Rev Cient Soc Esp Enferm Neurol 60 (2024) 100144

Enfermería Neurológica

www.elsevier.es/rcsedene

CASE REPORT

''No SMA can hold'': Nursing care for children with spinal muscular atrophy. Descriptive analysis of two case studies

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Received 19 March 2022; accepted 25 November 2022

KEYWORDS
Chronic illness;
Decision making;
Spinal muscular;
Atrophy;
Self-care;
Children;
Co-production

Abstract
Background: Spinal muscular atrophy (SMA) is one of the most common genetic causes of death in children affecting about one in 10,000 live births, while its prevalence is about 1–2 per 100,000 (live births). Recently, the European Commission (EU) approved a novel gene therapy based on the onasemnogen abeparvovec (Zolgensma) for the treatment of patients with SMA. In addition to drug treatment, it is essential that children with SMA apply self-care methods to maintain their health, monitor their weight and food intake, and use appropriate remedies. Self-care and co-production of health care services are crucial in the modern ecosystem, as they can improve survival and prevent hospitalizations. The aim of this work is to support healthcare professionals who may have to deal with patients affected by this disease.
Methods: The article uses two case studies of children with spinal muscular atrophy through the creation of a multi-professional research group composed of health professionals who provide direct care to SMA children. The collection and analysis of the data were carried out by involving different figures who interact with SMA children. Specifically, physicians, nurses, parents, physical therapists, social workers, and teachers were individually interviewed.
Results: The study aims to provide suggestions on assessing child self-care, believing it to be a valuable method to gather information on how the child performs daily activities and how much the surrounding environment affects self-care. This paper highlights how self-management behaviors depend on four basic aspects: the person (individual, cognitive, and social perceptions), the patient's family (level of knowledge of pathology, involvement in the management and quality of relationship with the patient), the community (relationships with external social contexts, such as school and other organizations), and the healthcare system (availability of resources and the degree of evolution of healthcare).

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https://doi.org/10.1016/j.sedene.2022.11.002

Conclusions: The experience conducted may be helpful to other health institutions to make the approach to children with SMA as most effective as possible, creating internal workgroups and collaboration with external experts on the subject. Moreover, it provides valuable information on caring for families with children with SMA.

PALABRAS CLAVE
Enfermedad crónica;
Toma de decisiones;
Atrofia muscular espinal;
Autocuidado;
Niños;
Coproducción

«No hay AME que aguante»: cuidados de enfermería a niños con atrofia muscular espinal. Análisis descriptivo de dos casos prácticos

Resumen
Antecedentes: La atrofia muscular espinal (AME) es una de las causas genéticas más comunes de muerte en niños que afecta a uno de cada 10.000 nacidos vivos, mientras que su prevalencia es de aproximadamente 1-2 por cada 100.000 (nacidos vivos). Recientemente, la Comisión Europea (CE) ha aprobado una novedosa terapia génica basada en el onasemnógeno abeparvovec (Zolgensma) para el tratamiento de pacientes con AME. Además del tratamiento farmacológico, es esencial que los niños con AME apliquen métodos de autocuidado para mantener su salud, controlar su peso y la ingesta de alimentos y utilizar los remedios adecuados. El autocuidado y la coproducción de servicios sanitarios son cruciales en el ecosistema moderno, ya que pueden mejorar la supervivencia y evitar las hospitalizaciones. El objetivo de este trabajo es apoyar a los profesionales sanitarios que puedan tener que tratar con pacientes afectados por esta enfermedad.
Métodos: El artículo utiliza dos estudios de casos de niños con AME, a través de la creación de un grupo de investigación multiprofesional compuesto por profesionales sanitarios que prestan atención directa a los niños con AME. La recogida y el análisis de los datos se llevaron a cabo con la participación de diferentes figuras que interactúan con los niños con AME. En concreto, se entrevistó individualmente a médicos, enfermeras, padres, fisioterapeutas, trabajadores sociales y profesores.
Resultados: El estudio pretende aportar una sugerencia sobre la evaluación del autocuidado infantil, considerando que es un método útil para recabar información sobre cómo el niño realiza las actividades cotidianas y en qué medida el entorno que le rodea afecta al autocuidado. Este trabajo pone de manifiesto cómo las conductas de autogestión dependen de cuatro aspectos básicos: la persona (percepciones individuales, cognitivas y sociales), la familia del paciente (nivel de conocimiento de la patología, implicación en el manejo y calidad de la relación con el paciente), la comunidad (relaciones con contextos sociales externos, como la escuela y otras organizaciones) y el sistema sanitario (disponibilidad de recursos y grado de evolución de la atención sanitaria).
Conclusiones: La experiencia llevada a cabo puede ser de utilidad para otras instituciones sanitarias, para hacer lo más efectivo posible el abordaje de los niños con AME, creando grupos de trabajo internos y colaborando con expertos externos en el tema y aporta información útil sobre el cuidado de las familias con niños con AME.

Introduction

Spinal muscular atrophy (SMA) is a genetic neurological disease caused by degeneration of the alpha motor neurons of the anterior horn cells of the spinal cord.[1] It is characterized, predominantly, by progressive muscle weakness and, in the most severe types, paralysis. There are several subtypes of SMA, classified according to the age of the patient at onset and the motor function achieved: (i) type 1, also called Werdnig Hoffmann disease, with onset before 6 months; (ii) type 2, with onset between 6 and 18 months; (iii) type 3, with onset after 18 months. In a more detailed classification, type 0 and type 4 were observed for prenatal and adult forms, respectively. International statistics of SMA show that its incidence is about one in 10,000 live births, while its prevalence is about 1–2 per 100,000 (live births).[1] Children with SMA experience a complex and risky health state. During their daily management, they must address their dependence on a ventilator, cough machine, and enteral nutrition while

trying to reduce the likelihood of contracting preventable diseases, such as lung infections. Although ventilator support has been shown to improve survival in infants with SMA1, the difference in survival between invasive ventilation via tracheostomy and NIV (non-invasive ventilation) is unclear.

SMA is one of the most common genetic causes of death in children. Without respiratory support, SMA1 has a mortality incidence of 50% at 7 months of age and 90% at one year of age.[2] Recently, the European Commission (EU) approved a novel gene therapy based on the onasemnogen abeparvovec (Zolgensma) for the treatment of patients with SMA 5q with a bi-allelic mutation in the SMN1 gene and a clinical diagnosis of SMA type 1, or patients with SMA 5q with a bi-allelic mutation in the SMN1 gene and up to three copies of the SMN2 gene. The approval covers infants and children with SMA up to 21 kg in weight, according to the approved dosage. Zolgensma is a once-in-a-lifetime gene therapy designed to address the genetic cause of the disease by replacing missing or non-functioning SMN1 gene function.[3] Through a single intravenous (IV) infusion, a new functional copy of the SMN1 gene is inserted into the patient's cells, halting disease progression.

Patients with SMA are primarily treated at home. In this context, it is essential that children with SMA also apply self-care methods to maintain their health, monitor their weight and food intake, and use appropriate remedies (e.g., airway interruption, secretion removal, and prevention of immobilization syndrome) when there are problems to reduce the need for hospitalization.[3]

Worldwide, optimizing self-care is crucial in the health care system because it can improve survival and prevent hospitalizations.[4] Self-care turns out to be an aspect of healthcare co-production, in which patients contribute to creating, jointly with healthcare professionals and parents that are the primary caregivers at home, the service they need, thus going on to co-create clinical value.[5] Self-care methods in patients with chronic diseases are associated with reduced hospitalizations and improved quality of life and patient perceptions of health. Previous studies have shown that it is possible for children to engage in self-care[6]; therefore, understanding whether and how children with SMA engage in self-care behaviors may impact clinical practices and educational interventions. Previous studies have focused primarily on self-care behaviors in adults with motor neuron disease,[7] whereas self-care in children with a chronic illness is less frequently investigated.

Self-care in children with disabilities is an active, daily, and flexible process in which children and their parents share responsibility and decision-making to gain control of their health and well-being through a wide range of disease-related activities. For a child with a disability, self-care is essential to living optimistically and overcoming the challenges that the disease presents every day. Ultimately, the goal is to introduce individualized, early intervention and to control the progression of the disease.[2] Added that quality of life is compared to the quality of experience, but children with low self-esteem who have had a negative social impact also have a low quality of life and low levels of self-esteem and self-care. As youth transition from childhood to adolescence, they increase their awareness of people with disabilities and their autonomy.[8]

Increased self-awareness and disease awareness are linked to higher levels of self-care. This occurs because patients make informed decisions.[9,10] Travors et al.[4] stated that the transition from youth to adulthood affects development because perceived healthcare engagement is given tools (such as knowledge and responsibility) to be used to maintain their finely balanced health. Nested in adequate social, emotional and physical support, patients take responsibility for creatively integrating health behaviors they feel suit their condition by credible knowledge gained primarily through personal experience.

A recent review of the literature pointed to six prevalent themes that can be defined as following a young person's ability to self-care (problem-solving, decision making, optimizing living with an illness, child-focused homecare, role of healthcare professional, and monitoring behaviors).[11]

In light of the above premise, the purpose of this study is to examine the clinical, neuropathological, and genetic aspects of spinal muscular atrophy through the analysis and description of two cases of girls with spinal muscular atrophy in order to systematically explore the characteristics of self-care in children with spinal muscular atrophy and the relationship with their parents and the collaboration of health care providers. This paper aims to provide and share useful information to health professionals who should take care of such patients, stimulating better care and the logic of health co-production.

Materials and methods

Taking into account the specific context and the objective of the work, we chose to develop two cases of girls affected by SMA, both residents in the province of Barletta Andria Trani (Apulia, Italy), taken care of by health professionals of the ''Mons. R Dimiccoli'' hospital, Asl Bat Health Agency. The operating units that deal with SMA pathology with a multiprofessional approach are the Intensive Care Units that, to date, have demonstrated specific approaches and knowledge, achieving a higher success rate in terms of survival and quality of care for their patients than non-specialized centers.[11]

The collection and analysis of the data were carried out by involving different figures who have interactions with SMA children. Specifically, physicians, nurses, parents, physical therapists, social workers, and teachers were individually interviewed[12] with the aim of reconstructing the clinical history of diagnosis and treatment of the two involved children (A.D. and M.ML).

To safeguard construct validity and data triangulation,[13] multiple reference sources were used for data collection. The collected material was validated by the scientific manager of the project. All results were regularly discussed within the research team. To study the data, all material was manually coded by one of the authors and then discussed within the working group. For data analysis, we used the open coding method, grouping the raw data that emerged from the interviews within well-defined conceptual subcategories, referring to the topics under investigation. The presence of clearly defined conceptual subcategories meant that there were no errors of interpretation, thus bringing conceptually similar information

Table 1 Interview method.

Method	Object of analysis
Interview	13 Nurses 7 Physicians 2 Pairs of parents 3 Social workers 2 Teachers
Analysis	Approach to self-care Operational modes Disease management Therapeutic resources Care interventions

back into the relevant subcategory. In order to analyze the case studies, the research team first decided to map all of the caregivers, patients, and health care professionals involved in direct care. Following the analysis, the clinical cases were divided into three sections, namely: history of the diagnosis, nutritional and respiratory needs, and therapeutic resources (understood as all activities that can be done to improve the quality of life of SMA children).

The methods and objectives of the analysis are summarized in Table 1. The authors considered it useful to create a research group with all the professionals involved in the direct care of SMA children to collect and document experiences, analyzing how health professionals approached the disease and favoring the child's self-care process. In order to describe the case studies, the research group considered it useful to investigate retrospectively the work done with particular reference to the care interventions carried out, the approach to the disease and the use and efficiency of the available resources.

The written informed consent was obtained from parents or legal guardians, and based on institutional guidelines and the child's age, participants provided consent as appropriate.

Results

Case study 1 A.D., born in 2005 in Barletta from a eutocytic birth. At the time of the study, the female patient was 13 years old. At birth, the psychomotor development appears normal until the second month of life, when the motor skills are slowed down. At the completion of the third month, following a pediatric and a specialist neuropsychiatric examination, it emerged hyposthenia and hypotonia spread with greater expression in the lower limbs. The picture was so oriented toward suspicion of SMA type I. Only after genetic counseling exon 7 and 8 deletions were identified in a homozygous state. These two mutations represent about 98% of mutations identified in patients with SMA. The analysis conducted on DNA showed that AD had both deletions in the homozygous state, compatible with the state of the disease. In the same year of her birth, the first hospitalization at the O.U. of bronchial pneumology of the Bambin Gesù Hospital in Rome was scheduled. The initial objective examination showed marked hypotonia in the lower limbs and neck, fair control of the head with the presence of dorsal hypercosis and the presence of a hint of paradoxical breathing. During the period of hospitalization a personalized program of respiratory physiotherapy was applied, which maneuvers aimed at bronchial disobstruction. The procedure was explained to parents. A program of non-invasive ventilation (NIV) at night through the use of BI-PAP at low pressures was then started. After a period of hospitalization and once the general clinical conditions had been reestablished, AD was discharged. Parents were provided with various devices such as BI-PAP ventilator, secretion aspirator, cough machine, and AMBU balloon and were taught how to use them. During the years, in concomitance with the administration of the vaccine, AD experienced an increase in Calcitonin CT, which caused the first respiratory crisis related to ab-ingestis. Following this event, the child was rushed to the hospital and, after a period of hospitalization, she was discharged with a critical clinical picture with difficulty in swallowing and sucking. On 31/12/2005 AD was admitted to the Bambin Gesù hospital as an emergency case following major febrile episodes associated with respiratory crises and cyanosis. During her hospital stay with her parents, she underwent treatment with the ''COUGH MACHINE and BI-PAP'' but, despite these interventions, AD continued to show progressive respiratory decompensation associated with frequent desaturations caused by the complete atelectasis of the left lung. Given the lack of improvement in clinical conditions, the doctors deemed it necessary to proceed with tracheal intubation with mechanical ventilation and admission to the intensive care unit of the same hospital. The following year, a PEG (Percutaneous Endoscopic Gastrostomy) was implanted to allow adequate nutrition and exactly one month later, a tracheostomy was performed. The diagnosis formulated at the discharge was that of a patient suffering from SMA I with chronic respiratory failure and subjected to continuous mechanical ventilation through tracheostomy cannula with home ventilation and feeding through PEG. Today, AD has stable general clinical conditions; she is alert and well-oriented to her surroundings. Currently, due to nutritional needs, AD does not have the ability to chew and swallow food and water safety and even presents difficulties in swallowing saliva. In fact, she is at high risk of aspiration of food and water (suffocation) and incurring respiratory problems if the leftover food gets into her lungs. AD has developed significant gastroesophageal reflux (a condition in which food returns from the stomach to the throat) over time. In fact, she uses a medication to treat the symptoms and nutrition is administered enterally via Peg. She also receives support from the dietitian/nutritionist and medical team to monitor her weight and nutritional status. For respiratory needs, the child uses invasive respiratory assistance through mechanical ventilation, which is provided through the tracheostomy. She also uses a cough machine (coughtAssist) to promote alveolar recruitment and lung expansion.

Therapeutic resources AD is included in various therapeutic care pathways provided by the Health Agency Asl Bat and the municipalities of Barletta, like physiotherapy, occupational therapy, and speech, often through interventions or programs run by experienced staff. For physiotherapy, which is completely passive, since the child does not move any muscle of the body, the healthcare professional uses balloons and feathers as toys to facilitate good stimulation and give her a sense of independence and achievement. AD

undergoes hydrotherapy weekly, which is very helpful as the hydrostatic buoyancy of the water allows her to move her arms and legs that she would otherwise never move.

Case study 2 M.ML. was born in Canosa di Puglia in 2007 by eutoconic delivery. At the time of the study, the female patient was 15 years old. Throughout her first year of life, she has been a little girl with regular muscular movements and reactivity and general psychic conditions in normal. At the completion of the sixteenth month of life, in M.ML., abnormalities in the development of muscle tone were observed, which appears to decrease. With the continuation of the days, the situation regressed, with the child unable to maintain an upright position, accusing greater heaviness to the head than the rest of the body, and being the victim of continuous and sudden falls. However, the family, worried about the decline of M.ML's general conditions, decided to request an orthopedic consultation at the Policlinic of Bari and subsequent neuro-psychiatric consultation with attached genetic tests at the ''Casa Sollievo della Sofferenza'' located in San Giovanni Rotondo. M. ML's situation is not understood from the beginning both by the family pediatrician, who showed indifference to the pathological picture and by the specialists to whom the family relied on. At this point, M.ML's family decided to go to the Bambin Gesù Hospital in Rome. The medical doctors involved, considering the situation, decided to carry on further genetic diagnostic investigations, which led to the identification of a deletion in homozygosity in exon 7 of SMN1 gene, confirming the diagnosis of SMA type 2. The reason for the delay in obtaining a correct diagnosis should be taken into account because the suspicion of the disease was hidden by the presence, even if minimal, of reactivity. The first complications arrived at only 4 years of age following a respiratory crisis due to pulmonary infection. For this reason, M. ML. was hospitalized in the U.O. of bronchial pneumology of Foggia, which, not having a pediatric department, would face the first case of SMA until that moment. The girl was intubated for a period of 10 days, during which the worsening became more and more acute because of the refusal by the doctor to transfer the patient at a hospital suitable for the treatment of the disease. The hospital stay continued in Foggia where, after 40 days, the tracheotomy and PEG were placed. During the hospitalization, there were rare consultations by specialists and equally sporadic interventions by the resuscitator. Subsequently, M. ML. was transferred to the U.O. of Pediatrics at the Monsignor Dimiccoli Hospital in Barletta. After a period of hospitalization, she was discharged without a therapeutic plan; this would involve occasional interventions performed by the mother. Following the discharge, the parents decided to turn to the staff for the resuscitation of the same A.D., whose team decided to take charge of her. Difficulties, in this case, were present, given the difficulty in obtaining the life-saving devices for M. ML. A further complication occurred after a few months from discharge with leakage of the PEG as a result of poor positioning of the same. M.ML. was transferred to Foggia and then, at her parents' expense, to Rome for PEG repositioning. Today M.ML's conditions are stable; the motility of the lower limbs is absent, but her cognitive functions are not compromised. Currently, M.ML. benefits from therapeutic resources such as physiotherapy and occupational therapy as well as hydrotherapy, and in some cases hippotherapy (therapeutic riding). The physiotherapy is provided primarily by the home service of the Asl Bt Health Agency. Moreover, her parents and nurses make her perform stretching and joint flexibility exercises directly at home. In order to promote comfort and maximum mobility, the child uses appropriate seating systems and/or braces to assume the upright position. Standing is important for her physical development as it allows for better respiratory function, improved bowel function, and increased mobility. M.ML. uses a lightweight manual wheelchair. Despite the anxiety that the idea and use of a wheelchair generate in her parents, it actually provides her with mobility, independence, and a feeling of ''adventure'', encouraging her to use her residual upper body strength to explore the environment. In some cases, the muscles of her arms are weak and tired. The weakness is usually greater in the legs than in the arms, so she uses a motorized wheelchair operated directly by a joystick. Scoliosis represents a big problem for her because it limits her breathing and lung function. In fact, to reduce the advancement, she uses customized seating systems, seat supports, and corsets. The nutritional needs of a growing child are essential, and proper nutritional planning is fundamental in SMA2. For nutritional needs, M.ML. has swallowing problems, difficulty taking enough food by mouth to maintain her weight and regular growth, and therefore uses a PEG gastric tube for nutrition. She also receives support from the dietitian/nutritionist and medical team to monitor her weight and nutritional status. For respiratory needs, M.ML., for pulmonary ventilation, has been fitted with a tracheostomy, with continuous ventilation h24, and also uses a cough machine (coughtAssist) to promote alveolar recruitment and lung expansion. In conclusion, we can say that M.ML. conducts a normal life, attending school, friends, and the parish. Many initiatives have been carried out to stimulate cognitive, physical, and emotional health.

Suggestions for clinical nursing practice

In the above-described experiences, nurses put the patient through many stretching, gripping, and carrying exercises. These are small daily activities that should help them achieve independence. The patient clears her bowl from the table. It is heavy for her, but it stands as a part of therapy. Lifting, holding, and carrying; it is all about these little things to do. Nurses should use foam from a craft store to adjust the depth or height of the wheelchair seat. Nurses bought a slightly larger wheelchair to allow the patient to 'grow into it,' adding padding behind her back to position her a little more forward. Nurses should adapt and find a new normal each time. Nurses should not make her think she is different. She is one of us and does exactly everything we do. When nurses draw or color on the board, they use tape to hold the paper in place and make it easier for her to do the activity. Local public libraries that rent support equipment are hidden gems. The facilities are extremely low cost and often have much of the necessary equipment (wheelchairs, adaptive toilet seats, adaptive lift platforms, etc.). Crayons and pencils are okay, but with markers, the patient sometimes finds it difficult to remove the cap. In the above-described cases, the nurse in charge checked every

single brand of markers until he found the ones with caps that come off very easily. She has 30 different golf balls decorated with a hole drilled in them. The nurse used them as joystick knobs, changing them on special occasions or just for fun.

In order to develop independence, in the bathroom, the family keeps a tray with a cup, where she brushes her teeth. This way, she can brush her teeth sitting up, and the caregiver just drains the contents of the tray. It is good for the caregiver's back, because it saves them from holding her up to get her to the basin, and it is also safer for her. The parents bought an inexpensive writing tablet (to hold on the lap), so children can easily sit on the floor (without any fear of falling off a chair or couch) and have a hard surface to write or play on. Putting a knee immobilizer on them while they are sitting on the floor with their legs stretched out in front of them can also become a therapeutic activity. A cup in the water dispenser is always available on the outside of the refrigerator door. If she is thirsty, she just has to go to the kitchen and drink without worrying about reaching the dish cabinets.

Feeding tips, Homemade popsicles are delicious and can be made with healthy ingredients. Children can bite or suck on them as they wish. Parents fill store-bought molds with a mixture of strawberries, peel-free orange slices, ice, and water, as an easy and healthy snack. Smoothies with all kinds of nutrients to them, including supplements, are particularly welcome, as they are easy to drink and nutritious.

Repurposing the home environment, The parents changed the bathroom sinks to allow wheelchair access and added a faucet with a longer arm so that her hands could reach the water jet. In one of the cases, the living room was completely refurbished. The parents looked for adequate furniture but could not find any that would allow the children to remain active in their wheelchairs. It was enough to allocate one room for a different purpose, but it works because the children have all the space they want to move around. As much as possible, parents switched from carpets to a hard, smooth floor type throughout the house, not only to eliminate friction problems and make it easier for children to move around but also to clean it more easily from wheel marks. In some areas, parents set up some rubber mats and mats so children could play on the floor on soft surfaces without having to worry about them falling and hurting themselves by hitting their heads on the floor.

Discussion

This article contributes to the current literature by reviewing the concept of home-based self-care in collaboration with parents and health professionals for SMA children. Because of the uniqueness of spinal muscular atrophy, parents/caregivers of children with spinal muscular atrophy generally become experts in their child's care and play a crucial role in the multidisciplinary care approach and decision-making process.

They develop a high level of knowledge about the disease and their child's progress. They are extremely active, frequently using the Internet and social media to seek medical and social support. They perform most of the health care interventions their children need on a daily basis, including respiratory support, mechanical cough assistance, and nutrition/feeding. Parents often represent the key factor in the interventions their children will or will not receive

Analyzing the cases presented, it clearly emerged the presence of different stakeholders and, consequently, various tools for the management of the pathology, from diagnosis to care and treatment.

The experience conducted may be useful to other healthcare institutions to make the approach to children with SMA as effective as possible, creating internal working groups and collaboration with external experts on the subject. In addition, the article describes helpful information on taking care of families with children with SMA and the possibilities of therapy and treatment of the symptoms of a disease

The narration of the stories can be useful to inform all nursing professionals who will approach this disease about the importance of neonatal screening, which is essential to diagnose the disease immediately and start the needed therapies as soon as possible. Such treatments can offer children a future in terms of survival free from severe disability. A diagnosis that arrives a few months after birth, without any suspicion, can be even more disruptive. Until a few years ago, there was no cure: telling a parent who would give their lives to give his child hope that a cure does not exist may be devastating. Fortunately, things have changed a lot, but the general sense remains. Diagnostic communication does not end when the pediatric neuropsychiatrist provides the diagnosis. It is a process that begins when parents realize their child is not sitting up on his or her own when they realize something is wrong. The tests, investigations, and waiting may be devastating, and the diagnosis may be even worse. Nursing health care professionals must be ready to support and educate the family on all choices. Patient-centered communication means sharing information, improving relationships, managing uncertainty, and recognizing and responding to emotions to pave the way for self-care. Moreover, it is necessary to focus on the nature of these relationships and the factors that can support or impede effective communication. A systematic review of the literature identified how the healthcare structure helps or hinders the implementation of the decision-making process.[14]

The study aims to provide suggestions on assessing a child's self-care, believing it to be a valuable method to gather information on how the child performs his or her daily activities and how much the surrounding environment affects self-care. A child's routine performance is not always equal to his or her abilities. Performance assessment provides a way to indicate how the environment in which the measurement is taken affects the child's daily activities.

In fact, self-care or co-care in children with SMA can be assessed based on the severity of the disease, based on the child's level of autonomy, and how he or she copes with the disease on a daily basis (e.g., anxiety, stress, depression, acceptance, etc.).[5] The level of self-care depends on improving patients' ability to collaborate and communicate and to establish an empathic relationship with healthcare professionals. Several studies using multidisciplinary approaches have shown that an individual's ability to self-care must be supported by the presence of relatives, and particularly parents, and the expertise of health care professionals.[15,16] In order to increase their skills and deepen

their knowledge, health care professionals must participate in a systematic way to implement educational initiatives that provide useful tools to increase the self-care process in children. The International Standard of Care Committee for SMA was formed in 2005 with the goal of establishing guidelines for care.

In 2007, it published the Consensus Statement for Standards of Care in Spinal Muscular Atrophy, which addresses common medical issues, diagnostic strategies, and recommendations for assessment, monitoring, and therapeutic intervention in each area of care. An updated standard of care document reflecting advances in treating spinal muscular atrophy is currently under development. Current guidelines recommend that the family meet with a multidisciplinary team of experienced clinicians as soon as possible. A care plan should include referrals to experienced clinicians who will direct care for the child. The first referral will often be a child neuropsychiatrist. The level of care can vary from proactive to palliative.

Proactive care refers to procedures that address acute problems and daily management of a child's medical needs, including but not limited to airway cleansing, coughing, secretion management, respiratory support (invasive or noninvasive), nutrition, and hydration. Palliative care focuses on the prevention and relief of suffering to support the best possible quality of life. They may include: managing distress and preventing unnecessary interventions, potentially life-saving measures for the patient and/or family, and psychosocial and spiritual support for the patient and/or his or her family.

Moreover, socioeconomic and demographic factors, life context, life events, cultural and religious aspects, psychological aspects, and emotional issues related to illness have a significant impact on self-care.[17,18]

Self-care in children with SMA can be evaluated according to the gravity of the disease, the level of a child's autonomy, and how he/she faces the illness each day (e.g., anxiety, stress, depression, acceptance, etc.). The level of self-care depends on improvements in the patient's ability to collaborate and communicate and establish an empathic relationship with their healthcare professionals. The analysis of the literature that has investigated self-care in children affected by SMA underlines that problem solving is also an important aspect in children who have the ability to face and find solutions to maintain their state of health, thereby demonstrating a high rate of literacy and knowledge of the illness. It was demonstrated that children with SMA develop a certain self-care ability, as they succeed in carrying out self-care behaviors.

Studies using multidisciplinary approaches have shown that an individual's ability to self-care must be supported by the presence of relatives, and in particular parents, and the competence of healthcare professionals. In order to increase their competence and deepen their knowledge, healthcare professionals must participate systematically in implementing educational initiatives that provide valuable tools to improve the self-care process in children.

We identified the key nursing diagnoses in order to evaluate collaboration problems by NANDA taxonomy and plan and implement care: **Oxygenation and circulation** Bradycardia (10027274); Heart rate within normal limits (10029229); **Nutrition and hydration** Impaired self feeding (10000973); Low weight (10027316); Impaired swallowing (10001033); **Elimination and excretion** Constipation (10000567); Constipation absent; Improved constipation; Bowel continence (10027741); Urinary continence (10027836); **Activity and rest** Effective active range of motion (10052082); Impaired active range of motion (10052095); Impaired ability to transfer (10001005); Able to mobilize (10028461); Able to move in bed (10029240); Able to transfer (10028322); **Social interaction and loneliness** Effective social support (10045794); Lack of social support (10022753); Social isolation (10001647); Risk for social isolation (10047213). **Prevention of risks to life and wellbeing** Health care associated complication (10042451); No health care associated complication; Aggressive behavior (10002026); No aggressive behavior (10035645); Comfortable (10025574); **Health promotion and maintenance** Impaired ability of caregiver to perform caretaking (10035414); Impaired family ability to manage regime (10000902); Impaired ability to groom (10029632); Ability to perform impaired hygiene (10000987).

The use of Orem's theory of self-care as a basis for the categorization of nursing diagnosis statements favors the implementation of comprehensive and individualized care, encouraging people to take responsibility for their own health care, which converges with the main objectives of rehabilitation. This way, nurses can use different methods according to the requisites arising from self-care deficits, being able to act or do for the patient, advise, guide, supervise, offer physical and/or emotional support, provide and maintain a personal environment or teach them how to deal with their limitations. Most statements were grouped into universal self-care requisites, emphasizing the physiological repercussions and complications related to the organic functions of maintaining an adequate supply of air, water and food, vesico-intestinal elimination, mobility, safety, social interaction, and health promotion. Spinal cord injury results in important motor, sensory and autonomic dysfunctions, vasomotor, respiratory, intestinal and genitourinary impairment, in addition to potential complications such as neuropathic pain, spasticity, pressure injury, deep vein thrombosis, autonomic dysreflexia, and urinary tract infection. This draws attention to the need to implement preventive, recovery, rehabilitation, and control actions in order to avoid complications and reduce the level of dependence through stimulation and training for self-care and preparation for care in the home context.

In conclusion, this work highlights how self-care behaviors depend on four fundamental aspects: the person (individual, cognitive, and social perceptions), the patient's family (level of knowledge of the disease, involvement in management, and quality of the relationship with the patient), the community (relationships with external social contexts, such as school and other organizations), and the health care system (availability of resources and the degree to which health care is evolving.[19] Families need to understand their rights when it comes to making decisions about extending their children's lives and to understand that this is a very personal decision. In these cases, no choice is more ''right'' than another, only more appropriate for that specific family. Members of a hospital's pediatric palliative care team can help a family make the most appropriate decision for their situation. Once the family has made a choice about

what to do (i.e., whether to use invasive ventilation or to forgo it), it is crucial to make sure that all medical staff (and all other people with whom a family wants to share this decision) are fully aware of the choice they have made. It is also important to remember that families can change their minds during their SMA experience and should feel free to talk with their medical team at any time.

The limitation of the study is the numerosity of the sample size recruitment since SMA is a rare disease, so further qualitative and quantitative studies seem to be necessary to understand the experience of self-care in children with SMA, and in particular, to explore the determinants of self-care in this population.

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