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A Care Plan For the Patient with Spinal Muscular Atrophy (SMA) — Case Report

Plan opieki nad pacjentem z rdzeniowym zanikiem mięśni (SMA) — opis przypadku

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Abstract

Introduction. Spinal muscle atrophy (SMA) is one of the most common neurodegenerative diseases of genetic background the the child's age. Regardless of the form, it is essential to draw up a suitable nursing care plan that will help in accepting disabilities as well as in the preservation of self-care and self-nursing nursing by the child and family.
Case Report. The case report refers to a 17-year-old patient diagnosed with a diagnosis spinal muscle atrophy (SMA) five years before. Anorexia has resulted in patient's underweight. There is a functional deficit in the child. The family and the patient do not have sufficient knowledge about the disease.
Discussion. Taking care of a child with SMA during hospitalization period requires from all staff to have broad knowledge on mitigation of the symptoms emerged, delaying the consequences of weakness and muscle atrophy, and therefore prolonging functional efficiency.
Conclusions. The care issues of the patinet with SMA cover all areas of his functioning. The child and the family must accept the new situation they are in. Particular significance is attributed to systematic rehabilitation, which slows down the progress of the child's motor impairment which promotes effective self-care and self-nursing care, and thus the quality of life. **(JNNN 2017;6(3):114–119)**
Key Words: spinal muscle atrophy, quality of life, care

Streszczenie

Wstęp. Rdzeniowy zanik mięśni (SMA) to jedna z najczęstszych chorób neurodegeneracyjnych o podłożu genetycznym w dziecięcym wieku. Niezależnie od postaci niezbędne jest zbudowanie odpowiedniego planu opieki pielęgniarskiej, który pomoże w akceptacji niepełnosprawności, a także w zachowaniu samoopieki i samopielęgnacji przez dziecko i rodzinę.
Opis przypadku. Opis przypadku odnosi się do 17-letniego pacjenta z rozpoznanym pięć lat wcześniej rdzeniowym zanikiem mięśni (SMA). Zaburzenia łaknienia doprowadziły u pacjenta do niedowagi. U dziecka występuje deficyt sprawności funkcjonalnej. Rodzina oraz pacjent nie posiadają dostatecznej wiedzy na temat choroby.
Dyskusja. Opieka nad dzieckiem z SMA w trakcie hospitalizacji wymaga od całego personelu szerokiej wiedzy na temat łagodzenia zaistniałych objawów, opóźniania konsekwencji osłabienia i zaniku mięśni, a tym samym przedłużania funkcji sprawności funkcjonalnej.
Wnioski. Problemy pielęgnacyjne pacjenta z SMA obejmują wszystkie sfery jego funkcjonowania. Dziecko i jego rodzina musi zaakceptować nową sytuację, w jakiej się znajdują. Szczególne znaczenie przypisywane jest systematycznej rehabilitacji, która spowalnia postęp upośledzenia ruchowego dziecka, co sprzyja efektywnej samoopiece i samopielęgnacji, a tym samym jakości jego życia. **(PNN 2017;6(3):114–119)**
Słowa kluczowe: rdzeniowy zanik mięśni, jakość życia, opieka

Introduction

Spinal muscular atrophy (SMA) is an autosomal recessive inheritance (SMN1 gene mutation) of motor neurons of the frontal horn of the spinal cord [1]. It is one of the most common genetically conditioned diseases. It is the second cause (just after cystic fibrosis) of progressive disability in childhood, which leads to death of the patient. The incidence of SMA is estimated at 1 case per 10.000 births [2].

There are three forms of spinal muscle atrophy distinguished:

— Type I — early childhood Werdnig-Hoffman form, where clinical symptoms occur within first six months of life and the patient's survival does not exceed 2 years. In this form there is a characteristic image of a flabby, weakly screaming child who cannot lift the head by itself or even hold it upright. The child is not able to sit on its own. With the exception of the diaphragm, all the respiratory, bulgeous, more rarely facial muscles are occupied, the gallbladder muscle activity is preserved. Respiratory failure and pneumonia cause death in the early childhood form of SMA.

— Type II — intermediate form, Dubowitz disease — the first clinical symptoms usually appear in the third trimester of life, and before the age of 18. Symptoms are the same as in the first type of SMA, whereas the course of the disease is slower and milder. The child can sit on its own, but never starts walking. Gentle shaking of fingers can be observed. In this form, it is important to have early rehabilitation procedures applied in order to prevent developing contractures, deformities of the spine and feet. The age of 25 is achieved by 70% of the patients.

— Type III — juvenile form, Kugelberg-Welander disease — symptoms appear after 18 months of life, most often between 5 and 15 years of age. The first symptom being impairment of the gait, lumbar lordosis progresses. In the cases of an early start, patients stop walking before they reach maturity or move on their own until the age of 30–40. The lifespan in this form is similar to that of the general population.

— Type IV — adult form, symptoms appear in adulthood. In the clinical course, this form is similar to type III [1,2].

In the SMA clinical picture there occur the following symptoms:

— flabby baby,

— limb movements, mainly those of lower ones, are limited,

— the child can not raise the feet from the ground and the hands above the shoulders [3].

All reflexes usually disappear:

— muscular atrophy is often masked by adipose tissue,

— the position of the child resembles the "position of the frog", that is the limbs spread in the hip joints and bent in the knee joints,

— when lifting the baby under the arms, the legs hang loosely [3].

Observed:

— difficulties in sucking and swallowing,

— children's crying is quiet and weak,

— sitting and walking on their own is impossible,

— there occur more frequent and more severe infections of the lower respiratory tract, which cause respiratory failure, often requiring respiratory support [3].

Until now, no effective drug for spinal muscle atrophy treatment has been implemented. Attempts have been made to find ways to protect remaining motoneurons, or to use the effect of the presence of a second coding copy of the gene for this disease. Symptomatic treatment has been implemented: physical therapy, physical rehabilitation for the purpose of preventing contractures, breathing support, as well as diet [4].

The purpose of the study is to analyze the main aspects of nursing care for a child with SMA.

Case Report

The 17-year old patient admitted to the pediatric neurology ward in an emergency mode due to a breathing disorder. Five years earlier the patient was diagnosed with spinal muscular atrophy (SMA type III). Current symptomatic treatment includes pharmacotherapy, massage, rehabilitation, which is individually adjusted to the patient's condition.

The patient's breaths are irregular and their number is 14 breaths/min. There are murmurs in the hearing test. Blood pressure (RR): 120/70 mmHg, body temperature: 36.6°C, heart rate: 90 beats/min. On the second day of hospitalization due to respiratory disorders, the patient was given oxygen (1 l/h) at the doctor's request. Stitching and positioning in the semi-high position were applied. The patient reports lack of appetite, he is underweight. The patient weighs 51 kg at 165 cm, giving the BMI — 18.75 result. In physical examination, skin, urinary system, sexual system, endocrine system, sensory organs — with no pathological features.

The patient has reduced muscle strength. The posture is bent. Due to the limited range of motion, the patient moves using orthopedic crutches or with another person's

assistance. The patient scored 15 points on the Tinetti Scale — the risk of falling is high.

The patient has difficulties in self-care, therefore the child is always looked after by his parents. Having talked with the boy and his parents, one can say that the family does not have sufficient knowledge about the disease, self-care in the course of the disease, treatment or rehabilitation. The mood of the patient is reduced. The child is disturbed by the state of his health as well as by prolonged hospitalization.

SMA Patient Care Plan

Patient's Discomfort: Patient's Discomfort due to Disturbed Respiration and Gas Exchange

Negative diagnosis acc. to ICNP:
- — discomfort [10023066] + disturbed gas exchange [10001177] + disturbed respiration [10001316].

The purpose of nursing activities:
- — reduction of patient's discomfort,
- — better gas exchange,
- — improvement of breathing.

Nursing activities:
- — monitoring of vital parameters, including breathing,
- — teaching and doing breathing exercises with the patient, exudating secretions to improve gas exchange,
- — oxygen supply at the doctor's request: flow (1 l/h).

Nursing interventions acc. to ICNP:
- — monitoring of the breathing status (capacity) [10012196],
- — encouraging the use of breathing or curettage techniques [10006834] + chest physiotherapy [10044826],
- — oxygen therapy [10039369] + coperration with the doctor [10023565].

Evaluation of activities:
- — correct breath,
- — corect gas exchange,
- — discomfort reduced.

Positive/negative diagnosis acc. to ICNP:
- — effective gas exchange [10027993] + effective breathing [10041334]+ balance of mood [10035792].

Patient's Problem: Self-care Deficit due to the Musculoskeletal System Disorders

Negative diagnosis acc. to ICNP:
- — Self-care deficit [10023410] + impaired function of the musculoskeletal system [10022642].

The purpose of nursing activities:
- — reduction of the self-care and self-nursing deficit,
- — improvement of the musculoskeletal system efficiency.

Nursing activities:
- — evaluation of the degree of the patient's self-care,
- — assisting the patient in hygienic and nursing activities,
- — participation in patient's improvement; performing active and passive exercises with the patient,
- — teaching the patient to move safe.

Nursing interventions according to ICNP:
- — self-care evaluation [10021844],
- — assisting in toilet activities [10023531] + assisting with hygiene [10030821],
- — strengthening the technique of exercising muscles or joints [10036512],
- — teaching about mobility techniques [10041489].

Evaluation of activities:
- — the self-care deficyt not reduced,
- — musculoskeletal system disorders sustain,
- — adequate knowledge on patient's care and rehabilitation.

Positive/negative diagnosis acc. to ICNP:
- — impaired function of the musculoskeletal system [10022642] + self-care deficit [10023410] + adequate knowledge [10027112].

Patient's Problem: Body Weight Disorders Caused by Lack of Appetite

Negative diagnosis according to ICNP:
- — body weight disordered [10013016] + lack of appetite [10033399].

The purpose of nursing activities:
- — improvement of patient's appetite,
- — increase of body weight.

Nursing activities:
- — daily weight control and self-control diary,
- — educating the patient and family about proper nutrition: the type, frequency of meals, and the body's requirements for full-value meals,
- — familiarizing the patient and family with the diet prescribed,
- — encouraging the patient to consume full-value meals more often and in smaller quantities,
- — enabling the patient and his family to meet the dietician for the purpose of developing an individual diet.

Nursing interventions acc. to ICNP:
- — cooperation in the diet regime [10026190],
- — teaching about nutrition [10024618],
- — teaching about feeding technique [10045411],
- — teaching about dietary needs [10046533],

— teaching the family about the diet regime [10026525],
— strengthening compliance with prescriptions [10024562],
— cooperation with the dietician [10040435].

Evaluation of activities:
— body weight disturbed,
— appetite improved.

Positive/negative diagnosis acc. to ICNP:
— body weight disturbed [10013016] + positive appetite [10040333].

Patient's Problem: Difficulties in Coping with the Problem Faced by the Family and Child due to the Lack of Knowledge About the Disease

Negative diagnosis according to ICNP:
— lack of knowledge [10000837] + disordered coping with the problem by the family [10034789] + difficulties in managing the problem [10001120].

The purpose of the nursing activities:
— reduction of the knowledge deficit,
— preparing parents and the child for appropriate disease management.

Nursing activities:
— identifying parents' knowledge and the degree of coping with the problem,
— educational conversation with the family about disease and its management, providing educational materials: leaflets, books, films about the disease;
— educating the family about pharmacotherapy, diet and rehabilitation of a child with SMA;
— supporting the family and child;
— enabling the family to contact a psychologist;
— making parents aware of the possibility of using various forms of institutional and non-institutional support in child's care, enabling contact with parents of children regularly rehabilitated and suffering from SMA.

Nursing interventions acc. to ICNP:
— assessing the degree of the family coping with the problem [10026600],
— teaching about the disease [10024116],
— teaching about the medicine [1019470] + teaching about nutrition[10024618] + teaching about rehabilitation [1033017],
— promoting family support [10036078] + supporting the process of family coping with the problem [10032859],
— supporting the psychological status [10019161];
— facilitating the family's ability to participate in care planning [10035927].

Evaluation of activities:
— the family has adequate knowledge about the disease and how to manage with the child with SMA, so that they will effectively deal with the current situation.

Positive/negative diagnosis acc. to ICNP:
— family's effective coping with the problem [10034770] + adequate knowledge [10027112].

Patient's Problem: Lowered Mood due to Current Health Conditio

Negative diagnosis acc. to ICNP:
— anxiety [10000477] + impaired acceptance of health status [10029480].

The purpose of nursing activities:
— improvement of mood,
— acceptance of current health situation.

Nursing activities:
— assessment of anxiety,
— evaluation of health status acceptance,
— making contact with the patient and providing emotional support,
— strengthening self-esteem,
— strengthening the sense of hope,
— promoting the acceptance of health status,
— providing mental support.

Nursing interventions acc. to ICNP:
— assessment of anxiety [10041745],
— evaluation of health status acceptance [10026249],
— providing emotional support [10027051],
— promoting self-esteem [10024455],
— counseling on hope [10026212],
— promoting health status acceptance [10037783],
— mental status support [10019161],
— mood management [10036256].

Evaluation of activities:
— patient's mood has improved,
— the patient has accepted his health status.

Positive/negative diagnosis acc. to ICNP:
— balance of mood [10035792] + health status acceptance [10023499].

Discussion

Spinal muscle atrophy (SMA) is a heterogeneous group of neuromuscular diseases that results in the loss of motoneurons of the spinal cord. Loss of motoneurons leads in consequence to progressive paresis and muscle atrophy. According to sources, the incidence is 1:7000–1:10 000 live births, and the risk of SMA in the offspring of the ill is aproximately 1%. There are four forms of the disease distinguished: from SMA Ia to SMA IIIb,

where SMA Ia is the most severe whereas SMA IIIb is the mildest [5].

SMA is the second most common genetic disease leading to infants' deaths after cystic fibrosis. For several years, it has been in the phase of intensive research on molecular basis of disorders and of the search for effective therapy [6].

Molecular verification of SMA diagnosis is possible by identifying the SMN1 gene the mutations of which are responsible for the symptoms onset of the disease. Molecular research allows full genetic counseling, prenatal diagnosis and carrier testing. At present, innovative therapies plans are at the stage of clinical trials that are made possible thanks to better understanding of the pathogenesis of molecular disease [7].

When a patient is diagnosed with SMA, many questions regarding further care arise. The therapeutic team should as soon as possible implement the assumptions of interdisciplinary care for the patient. During hospitalization period, the members of the therapeutic team focus on evaluating disorders, particularly in the areas of motor and respiratory issues. It is necessary to implement systematic, planned improvement measures and to introduce to parents and children the importance of their regular use. Taking into account the complex problem of patient's disability, we should always consider its individual, social as well as interactive dimension [8].

The purpose of emotional support, also reinforced by contact with a psychologist, is to improve the child's mental condition and help with the acceptance of disability, by changing the worldview and adopting adequate attitudes and social roles. It is necessary to strengthen the sense of effectiveness and active attitudes [9].

The child described in this study requires help due to the lack of self-care and self-nursine care, family support, and a therapeutic team, as well as compensation for the lost and limited motor functions.

The task of the nurse is to prepare the child and the parents for self-control, self-care self-nursing care. However, in order to consider the patient and his family as partners for cooperation in the nursing process, the nurse bases on the understanding and getting acquained with the relationship between the ill child and his family [10].

In the professional approach, the most desirable relationship between the family and the ill, disabled child is the balance between the care and nursing needed, and the ultimate assistance obtained. This should be referred to the sphere of care and nursing care the child receives in the event of the disease as well as to the help in recovery. In the conditions of hospitalization, it is more advantageous to find some kind of deficit of assistance rather than its excess. This regards a minor deficiency of help, which can motivate the child when making an attempt to cope on his own in the situation emerged. Carrying out bedside rehabilitation and participating in the rehabilitation of the patient by a team of physiotherapists is another significant task for a nurse caring of a child with SMA [11].

The nurse has the task of providing counseling on the nutrition of healthy and sick children and should therefore pay particular attention to any deviation from the standard weight values of their bodies. Underweight includes children whose usual food intake is smaller than the required supply, for example due to lack of appetite or other reasons. Thanks to percentile norms, BMI, or weight-to-height ratio, we can determine whether the child has underweight problems. The situation of significant underweight is accompanied by a deficiency of nutrients, as well as by weakened immunity, more frequent infections, which can lead to worse outcomes of treatment. Depending on physical activity, gender, maturation stage, physiological energy demand of the child is changing. Therefore, food consumption should be adjusted individually, depending on the appetite and the growth rate of the small patient. For a few days during the hospitalization period it is important to record accurately what the child ate and drank, calculating at the same time the calorie intake of the meals and any possible weight fluctuations, and then add 500 calories to the estimated average caloric value [12,13]. The child should consume 5–6 regular meals a day. The interval between them should not be longer than 4 hours. Cereal products should be served at every meal during the day. It is necessary to use varied types of bread. Full-quality protein is provided to the child through meat, fish, milk and its products as well as eggs [14,15].

The results of the therapeutic actions during hospitalization and later period largely depend on the involvement of the parents. They require assistance and support in the care of the child. Very often they cannot satisfy basic needs of the child. They have no knowledge on the benefits of systematic activity and rehabilitation in the course of the disease. They also cannot obain information about SMA. This attitude of the parents does not create favourable conditions for the individual development of the child and it does not help in solving the current problems of the patient. The present case study of the situation of the child and his parents in the actions taken by the medical staff has included the belief of parents as to the correctness of regular child rehabilitation and updating of knowledge about SMA. The study also presents the benefits resulting from continuity of rehabilitation and the suggestion of regular contact with the parents of children subject to regular rehabilitation.

A child ward patient is a special nursing care receiver. Time spent in hospital is a kind of difficult experience

for the child. Hospitalization can be a reason for strong anxiety reactions and concern. Adaptation to a new environment is associated with a high physical and mental burden on the child. The person closest to the small patient is the nurse; it is her responsibility for the best possible adaptation of the small patient to hospital conditions. The actions that are carried out by the nurse towards child's acceptance of the current situation, depend on the child's age, as well as on the way of dealing with difficult situations. These aims are supposed to help minimize concern and anxiety and also establish and maintain contact with the child and the parents.

Children during hospital stay should be able to express their emotions and be informed by the nurse about their rights. An essential function of the therapeutic team for the adaptation of a small patient to hospitalization is the organization of leisure time and the presence of staff with the child in the absence of parents [9]. All these measures are meant to prevent the negative effects of child hospitalization and help in forming the skills to cope with life the situation has changed due to the disease.

Conclusions

1. Physiotherapy applied in SMA symptomatic treatment in a child slows down the progression of motor impairment, supports motor functions, and maintains maximum independence of the patient.
2. Early diagnosis of respiratory failure in patients with neuromuscular disease allows to implement non-invasive ventilation on time, which significantly increases the comfort of care and reduces the risk of complications related to tracheostomy and invasive ventilation.
3. The child and his or her family during care must confront the fact of an unfavourable diagnosis and make an attempt to accept the new situation.

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