in which combined pharmacological and psychosocial interventions are implemented and evaluated (Nabbout et al., 2020).

On the one hand, the regular screening advocated by Gonçalves et al. (2021), would assess anxiety and depression so that caregivers would have the opportunity to attend psychological support groups to express their concerns about their child's possibilities and future. In addition, they suggest that larger studies and approaches will be undertaken on the development of depression, anxiety and associated stress and poor quality of life in DS caregivers (Gonçalves et al., 2021).

Moreover, support services are appropriate, including a community education program to address caregiver' information needs and concerns about DS and a trained respite service to represent caregivers when needed to care for their children with DS (Gonçalves et al., 2021).

On the other hand, another intervention would be to introduce the family to one of many online Dravet support groups. These groups provide parents up-to-date information and help families cope with a difficult reality (Camfield et al., 2016). In addition, Ceulemans (2011) suggests contact days for children with DS to share their experiences and issues.

To support the family, respite care or babysitting is important so that the couple can have time for themselves (Camfield et al., 2016). Health policies that give these caregivers access to

appropriate support services, including trained respite care providers who can substitute them when needed to care for their children (Gonçalves et al., 2021).

Parents should be encouraged to take time for themselves, both individually and as a couple. This may require psychological support and counseling to cope with the challenges of daily life. Parents should be encouraged to share the care of their sick child, maintain work and activities outside the home, and be given personalized and coordinated assistance in finding remedies mechanisms (Granata, 2011).

In most cases, caregivers received child care support from social welfare services, their partners and other family members. The average annual cost of an out-of-pocket caregiver was $3371. Other caregivers received child care support from social services (Lagae et al., 2019).

According to techniques to help the siblings of DS children, there are specific interventions for them. For example, spending more time with their friends and siblings, special outings alone with their parents, fun games or apps to distract them when they feel stressed and having more information about their siblings.

They should also need to learn how to cope with stress, worry and sadness and get emotional and social support for the whole family including their siblings. For siblings of children with a variety of chronic illnesses, there are emotional and social services for sharing positive and negative experiences in a safe and compassionate environment to improve coping and functioning.

Another option would be to expand such resources in DEE (Developmental and Epileptic Encephalopathies) specific areas to provide additional opportunities for siblings of DEE children to express their concerns in a supportive environment. Sibling programs or psychotherapeutic interventions (e.g., cognitive behavioral therapy) have not yet been performed on DEE family units (Bailey et al., 2020).

Another ingenious solution is for one parent to “stand by” with the child with DS daily and go to the emergency room as needed or to stay home taking care of multiple seizures while the other parent continues carrying out the activities with the unaffected sibling(s) (Camfield et al., 2016).

Siblings usually have a warm and compassionate attitude, but they also often feel jealous and miss out on enjoyable experiences due to the illness. The siblings are the ones that request age-appropriate information. In addition, some families have reported that involving siblings in emergency response plans (e.g., assigning them a specific role) reduces their anxiety in an otherwise terrifying situation (Granata, 2011).

Finally, taking into account financial support, health policies need to ensure that these caregivers have access to appropriate support services, including trained caregivers who can substitute for them in caring for their children when needed, as mentioned above (Gonçalves et al., 2021).

Caregivers were asked if they received financial support for medical fees, antiepileptic medications, therapies for comorbidities, emergency visit costs and home adjustments related to child care with DS. The majority of caregivers reported that they were fully reimbursed (i.e, patient copayments were 0%) for epilepsy specialists (80%), epilepsy medications (59%) and therapies (therapies for motor impairments [68%], speech impairments [74%], learning disabilities (other special education) [52%], autism [57%], ADHD [74%], and behavioral disorders [48%]). However, co-payment requirements vary widely across countries and many patients require out-of-pocket costs.

While most caregivers reported that they received either full (i.e 0% copayment) or partial reimbursement for epilepsy specialists, epilepsy medications and therapies, support varied depending on the country and those who did not receive reimbursement were required to pay a range of costs on the patients' needs (Gonçalves et al., 2021).

Very few adult patients (36%) reported benefiting from interventions such as supported

employment (14%), continuing education and training (15%), supported housing (18%) or other services to support independent living (21%) (Lagae et al., 2019).

4.3.3. Emergency protocol

Appropriate emergency treatment is to limit parental anxiety, provide punctual and effective medical treatment, and reduce the need for hospitalization. To achieve this, it may be useful to juggle various protocols. For example, a well-known protocol could be figured so that each member is responsible for handling a particular emergency. If it is known that seizures do not respond to rectal antiepileptic drugs, it may be suitable to think about inserting a percutaneously accessible permanent intravenous catheter for immediate treatment by the emergency physician (Granata, 2011).

A personal emergency protocol should be established for each patient to enable for proper and prompt treatment, even in unknown or unfamiliar hospitals. This personal protocol should contain demographic data, referencing doctor/hospital contact information, a brief medical history, diagnosis, allergies, daily medications, and specific strategies that have proven to be effective in emergencies. Finally, it may be helpful to build a portable microenvironment with everything needed to treat seizures, including a diazepam rectal kit, portable oxygen, inhalation, and daily medications (Granata, 2011).

5. Discussion

Through this review, it has been possible to know the existence of different interventions for the improvement of the quality of life of families with children with Dravet Syndrome. On the one hand, there are findings about the improvement of quality of life with the early diagnosis of the disease. On the other hand, the Person-Centered Health System helps the family in its different aspects such as support for the family, techniques for siblings, financial support, emergency protocol… Based on the results analyzed, it is difficult to recommend which of them is the best for the improvement of quality of life. However, it should be noted that, taking into account the different interventions, the use of them as a whole would improve the situation of the family.

The majority of the studies said that diagnostic delays often lead to the use of medications that are contraindicated for DS in which 42% of patients with diagnostic delays greater than four years took three or more of these contraindicated drugs. Consequently, this data reflects the need for improvements in early diagnosis, coinciding with the opinion of DS specialists (Aledo-Serrano and Mingorance, 2020).

Other studies have confirmed that early diagnosis helps families organize their lives with the knowledge that their child will have an ongoing problem (Camfield et al., 2016). In addition, Sanchez Carpintero (2011) corroborates that information by saying that the consequence of an earlier diagnosis could improve the cognitive development of these children and opens up the prospects for improving prognosis. According to Lo Barco et al. (2021), because seizure control may be partially associated with improved cognitive performance and improved quality of life, early diagnosis to avoid deterioration of treatment and to establish an optimal treatment protocol is required.

In contrast, further studies show that there is no evidence to support the benefit of early diagnosis of Dravet Syndrome. Thus showing the need for further studies due to the lack of evidence in this study (Wirrell et al., 2017).

The results of this research also suggest that one parent need to "stand by" with the child with DS daily and go to the emergency room as needed or to stay home taking care of multiple seizures while the other parent continues carrying out the activities with the unaffected sibling(s) (Camfield et al., 2016).

Other research has shown similar results, with some families implementing plans for one to take responsibility for one child at any given time. When their son had a prolonged seizure, the parent on duty took him to the hospital. The other parent is with their child and continues his/her normal activities (Nolan et al., 2008). Moreover, other studies confirmed that almost half (46%) siblings had missed recreational opportunities in the last four weeks to allow caregivers to take care of their siblings with DS (46%). Attendance at school had less impact

and have had few absenteeism in the last four weeks (Lagae et al., 2019).

The main limitations of this study are related to the article search strategy. One limitation to take into account is that only articles published in English and Spanish were included and that the literature published in the three databases mentioned above was taken into account. An additional limitation were articles whose publication date exceeded 2011 and 2021 have been excluded, as well as those that could not be accessed. Finally, it should be noted that although the results of all the selected articles were analyzed in depth, their grouping into the present categories depends to a large extent on the criteria established by the authors of this work and their interpretation of the results.

It is evident that the results of the present investigation have implications for clinical practice, teaching and research. Professionals will have a broader vision of the importance of the quality of life of the family in diseases such as Dravet Syndrome, so that the promotion of the family unit in nursing care and health care will be put into practice. Taking into consideration the limited existing bibliography, new research would be necessary to know how the individual and internal spheres of these families are currently being addressed and to promote new strategies for the management of this disease, as well as a more holistic attention to all those families who suffer from it.

6. Conclusions

In conclusion, the knowledge of interventions to improve the quality of life of families who have children with Dravet Syndrome can be useful when establishing strategies to address the social and health care needs of these caregivers, thus contributing to improving the quality of life of the caregivers and, consequently, of the patients.

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8. Annex

Table 1. Table with search keywords, synonyms and boolean connectors

INTERVENTION	A	*POPULATION*	A	*SITUATION*
Program	N	Family	N	**Quality of life [MESH]**
OR	D	**OR**	D	
Intervention		Parents		
OR		**OR**		
Strategy		**Siblings [MESH]**		
OR		**OR**		
Method		**Caregivers [MESH]**		
OR				
Plan		"Dravet Syndrome"		
OR				
Nursing intervention				

Table 2. Table with the inclusion and exclusion criteria

INCLUSION CRITERIA	EXCLUSION CRITERIA
→ Families with children with Dravet Syndrome	→ Articles that only talk about the disease
→ Children from 1 to 18 years of age of both sexes	→ Articles that do not include the family
→ Articles that include the improvement of quality of life	→ Articles that do not talk about interventions to improve the quality of life

Figure 1. Flowchart for the selection of studies