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NURSING CARE AND EDUCATION OF THE FAMILY OF A CHILD WITH DUCHENNE MUSCULAR DYSTROPHY

OPIEKA PIELĘGNIARSKA I EDUKACJA RODZINY DZIECKA Z DYSTROFIĄ MIĘŚNIOWĄ DUCHENNE'A

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Summary

Some individuals suffer from a little-known, rare genetic disease called Duchenne muscular dystrophy. In Poland, approximately 40 boys are born with this condition every year. Therefore, paying attention to people with this rare disease was the motivation to undertake a qualitative study in order to identify the tasks of the nurse in terms of medical care and education of both the patient himself and his family, based on a case study. An interview technique, observation and analysis of medical records were used to develop the case study. As a result, the 2023 research material necessary for the case study was collected, which included: information on the patient's medical history, observation of the patient, the problems faced each day, the planned goals of assistance, the nursing interventions and their evaluation, the time of their implementation and their effects. Based on the study, it should be concluded that caring for a person with Duchenne muscular dystrophy presents a number of challenges. In such a case, a holistic approach to the patient is required that encompasses all areas of life and all aspects of the disease. The nurse not only deals with medical problems, but also provides support with mental, social and spiritual problems. The patient also needs care and support from the family, and education is helpful in preparing them to fulfil these tasks.

Keywords: Duchenne muscular dystrophy, nurses, family, patient, care

Streszczenie

Niektóre osoby chorują na mało znaną, rzadką chorobę genetyczną o nazwie dystrofia mięśniowa Duchenne'a. W Polsce co roku rodzi się około 40 chłopców z tą jednostką chorobową. W związku z powyższym zwrócenie uwagi na osoby z tą rzadką chorobą było motywacją do podjęcia badań o charakterze jakościowym w celu określenia zadań pielęgniarki/pielęgniarza w zakresie opieki medycznej i edukacji zarówno samego pacjenta, jak i jego rodziny, bazując na studium przypadku. Do opracowania studium przypadku wykorzystano technikę wywiadu, obserwacji i analizy dokumentacji medycznej. W efekcie zebrano materiał badawczy w 2023 roku niezbędny do opracowania studium przypadku, który obejmował: informacje dotyczące historii choroby pacjenta, obserwację pacjenta, problemy, z jakimi spotyka się na co dzień, planowane cele pomocy, interwencje pielęgniarskie i ich ocenę, czas ich realizacji i efekty. Na podstawie przeprowadzonych badań należy stwierdzić, że opieka nad osobą z dystrofią mięśniową Duchenne'a wiąże się z szeregiem wyzwań. W takim przypadku konieczne jest holistyczne podejście do chorego, które będzie obejmowało wszystkie obszary życia oraz wszelkie aspekty choroby. Pielęgniarka/pielęgniarz zajmuje się nie tyko problemami natury medycznej, jest także wsparciem przy problemach natury psychicznej, społecznej, duchowej. Choremu potrzebna jest również opieka i pomoc ze strony rodziny, a w przygotowaniu do wypełniania tych zadań pomocna jest edukacja.

Słowa kluczowe: dystrofia mięśniowa Duchenne'a, personel pielęgniarski, rodzina, pacjent, opieka

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Introduction

Some male children are affected by one of the rare little-known disease entities called Duchenne muscular dystrophy (DMD). In Poland, approximately 40 boys are born each year with this type of disease entity. The incidence of DMD is defined as 1 case per 3,500 boys born. However, there are cases of its occurrence in girls with Turner syndrome or in the case of X chromosome translocation [1,2]. The aforementioned disease has a genetic basis and is passed on from the carrier mother to her son. Boys are most often diagnosed between 2 and 4 years of age. The disease is confirmed through a genetic test, reimbursed in Poland by the National Health Fund.

DMD belongs to a group of disorders called "muscular dystrophy", characterized by a degenerative process of striated muscle fibers and connective tissue, which leads to atrophy of muscle mass and progressive weakness of skeletal muscles controlled by the patient. However, the pathological process does not result in sensory impairment. It was first described by the French neurologist Duchenne, and Mayron, in England in 1861. For several years, 7th September has been celebrated worldwide as Duchenne Dystrophy Awareness Day. Progressive DMD is caused by mutations in the DMD gene, located on the shorter arm of the X chromosome, and is the most common genetic dystrophinopathy, recessively inherited and sex-linked [3]. It is transmitted from carrier mothers (obligate carrier) or is the result of a *de novo* mutation. The symptoms of this disease are caused by abnormal expression of a protein called dystrophin, found in many tissues, including skeletal muscle and heart, which, as an essential component of the cell membrane, is responsible for the normal structure of muscle cells. It is essential during muscle contraction and diastole – it forms a bridge between intracellular proteins and the extracellular environment, and stabilizes muscle cell structure and function [3-5]. Lack of dystrophin results in abnormal muscle function and cell membrane damage, which gradually leads to muscle cell death. Recent studies indicate that dystrophin is also present in areas of the central nervous system, which may explain the cognitive impairment found in some individuals with muscular dystrophy [1,6].

Symptoms and diagnosis of DMD

The first visible symptom of DMD, present in the first years of life, is physical weakness. The progressive loss of muscle strength involves symmetrically the large muscles of the pelvic girdle and then the smaller muscles of the shoulder girdle. Clinical signs usually appear between the 2nd and 4th years of a child's life, sometimes slightly later, in which case the disease is expected to progress rapidly. The majority of newborns are asymptomatic, with a few cases of decreased muscle tone (hypotonia). Difficulties in getting up from the lying position occur when the baby starts to walk. When standing up from this position the child gradually supports himself against the ground and then straightens up, climbing over himself [7,8]. Common symptoms include tiptoeing, clumsiness when running and hopping, falling when walking on flat ground, followed by lumbar spine hyperlordosis and gait abnormalities. Typical symptoms also include difficulty walking up and down stairs and getting up from the floor, as well as pseudo hypertrophy of the calves 6. Between the ages of 10 and 14 many patients lose the ability to walk unaided. Curvature of the spine worsens (lordosis and scoliosis), the muscles of the upper limbs weaken, and pulmonary complications develop, which may in some cases require nocturnal non-invasive respiratory support. After the age of 15 years cardiovascular and pulmonological complications increase [9,10]. Limited initial physical activity, as well as low exercise tolerance and loss of independent walking ability make it difficult to detect early signs of heart failure. Typical symptoms of heart failure become apparent at a late stage of the disease. Patients with DMD are at higher risk of arrhythmia – either asymptomatic or causing palpitations, syncope, dizziness and, in rare cases, potentially leading to sudden cardiac death. In the early stages

of the disease, many patients present with sinus tachycardia, which increases in frequency with the progression of muscular dystrophy and deterioration of left ventricular function. However, it should not be considered an exponent of disease progression [11,12]. Among the most common endocrine disorders in patients with DMD is short stature. The rate of growth is slower than expected already in the first years of life and then runs along the lower centile values during childhood and adolescence. With the long-term steroid therapy used for patients with DMD, there are also risk factors for the development of osteoporosis [13].

Nursing care for children with DMD

As the disease progresses, the patient with DMD experiences problems that are the result of disorders of the circulatory, digestive as well as respiratory systems. As such, he or she requires specialized care, tailored to his or her individual needs, aimed at the early detection of symptoms as well as maintaining the patient in the best possible condition [14].

In the indicated care of the patient with DMD, many tasks are performed by the nurse and they cover many areas. It would seem that the first place would be taken by typically medical tasks. However, as research indicates, educating the family and the patient about the nature of the disease, its causes, treatment, prognosis, and emergency management comes first [15]. Educating the family is also about adapting the home to the patient's needs (non-slip carpets permanently installed, elimination of thresholds, grab bars in the bathroom, full shower or bath seat, specialized equipment and bed, ramp in front of the house – depending on the family's financial situation), and providing the patient with psychological support and motivation to continue exercising, learning, and maintaining relationships with friends and peers [16].

Therefore, it is important to inform the family about the functioning of support groups (Polish Neuromuscular Disease Society, Parent Project Muscular Dystrophy Foundation) and the possibility of financial support [16,17].

Secondly, tasks were identified to assist the patient with daily activities and, in the case of disease progression, also with basic activities, and to observe the patient for excessive muscle fatigue during exertion, as well as possible factors that exacerbate fatigue (infections, activity, stress). Further tasks that the nurse performs are to collaborate with the patient in performing improvement exercises – systemic, strengthening, isometric, active, passive, respiratory – according to the current stage and severity of the disease. If the patient requires feeding – preventing choking while eating, feeding the patient in a high or semi-high position and leaving the patient in this position for a minimum of 30 minutes after the meal, which is also accompanied by observation. Selecting foods in terms of consistency, as well as choosing an appropriate feeding method depending on the route of administration (oral feeding, gavage, gastrostomy, parenteral nutrition) – the patient should receive small portions of warm food at short intervals [18].

Paying attention to oral hygiene in the morning, evening and after each meal. Monitoring the patient for possible respiratory and circulatory complications – observing, checking pulse, temperature, blood pressure, saturation, and assessing pupils and consciousness. Chest percussion, cauterizing, performing breathing exercises, placing the patient in the drainage position, draining saliva and secretions from the bronchial tree to maintain airway patency. From the tasks listed, the multidimensional role of the nurse is evident, which includes medical care, education, support, observation and assessment of the patient's needs.

Family participation in caring for a sick child

A terminally ill child in a family implies a number of challenges not only typical of family dynamics, but primarily related to the disease. Such families face numerous stressors.

Financial stressor: the terminal disease of a child often means financial problems – it is often the case that one parent has to give up his or her job in order to be able to care for the child. The financial demands associated with

the disease include, among other things, special furnishings for the home, the purchase of medication, as well as equipment necessary for the patient's daily functioning and rehabilitation. Daily requirements: the challenges of daily life mainly concern the child's primary caregiver (usually the mother), who is at great risk of physical and mental overload and exhaustion due to the tasks placed on her.

Social demands: the social functioning of a family in which one member suffers from a chronic, terminal disease is necessarily disrupted – such a family faces restrictions on privacy, spontaneity of action, as well as on the time devoted to healthy family members [19-22].

The family's relationship with the external environment also changes. In this aspect, two attitudes can be distinguished – an isolationist one (the family closes itself in at home and avoids contact with the outside world) and an integrative one (striving to maintain existing relationships and bonds, and even expanding the network of contacts to include families also having chronically ill children). The ability to cope with a child's chronic disease depends on factors such as the type of disease and its severity, its visible features, as well as the prognosis and course of the disease (permanent, worsening, possible periods of remission), and the presence of pain [22-25]. An extremely important factor is the family's resources – both personal (socioeconomic status, ability to face difficult situations, self-efficacy, health status of individual family members) and social resources (which include closer and distant relatives, friends and neighbors), as well as environmental resources (relationships between the family and the interdisciplinary hospice team). Parents of a chronically ill child have a great need for information about the disease. Not being familiar with medical terminology, they try to assimilate as much knowledge as possible about the condition their child suffers from. During the course of the disease, both parents and other family members need to acquire the necessary skills to be able to provide proper care for the patient. The long-term education process already begins during the child's hospital stay and continues at home through collaboration with local primary care staff [26-28].

The most important tasks facing the family of a chronically ill child, including those suffering from DMD, involve: lowering the level of anxiety and sense of chaos, effective reorganization of home life, searching for new values and adjusting life plans to them, strengthening bonds between individual family members, counteracting the sense of loneliness, helplessness and powerlessness that accompanies the patient as well as his or her loved ones, combating the sense of hopelessness, searching for new areas of activity and action that give the patient satisfaction [29].

With the essence of the disease DMD and the need for care, necessarily outlined in brief, as well as our own professional experience, we were motivated to undertake a study to explore the nursing and educational care of a patient with the aforementioned disease and his/her family. The research topic is important because DMD is a so-called rare disease and, due to the way it is inherited, almost exclusively affects boys. The care and treatment of patients with DMD presents a number of challenges.

The main research problem was framed by the question: What is the nursing care and education of the family of the patient with DMD?

The main problem of the research was detailed in several questions:

1. What is the role of the nurse when caring for the patient with DMD?
2. What are the health care problems experienced by the person with DMD?
3. Is the family properly prepared to care for the patient with DMD?
4. What is the education of the family of the person with DMD?

The research used the case study method, which belongs to qualitative research because in the case of people with DMD, each person requires individual care, rehabilitation and therapy. The individual case study method involves analyzing the situation of the patient in question, and his or her social, motor or cultural situation, which may contribute to the improvement or deterioration of health, resulting in the process of care and treatment

[30]. An interview technique, observation and analysis of medical records were used to develop the case study. As a result, the 2023 research material necessary for the case study was collected, which included: information on the patient's medical history, observation of the patient, the problems faced each day, the planned goals of assistance, the nursing interventions and their evaluation, the time of their implementation and their effects.

Case description

A male patient aged 30 years with diagnosed muscular dystrophy. The patient lives with his parents and siblings in a single-family house in the countryside. The father, 51, with secondary education, works on his own farm. The mother, 50, with vocational education, takes care of the household, the care of her sick son, and the upbringing of his younger siblings. An atmosphere of friendship, calmness and mutual support is observed at home. No addictions, or smoking or alcohol abuse are observed among the household members. The patient is the child of the first pregnancy and first birth, born by natural causes at 39 weeks' gestation. He received 10 points on the Apgar scale; he was breastfed on demand until 6 months of age. He was not ill, developed normally, and underwent vaccinations according to the vaccination calendar.

During the next stages of the baby's development – the stage of standing, a strange pose of bending the body and climbing on the toes aroused the mother's concern. The child was lively and joyful. The son's rolling over when trying to walk was still not associated by the mother with the disease. At the age of 4, the boy was referred to a specialist clinic in Warsaw, Poland, where he was diagnosed with DMD. He was under the constant care of the neurological outpatient clinic for children in Biała Podlaska and the Neurology Clinic in Warsaw, Banacha Street, with recommendations for systematic rehabilitation and periodic check-ups at the muscle clinic.

His general condition continued to deteriorate despite his parents' efforts, exercises and rehabilitation in the pool. Once a year, the patient went on rehabilitation holidays from PFRON. At the age of 8, problems with mobility and initial contractures in the Achilles and knee tendons began. And already during this period, due to the boy's weak immunity and worsening disability, home education began. During the period of primary school education, when the patient was already in a wheelchair, he suffered two fractures of his lower limb. Although efforts were made to isolate the boy from clusters of people, the problem of respiratory failure became increasingly frequent. From 2010 onwards, as a result of a decline in the patient's immune system, there were inflammations of the respiratory tract, bronchitis and pneumonia, and influenza. There were major problems with lingering secretions. There was frequent choking. Choking of secretions was also occurring – there were increasing problems with effective expectoration.

In 2014, the patient was admitted to the intensive care unit of a nearby hospital with developing respiratory failure, swallowing disorders and airway obstruction. An emergency tracheostomy was performed and he was qualified for chronic home mechanical ventilation with a TRILOGY device with TV 0.3, PEEP 4, FiO2 0.21. In 2015 and 2018, the patient was again hospitalized due to an unspecified bowel obstruction.

He is currently a contact patient, in poor verbal contact (speaking in whispers), and still mechanically ventilated. He complains of recurrent abdominal pain. Despite exercise and massage, contractures are increasing. The patient is now an adult 30-year-old man who requires constant full-time nursing care, which his family is able to provide in collaboration with a long-term care nurse. His passion is computer games. He is currently developing his software and computer games, which he is thinking of releasing in the future. Despite his severe dysfunctions, this man has a lot of motivation to fight his disease. He is a well-groomed patient. No bedsores or sores are observed on the man's body. A slight redness on his right hip is caused by lying on his right side while operating a computer.

Case analysis

One of the main diagnoses is the difficulty in expectoration and the retention of large amounts of secretions in the airways, resulting from the dysfunction of the respiratory apparatus as a consequence of muscular dystrophy, which impairs their patency.

The aim of the care measures is to ensure better airway patency and better lung ventilation. In the implementation of measures to achieve this goal, it is necessary to assess the patency of the tracheotomy tube, perform bronchial tree toileting as required, carry out physiotherapeutic procedures, patting, suctioning of secretions from the tube, moistening the respiratory mixture, teaching effective coughing, checking vital signs and performing nebulization with 0.9% saline. After such measures, airway patency improved after about fifteen minutes.

Another nursing diagnosis made was impaired verbal communication due to the insertion of a tracheotomy tube and mechanical ventilation, often manifested by the patient becoming upset due to the caregiver's failure to understand his message. The aim of the activities in solving this problem was to establish non-verbal contact. The easiest way to solve this problem was for the mother to establish contact with her child by informing him in clear and understandable, short sentences. She explained to the patient that the cause of the discomfort was the inserted tracheotomy tube, encouraging him to gesture with his head. The mother tried to obtain information on how to communicate with her son, studied the literature on the subject, and participated in webinars on how patients with a tracheotomy tube communicate. After the mother's actions, the patient was more willing to communicate with the nurse and family, and expressed his needs non-verbally. The first results of the improvement in communication could be seen after just two weeks of intensive work between the mother and her son. The patient now communicates more readily with the nurse/caregiver. He reports his needs by gesturing.

A major problem in the patient was chronic constipation and flatulence related to long immobilization and slowed metabolism, manifested by abdominal pain and high nervousness. In order to solve this problem, efforts were made to establish the regulation of bowel movements by assessing the food served, including more liquids in the diet, a rich-belly diet, eliminating heavy and bloating foods, giving the patient anti-constipation medication, and giving the patient an abdominal massage with a warm hand for a few minutes to accelerate intestinal peristalsis. The aforementioned measures contributed to the patient having a partial bowel movement after 3 hours.

Another diagnosed problem was reluctance to take liquids and food resulting from recurrent abdominal pains manifested by aggression towards caregivers. The aim of the measures to address this problem was to get the patient to take food and liquids without aggressive behavior towards caregivers. In these measures, efforts were made to control the amount of liquids and food intake, meals were served according to the patient's taste preferences, ample meal time was provided so that he would eat in peace and without haste, and meals were prepared and served in smaller quantities but more frequently (5 times a day). After a week, it was observed that the patient ate the prepared meals willingly. The young man was increasingly observed to feel unwell and ashamed due to his inability to perform hygienic activities related to his mobility disability. The goal of the activities was to improve well-being and reduce embarrassment and restraint during hygiene activities. In an attempt to solve this problem help was given during hygiene activities to ensure intimacy by closing the bathroom door. During these activities, the patient was accompanied by one person – his father. After these types of measures were taken, it was observed after just one week that the young man willingly participated in daily full-body toileting, and even reported the need and desire to bathe. The patient also often developed general weakness, breathlessness and apathy, which contributed to his decreased appetite and lethargy. The

aim of the measures in addressing this problem was to improve the patient's wellbeing by maintaining the his nutritional and hydration status. These were activities carried out systematically. As part of these activities, a set of physical exercises was prepared, which the patient performed with his brother every day at a fixed time. Meals were prepared that the patient liked best. Every day an effort was made to take the patient out for at least a fifteen-minute walk. As a result of the measures taken, the young man's improved well-being was observed after just one week. He was more eager to eat his meals and did not fall asleep during the day. He reminded himself of the desire to go for a walk or do daily exercises with his brother. His state of well-being was improving significantly.

In the presented description of the diagnosed problems, the need for education and the role of the nurse in its substantive implementation can be seen. Education involves both the patient and his family, and mutual interaction.

Conclusions

The aim of the study undertaken was to learn about and present the nurse's actions in caring for the person-patient with DMD, and to highlight the importance of educating the family as well as the patient themselves. The care of the person with DMD and the process of treating it presents a number of challenges. In such a case, a holistic approach to the person with the condition is required, which will include all areas of life and all aspects of the disease. The nurse not only deals with medical problems, but also provides support with psychological, social and spiritual problems. The patient needs the support of the family, the nurse and specialists (dietician, psychologist). There is currently an increase in the number of patients mechanically ventilated at home. The home ventilation programme allows the patient to be at home among family and loved ones. Home treatment with the family reduces treatment costs and viral exposure (which was particularly important during the SARS-COV2 virus pandemic). It is also the nurse's task in caring for the patient with DMD to adequately support and prepare the family for care, which is ensured by appropriate education, especially providing all necessary information, answering questions and dispelling doubts, as well as providing possible guidance to enable family members to actively participate in activities to improve the patient's quality of life. So far, medical developments have not led to the development of an effective therapy for patients with this condition. Therefore, rehabilitation, as well as psychological and content-related support provided by the nurse play a huge role in the life of the patient with DMD and his or her family.

On the basis of the collected medical records, nursing observations, diagnosed problems of the person with muscular dystrophy and implemented activities, the following conclusions were made:

- The nurse caring for the person with muscular dystrophy should be prepared to overcome many barriers of a therapeutic, caring and psychological nature. In order for the patient to want to cooperate with him/her, he/she should be prepared in terms of content and should gain his trust, which will be ensured by such personal qualities as empathy, forbearance towards the patient's mood changes, kindness, and communicativeness.
- The nurse is a support for the patient and his family. He/she is often the first person to direct what action to take during the patient's hospitalization. In addition, he/she educates, observes, supports and provides assistance. All the actions he/she takes are aimed at the patient's welfare.
- The family is not always prepared to care for the patient with muscular dystrophy. Only after being educated by the nurse do they know that they can count on the support of an organization or a specialist (psychologist). They realize that it will be necessary to adapt the home to the needs of the disabled person. More often than not, one of the parents makes the decision to give up their job.

In conclusion, the correct cooperation of the nurse with the patient and his or her family, the diagnosis of problems, appropriately selected methods of care and education, and the assistance and support of the family allow the person with muscular dystrophy to live, learn and develop interests.

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